VIEWED AS A WHOLE:
SYNTHESES OF RESEARCH EVIDENCE AND TEACHING AND LEARNING SUPPORT RESOURCES RELATED TO GENOMICS EDUCATION IN SCHOOLS
Viewed as a whole: Syntheses of research evidence and teaching and learning support resources related to genomics education in schools

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Citation
This report should be cited as:

Every effort has been made to ensure that the contents of this report are accurate. If you have any comments on matters of factual accuracy, we would be happy to discuss these with you. Please contact the report team leader, Jeremy Airey, at the address below or at jeremy airey@york.ac.uk.

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The science and technology of genomics is developing at pace, with a myriad of new applications emerging all the time. But what do school students know about genomics and what should they be aware of? Two decades after the Human Genome Project published its first draft of the complete DNA code for human life, there remains a lot of questions to explore in genomics education.

How do students distinguish fundamental concepts, such as DNA, genes and genomes? Is genomics taught in a relevant way, keeping abreast of latest developments? Is there space to discuss societal and ethical aspects of genomics? In an already tightly packed curriculum, how much time should be given to this pervasive and emerging area of science?

We can’t (and don’t) expect all schools students to become genomics scientists. However, might we expect school leavers to be genomically-literate citizens? Working with the University of York Science Education Group, we have explored the landscape of genomics education in schools and the range of resources available to teachers. This report provides a lens through which we can take a critical look at the onward development of learning resources and wider education programmes, to help adapt and improve genomics learning journeys. Genomics will continue to weave itself into our health and wellbeing, playing a role in vaccine development, virus tracking, personalised cancer treatments, unveiling family histories, solving crime and tackling the planet’s fragile ecosystems. It’s never been more important to equip and prepare students for the opportunities and societal debates that lie ahead in this stimulating, and ever changing, area of biology.

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This report is the outcome of a landscape review of research and resource provision in the field of genomics education in schools. Genomics education is a developing field, catching up with the rapid scientific and technological developments that have been achieved in recent decades. It intersects with several challenges in science education with which stakeholders are concerned. These include representation of contemporary science, education about socio-scientific issues, dual-audience notions of science for all and more science for some, achieving changes in practice, and securing adequate learning outcomes.

The key objectives of this exercise were focused on identifying key conclusions and unanswered questions from the research, characterising and evaluating the range of resources available to support teaching and learning about genomics, and thereby recommending priorities for further research and development of provision. The study identified and analysed approximately 150 papers, published in English between 2000 and 2015 and focusing on aspects of genetics and genomics education for young people between the ages of 9 and 18. Of these papers, 29 were taken to in-depth review.

Aspects identified as being important from the research review include the lack of consensus on what genomics education is intended to encompass or achieve, which contrasts with very clear dissatisfaction with traditional genetics education. There are some concrete views on how genetics education itself needs to develop, particularly by shifting focus away from Mendelism. Recognition of the need to prepare learners for personal and societal decision-making about genomics is not well-balanced with understandings of how to achieve it. There are research gaps, particularly regarding the needs of primary and lower secondary learners for suitable ‘pre-genomic’ learning, the views and needs of teachers, and the implementation and effectiveness of evidence-informed innovation.

The survey of teaching and learning resources examined the National Curriculum and post-16 biology specifications in England, and aligned textbooks, for genomics content; it also compiled and analysed almost 300 freely available resources for genomics teaching and learning. In the textbooks, structured evidence was sought as to whether the contents ranged beyond traditional genetics education. Resource analysis employed two theoretical frameworks, the BSCS 5Es instructional model and a framework (from Miller and Abraham) for analysing the purposes of practical work, to analyse the affordances of each resource.

From the review of resourcing, findings have been identified which complement those from the research review. There are opportunities for specialist providers to influence genomics education policy and practice, to bring both lesson content, textbooks and pedagogy more up to date and to ensure that it is evidence-informed. There has been a tendency for resource provision to focus on a limited range of resource types and formats and to be targeted to a narrow age range of learners. Opportunities exist to promote active learning and effective formative assessment. Providers could work with practitioners and researchers to develop and trial new approaches.

The report includes 24 recommendations for the science education research community, curriculum developers, teacher educators, specialist resource providers and other stakeholders.
SUMMARY OF RECOMMENDATIONS

Recommendations relating to research on genomics education

The research community, including funders, should

1. review the balance between empirical and non-empirical work in the field, to promote more robust study designs, be they qualitative or quantitative, and to promote further research towards solutions to problems;
2. facilitate collaboration and consensus-building in the genomics education research community;
3. address the relative lack of attention that has been given to research with primary and lower secondary age learners and with teachers.

Recommendations relating to the nature and purpose of genomics education

Stakeholders should

4. make efforts to achieve greater clarity about the purposes of genomics education (individually and collectively), aiming if possible to confirm the positions which are emerging; to add detail to establish the necessary and sufficient knowledge, understanding and skills that are required to meet participatory citizenship goals; and to agree what specific additional content is needed for the sub-set of young citizens who will go on to become scientists;
5. as part of the above, seek to establish and articulate a shared understanding amongst stakeholders of what genomics is taken to encompass, in relation to different groups of school-age learners and in terms of content and perspective.

Recommendations relating to curriculum development

Stakeholders should

6. delineate the knowledge, understanding and skills that are necessary and sufficient to meet participatory citizenship goals for all, to fulfil the requirements of those young citizens who will go on to become scientists, and to allow citizens to engage with further information at the point of need;
7. consider the implications of different paces of change in genomics and in education for a core curriculum requirement that is reasonably stable over time;
8. use the channels open to them, including those which could influence specified and assessed curricula, to promote educational adoption of evidence-informed learning progressions, appropriate terminology and contemporary examples of genomics technologies.

Recommendations relating to pre- and pro-genomics education

Researchers, developers and teacher educators should

9. review the research evidence base regarding naïve concepts about genetics and inheritance, with the aim of assessing their impact on genomics learning and thereby considering actions that need to be taken;
10. consider how to ensure that pre-genomics learning in primary and lower secondary schools is pro-genomic, through curriculum and resource development and through teacher professional development for the enhancement of pedagogical content knowledge.
SUMMARY OF RECOMMENDATIONS

Recommendations relating to pedagogical support for genomics and pre-genomics education

Those who develop and support school-level genomics education should

11. engage with the evidence base regarding effective practice in science education for personal and civic engagement with socio-scientific issues;
12. facilitate research to
   • establish the range of ideas that schoolteachers associate with ‘genomics’ and ‘genomics education’ and compare them with those of specialist stakeholders in the field
   • explore the factors influencing teachers’ approaches to teaching genomics, including subject specialism, time elapsed since training and engagement with professional development activities
   • strengthen the evidence base regarding effective pedagogical practice for genomics education, including the development of validated assessment tools;
13. facilitate access to teaching resources that are evidence-informed, congruent with curriculum and assessment models, and that meet teachers’ perceived needs;
14. facilitate access to professional development that supports teacher acquisition and application of subject knowledge and pedagogical content knowledge for genomics education, including approaches to teaching about socio-scientific issues.

Recommendations relating to specialist resource provision for genomics and pre-genomics education

Specialist teaching and learning resource providers should

15. Develop evidence-informed guidance on genomics-related content in preparation for future reform of the school science curriculum, including guidance on sequencing and age-appropriateness of ideas, with a view to influencing policy.
16. Work with publishers and textbook authors to develop appropriate and up-to-date content in textbooks that is aligned with (and helps to define) the intended learning outcomes in the curriculum.
17. Work with science education researchers to develop and trial new resources for genomics education to support teaching of the recommended learning progressions.
18. Work to make existing resources more useful and diverse in type, for example by adding questions to ‘Explain’ resources to facilitate the collection of evidence of learning, and by clearly labelling the resource with information about the target age range and date of last update.
19. Consider the publication of a catalogue of available resources for genomics education, alongside an analysis, in a form that may help teachers to locate resources to fulfil particular classroom needs.
Specific recommendations for YourGenome.org

The team creating and curating YourGenome.org should

20. Add a target age range to resources currently lacking this information, e.g. articles, and check that the target age ranges stated on existing resources are appropriate for students’ conceptual development.

21. Develop new resources targeted at young students up to age 11, and suitable for use by non-science-specialist teachers in primary schools, with the specific aim of supporting pre-genomic learning.

22. Develop new resources to expand the variety of activity types, including activities that involve practical or experimental work and activities that develop students’ numeracy or quantitative data analysis skills.

23. State the learning objective of each resource – i.e. what key concept they are intended to develop or test understanding of, or what key competency they are intended to develop or test.

24. Ensure that new and existing resources:
   a. promote ‘active learning’ (in which students do more than simply receive information);
   b. include, for example, questions that test understanding of the key concept, or tasks that test a key competency, so as to provide evidence of what students have gained from the activity and whether they have met the learning objective.
INTRODUCTION

How should we teach children about genes and genomes, sixteen years after the completion of the Human Genome Project? What should they be learning? What resources are available to support this learning, and to support teachers in securing it? Genomics is a field of science at the forefront of modern biology, concerned with understanding the role of genomes in the development and functioning of organisms and with understanding the structure and evolution of genomes. It utilises cutting edge technologies. In everyday life it is common to encounter news and information about genomes and genomics and about how society interacts with this science.

There is a challenge, therefore, for school science, with its twin aims of securing scientific literacy for all and educating some for careers directly involved with the use and production of scientific knowledge. There appears to be considerable contemporary interest in the teaching of genomics in schools. For example, in England and Wales, ideas about genomes were added to the statutory national curriculum for 14-16-year olds in 2014. This reflects scientific understanding that genes represent a small proportion of genomes, that DNA once thought of as ‘junk’ may have important functions and that complex interactions occur between multiple genes, non-coding regions of the genome and environmental factors in the development and functioning of organisms. The challenge for school science, then, is to move beyond the traditional narrow and atomistic focus on the functions and inheritance of single genes (‘genetics’) and to help learners acquire more sophisticated and holistic understanding of genomes and genomics.

At the onset of this study, there was no clear research-informed understanding in the UK curriculum development and informal science communities of pedagogical or instructional approaches that may help (or hinder) teaching and learning about genomics in schools, of the availability of teaching and learning resources, or of factors influencing their design and use. This study aims to address that gap.

A strategic aim of the Public Engagement team at the Wellcome Genome Campus (WGC) is to engage school children and teachers with genomics through accurate, inspiring resources. The team identified a need to work with education researchers at an early stage in resource design and development, building an evidence base on introducing abstract genomics concepts, informing curriculum development and designing pedagogically sound materials. The University of York Science Education Group (UYSEG) is a leading developer of research-informed science curricula; the group recognises genomics as an emerging area where research is needed to inform development.

The rationale of this collaboration is to survey and analyse research literature in genomics education and similarly to survey and analyse genomics teaching and learning resources. Key findings, conclusions, and recommendations will accrue from what is known and what is available (including the identification of what is not known and what is still needed). This is intended to lead to, and inform, future research in genomics-related education, and to foster research-informed curriculum development work, including the production of next-generation learning resources. It is also intended to complement other efforts to move genetics education forward towards genomics education, for example the stakeholder workshop that led to the ‘Nowgen Manifesto’ (Finegold and Starling, 2012), by summarising, analysing and critiquing the evidence base.
KEY OBJECTIVES

• To complete a landscape review of research regarding teaching and learning of ideas related to genomics, identifying key conclusions and unanswered questions.

• To survey resources available to support teaching and learning about genomics-related biology, identifying archetypes, investigating the extent to which they afford opportunities for supporting research-informed curriculum design and delivery in genomics education, and generating a typology of resources for use in further research and development.

• To recommend further research, resource provision and curriculum design work that may be necessary to optimise learner outcomes in genomics education in England (and Wales) as well as elsewhere.
1.1 IDENTIFICATION OF RELEVANT RESEARCH STUDIES IN THE DOMAIN

1.1.1 Review Strategy

The following steps were used in this adapted (streamlined) version of a systematic literature review strategy:

- Identifying the area for review
- Formulating the research question(s) for the review
- Identifying keywords for searching and for including papers
- Searching for papers which might be suitable for inclusion in the review
- Formulating exclusion criteria for screening
- Screening papers and removing them if they meet the exclusion criteria
- Extracting data from papers
- Applying a score to papers based on lack of bias, quality and relevance of study
- Evaluating distribution of scores in order to identify cut off points for shortlisting and rescreening
- Rescreening to identify high quality papers that should be further reviewed
- Combining shortlisted and rescreened papers to form a set for further review
- Papers subjected to final screening process to identify the papers suitable for in-depth review
- Data extraction from in-depth review set

In conversation amongst the research collaborators, including the Wellcome Genome Campus Public Engagement Team, it was determined that the specific area for review should be published research studies pertaining to the teaching in schools of topics in science closely related to genomics (including precursors and applications). The review would be restricted to studies published in English since 2000 (broadly coinciding with the release of the initial draft of the human genome sequence) and concerning learners from upper primary (aged 9+) to the point of transition into Higher Education (aged 19).

The research questions were formulated as:

- What research has been published in this area?
- What are the collective characteristics of this research in terms of such aspects as geographical distribution, authorship, research design, and research focus?
- What are the key papers in the area, in terms of indicators of the quality of evidence and the perceived relevance to the field?
- What, collectively, are the overall conclusions and recommendations that may be drawn from these key papers?

Keywords were identified for searching databases, formulated amongst the research group and the WGC colleagues. The keywords used are listed in Appendix A1. Together with the definition of the area for review, these constitute inclusion criteria for the literature search.
1.1.3 Identification of potential studies: search strategy

The search strategy for identifying potential studies to be used in the review had three components. The first component was to search the bibliographic databases ERIC, BEI and Web of Science as well as Google Scholar, using the list of keywords. Keywords were used in Boolean searches of the databases, using strings such as: ‘genomic’ AND ‘high school’. Search strings included a scientific component and an educational component from the keyword list. US and UK variants were addressed (for example searching for ‘high school’ and for ‘secondary school’). Not all possible combinations were used systematically – once a stage had been reached where further new searches returned no new articles, this searching strategy was discontinued.

In parallel, the group solicited recommendations from UYSEG and Wellcome Genome Campus Public Engagement team members. Finally, citation and reference searching (‘snowball searching’) were used to identify additional papers from the set already gathered.

The Review Group (RD and DS) set up a database system using EndNote software to keep records of studies found during the review.

1.1.4 Defining relevant studies: exclusion criteria

To be included, a study must NOT fall into any one of the following categories:

Exclusion on scope

1. Not reporting on learning/teaching of genomics within biology
2. Not reporting on teaching or learning in the context of schools
3. Not with a main focus on learners aged 9-undergraduate

Exclusion on setting in which the study was carried out

4. Not published in English
5. Not published in the period 2000-present

Therefore, the exclusion criteria formulated for the review are:

1. Exclusion 1: exclusion on topic (i.e. not relating to the teaching of genomics and closely related science)
2. Exclusion 2: exclusion on context (i.e. not reporting on teaching in schools)
3. Exclusion 3: age (not 9-18)
4. Exclusion 4: language (not English)
5. Exclusion 5: date (not 2000-2015)

1.1.5 Screening studies: applying exclusion criteria to search results

Exclusion criteria were applied to the titles and abstracts of studies found in the search, using the exclusion criteria to decide whether a paper was suitable to be added to the database.

148 papers were identified as potential studies for inclusion in the database. 1 of these studies was excluded because it was unobtainable.

Exclusion criteria were then re-applied to the full papers, and those which did not meet the criteria were excluded (N=35).

The process of searching and screening therefore yielded a total of 112 papers.

1.1.6 Characterising included studies

In order to analyse the papers, the following data were extracted from them and used to populate a ‘Review Pro Forma’ spreadsheet.

- Author
- Year
- Title
- Journal
- Source (e.g. URL)
- Country of Study
- Details of Researchers
• Nature or Category of Study (e.g. experimental, ex post facto, non-empirical)
• Aims of Study
• Research Question(s)
• Age of Learners
• Summary of study design, including details of sample
• Data collection methods, including details of checks on reliability and validity
• Data analysis methods, including details of checks on reliability and validity
• Summary of results
• Conclusions
• Reviewer’s Notes

The reviewer’s notes section was used to add any information regarding the respective paper further to that already encapsulated in the review pro-forma.

As well as undergoing data extraction papers were scored against the following criteria:

• Weight of evidence A (lack of bias in relation to study questions)
  • Judged by assessing the background of the author(s) and purpose for the study
  • 0–2: clear evidence of bias
  • 3–4: definite potential for bias
  • 5–6: some risk of bias
  • 7–8: no reasonable doubt of bias
  • 9–10: no potential for bias

• Weight of Evidence B: appropriateness of research design and analysis
  • Judged by assessing how appropriate the design and implementation of the study is in order to answer the respective research question
  • 0–2: question not answered
  • 3–4: question poorly answered
  • 5–6: question answered partially
  • 7–8: question well answered
  • 9–10: question unequivocally answered

• Weight of Evidence C: relevance of focus of study to review
  • Judged by assessing how relevant the study criteria are to our own review criteria
  • 0–2: irrelevant
  • 3–4: only slightly relevant
  • 5–6: moderately relevant
  • 7–8: strongly relevant
  • 9–10: extremely relevant

Two reviewers (RD and DS) worked on the data extraction and scoring of the 147 papers. In order to calibrate the scores given by reviewers a random sample (N=5; 3.4%) was taken and marked according to the marking guidelines by both reviewers. Reviewers then justified the score they gave to each paper in the sample.

Once data extraction and marking of the 147 papers was complete, a 10% random sample (N=15) was analysed by both reviewers in order to estimate inter-rater reliability.

Correlation of both reviewers’ mean weight of evidence scores for each paper revealed a very strong correlation coefficient of 0.961
(Spearman’s Rank; $p<0.001$) and the two reviewers’ sample scores were not significantly different when tested in a Mann-Whitney U Test ($U=86.5$, $p = .285$).

The process from searching for papers to establishing the in-depth review set is summarised in Figure 1.1. The identification of the set of papers for in-depth review is described in Section 1.3.

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**SECTION 1: RESEARCH EVIDENCE**

1. **Identification of potential studies**

   - Papers identified where there is no immediate screening (e.g., electronic searching where criteria for exclusion is recorded)
   - Citation/reference searching
   - Electronic database searching

   **Potential includes**
   - $N=148$

   **Papers not obtained**
   - $N=1$

2. **Application of inclusion and exclusion criteria**

   - Full document screened
   - $N=147$

   **Papers excluded by age of learners**
   - $N=35$

   **Papers excluded by review score**
   - $N=91$

   **Data protection and scoring of papers**
   - $N=112$

   **Shortlist**
   - $N=21$

   **Papers excluded by score rescreened between a set range**
   - $N=12$

3. **Characteristics**

   - Further review group
   - $N=33$

   **Papers excluded after re-evaluation of the further review group**
   - $N=4$

   **In-depth review**
   - $N=29$

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**Figure 1.1. Flow-chart to summarise the search strategy and outcomes**
1.2 LANDSCAPE ANALYSIS

1.2.1 Weight of Evidence Analysis

Figure 1.2 illustrates the pattern of distribution of the mean Weight of Evidence scores in the ‘long-list’ of 112 papers identified as relevant to this review by the application of inclusion and exclusion criteria to the search results.

18.8% (N=21) of the papers scored highly – between 8.0 and 10.0 – and were immediately put forward for the in-depth review stage. 15.2% (N=17) were considered of low overall quality (below a score of 5.0). The majority of papers (54.5%, N=61) were considered to be of reasonably good quality (scores between 6.0 and 7.9); these were rescreened for potential incorporation into the in-depth review set, as described in Section 1.3.

There were, inevitably, some papers (N=32, 28.5%) where the relevance to the focus of this review on teaching of genomics in schools was relatively poor compared to the lack of bias and the methodological robustness of the study (defined by Relevance Score minus Mean Score < -1). Some of these were included in the in-depth review set by virtue of good overall rating and qualitative judgements at the re-screening and validation stages.

Unfortunately, but also perhaps inevitably, there were some studies (N=9, 8.0%) which were rated as clearly relevant to this review, but where the levels of potential bias and/or low methodological robustness detracted from the overall judgement of value to this review. (This was defined by Relevance Score minus Mean Score > +1). One such study remained in the in-depth review group after re-screening and validation. It is, overall, pleasing that few studies relevant to this field are clearly let down by potential bias or questionable methodology.

1.2.2 Category of Study Analysis

Analysis was performed on the category of study (Figure 1.3). Categories were defined by the reviewers, as listed below:

- Experimental – study involving use of at least one control group
- Intervention, design and evaluation – use of pre/post-tests to quantify learning of content
- Ex Post Facto – use of post-test without pre-test to quantify learning of content
- Empirical – no intervention, but data collected such as survey, questionnaire or interview data
- Non-Empirical – literature without original research
- Review – evaluation of data from multiple studies
- Case Study – study in which data is gathered from a specific cohort on multiple occasions over a period of time
- Teaching resource – focused on the design and/or use of a resource to inform teachers
- Teacher analysis – study focused on teachers as opposed to students
- Resource Analysis – analysis of teaching resources
Some studies could be allocated to more than one category, for example an ex post facto teacher analysis; for the purpose of this analysis, a judgement was made as to the most appropriate first-level category under which to classify such a study.

Almost a third of the papers (28%, N=31) are non-empirical, where no original data have been collected and analysed – in general these represent theoretically-derived arguments and points of view in relation to the field. There are 21 studies (19%) using pre-/post-testing methodologies in the design and evaluation of interventions, and 18 (16%) empirical studies involving surveys, interviews and similar approaches. The other categories are more sparsely represented in the collection of papers, with notably few experiments (methodologically challenging in practice but regarded by many as heading towards a ‘gold standard’ for determining ‘what works’): of the four experimental studies in the set, one is a natural (quasi-) experiment. On the other hand, there are also few ex post facto studies, which are often regarded as methodologically weak (as there is no collection of baseline or other control data). It is perhaps unsurprising that there are very few meta-analytical reviews in this rather young field of study.

1.2.3 Country of Origin Analysis

Country of origin of the studies was also investigated (Figure 1.4). This revealed five dominant countries in this field of research: USA, Netherlands, Australia, UK and Sweden (collectively, N=87, 77.7%). The USA alone has contributed 43.4% of the papers.
It is clear to see when viewing the group of papers collected that a large proportion originate from five main groups of researchers, with some key authors from within these groups highlighted. For example, all of the papers from England feature Jenny Lewis as an author, amongst other associates mainly from the University of Leeds.

A similar pattern emerges when looking at papers from Australia where Grady Venville is an author on 7 out of the 9 papers. In the Netherlands 10 out of the 11 papers originate from the Freudenthal Institute for Science and Mathematics Education group at Utrecht University. Key authors from this group are Arend Jan Waarlo and Dirk Jan Boerwinkel. Of the 7 papers from Sweden, Niklas Gericke is an author on all. Gericke is also one of the few authors to collaborate internationally several times, contributing to international papers with authors from the USA and Brazil.

In regard to the most prolific country, the USA, Ravit Golan Duncan and Mike U. Smith were the only authors to feature more than twice in the review. Despite producing nearly five times more papers in this field than the next most productive country, there is little collaboration between researchers from the USA, unlike that seen in the Netherlands – presumably this is at least in part attributable to geography.

1.2.4 Age of Learner Analysis

One of the key pieces of information recorded in the review pro forma was the age of the learners in each study (Figure 1.5). A large proportion of the papers that were included were focused on learners of high school age.

The papers were selected, through inclusion and exclusion criteria, on the basis of being focused on learners aged 9-18. However, two points emerge as noteworthy.

1. There is some focus on younger primary-aged learners.
2. The great bulk of the attention has been on learners in upper secondary education (14+) and on post-compulsory (but pre-tertiary) education. This is despite the hypothesis that there are valuable opportunities to teach younger children about genomics-related science, from a scientific literacy point of view.
1.3 METHODS FOR THE IN-DEPTH REVIEW

1.3.1 Identification of studies for in-depth review

The mean weight of evidence score was used to rank the 112 long-listed papers (a mean score of 10 being most valuable to the study). The distribution pattern of these scores (Figure 1.2) allowed visualisation of the natural cut off points for the further review group to be identified. In this fashion, papers scoring 8.0 and above were immediately shortlisted (N=21).

Papers scoring means of ≥ 6.0 and < 8.0 were rescreened. Reviewers (JA, RD and DS) chose to retain further papers (N=12) which they deemed valuable to the study. Factors considered were:

- strong scores for **Weight of Evidence C: relevance of focus of study to review** in comparison to the mean score
- weak scores for either **lack of bias or design and analysis** in comparison to the mean score
- papers felt to be important or noteworthy for other reasons (such as widely cited papers).

These two groups were then combined to form a combined group for a further validation review (N=33). After validation of this combined group by the wider research team, four papers were removed, and the in-depth review set was decided upon (N=29). The four papers removed at this stage were assessed as being of insufficient relevance to the focus of this landscape review, despite being strong in at least one other aspect.

1.3.2 Data extraction from the in-depth review set

Qualitative data were extracted from the 29 papers in the in-depth review set by close reading and recording of notes about each paper using pro-forma. The pro-forma was bespoke – designed specifically for this review.

Observations and comments were noted for each paper in relation to the following aspects:

- Definitions of genomics (implicit or explicit)
- Goals of genomics education (implicit or explicit)
- Key assumptions made by the authors (implicit or explicit)
- Major findings and conclusions
- Key recommendations
- Key strengths
- Notable weaknesses
- Any further work proposed
- Other points

Early in the detailed review process, it became clear that an additional aspect was also relevant and important:

- **Aims, objectives and research questions of the empirical work reviewed**

An informal thematic analysis was then undertaken to identify common threads that can be identified within these aspects. No a priori theoretical framework was imposed on this thematic analysis, beyond the headings above; rather, in the spirit of a grounded theory approach, common themes were constructed from the analysis. Dissenting ideas – points that seem at odds with any emerging consensus – were also noted.

The thematic analysis is qualitative – quantitative data (for example frequency counts for particular points) were avoided as being invalid in a heavily selected sample, and where minor or dissenting views may nonetheless be important and insightful.
1.4 RESULTS AND ANALYSIS OF THE IN-DEPTH REVIEW

1.4.1 Quantitative analysis

The subset of 29 papers for in-depth review was analysed in terms of the same features used to characterise the ‘long-list’ set. Results are shown and discussed in the following five subsections, examining weight of evidence, study types, countries of origin, learner age ranges, and title key words.

Following the characterisation of the in-depth review set, the qualitative analysis is reported and emergent themes from the 29 papers are discussed.

1.4.1.1 Weight of evidence

The distribution of mean Weight of Evidence scores for the in-depth set is shown in Figure 1.6 below, for comparison with Figure 1.2.

Figure 1.6 illustrates how the in-depth review set is, as intended and expected, biased in favour of those papers scoring more highly. A small number of papers were included despite having low overall scores, as described in section 1.3. Table 1, below, emphasises this point by showing the percentage of papers in different sections across the spread of mean Weight of Evidence scores that were retained between long-list and short-list.

<table>
<thead>
<tr>
<th>Mean Weight of Evidence score range</th>
<th>Retained papers from the large set (N=112)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 6.00</td>
<td>0%</td>
</tr>
<tr>
<td>6.00–6.74</td>
<td>7% (n=2)</td>
</tr>
<tr>
<td>6.75–7.49</td>
<td>19% (n=4)</td>
</tr>
<tr>
<td>7.50–8.24</td>
<td>48% (n=10)</td>
</tr>
<tr>
<td>8.24–8.99</td>
<td>100% (n=7)</td>
</tr>
<tr>
<td>9.00–9.74</td>
<td>100% (n=6)</td>
</tr>
<tr>
<td>≥9.75</td>
<td>(no papers)</td>
</tr>
</tbody>
</table>

Table 1: retention into in-depth review group related to Mean Weight of Evidence

As confirmed in Table 1, all papers in the long list scoring a Mean Weight of Evidence above 8.0 were retained, with the exception of one study by Venville and Donovan (2008). This paper scored 8.0 but was felt to be only peripherally relevant although trustworthy and robustly designed as a set of multiple case studies of the use of a wool model for DNA/genes/chromosomes in use with pupils of different ages.
1.4.1.2 Category of Study

The categories of study represented in the in-depth review set are displayed in Figure 1.7, below, for comparison with Figure 1.3.

Notable differences between the long-list and in-depth list characteristics indicate that resource analyses were particularly likely to be taken through for in-depth review, as shown in Table 2. Four out of the five resource analysis papers taken through had Mean Weight of Evidence scores below 8.0; they were retained because of specific features of interest in the papers.

<table>
<thead>
<tr>
<th>Study Category</th>
<th>Retained papers from the large set (N=112)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Resource Analysis</td>
<td>50% (n=5 out of 10)</td>
</tr>
<tr>
<td>Empirical</td>
<td>39% (n=7 out of 18)</td>
</tr>
<tr>
<td>Non-Empirical</td>
<td>32% (n=10 out of 31)</td>
</tr>
<tr>
<td>Experimental</td>
<td>25% (n=1 out of 4)</td>
</tr>
<tr>
<td>Teacher Analysis</td>
<td>25% (n=1 out of 4)</td>
</tr>
<tr>
<td>Case Study</td>
<td>18% (n=2 out of 11)</td>
</tr>
<tr>
<td>Intervention, Design and Evaluation</td>
<td>14% (n=3 out of 21)</td>
</tr>
<tr>
<td>Teaching Resource</td>
<td>0% (n=0 out of 9)</td>
</tr>
<tr>
<td>Ex Post Facto</td>
<td>0% (n=0 out of 3)</td>
</tr>
<tr>
<td>Review</td>
<td>0% (n=0 out of 1)</td>
</tr>
</tbody>
</table>

Table 2 Differential retention rates of studies in relation to study category.

On the other hand, case studies, intervention designs and evaluations, teaching resource design and use, and ex post facto studies were relatively unlikely to be taken through to full review. Their Mean Weight of Evidence scores tended to be depressed as a result of perceived weaknesses in design or queries about the objectivity or independence of the study in relation to the evaluation of interventions and resources.

However, overall, chi-squared analysis indicates no statistically significant difference between the two distributions (long list and shortlist), indicating that differences are within the range that would be considered reasonably probable due to chance alone when selecting a 26% sample of this size.
1.4.1.3 Country of origin of study

The countries of origin of the 29 studies for in-depth review are displayed below in Figure 1.8, for comparison with Figure 1.4. There is a roughly similar distribution. The major countries and centres of research are represented in the in-depth set. It is notable that no international studies (i.e. papers where there is clear multinational input to both the research and the authorship) were carried into the in-depth review set. However, this is a categorisation artefact: there are several studies in the detailed review that have an international flavour to them, most often in the form of non-empirical studies that draw on international perspectives, including conference proceedings.

The dominance of the USA as a focus of research activity may appear to be somewhat challenged, when focusing on the most relevant and/or highest quality research. However, chi-squared analysis again indicates no significant difference between the two distributions. The in-depth set is broadly representative of the long list.

1.4.1.4 Age of Learner

Figure 1.9, below, builds on Figure 1.5 by showing the age-of-learner profiles of the long list and the shortlist sets side-by-side. The figures illustrate that the two distributions are very similar, and therefore that the age-range foci of the studies in the in-depth review are broadly representative of the field in general. Chi-squared analysis gives no cause to reject the null hypothesis that the two distributions are equivalent.
1.4.1.5 Title Key Words Analysis

An analysis of the titles of the 29 papers in the in-depth review set reveals some noteworthy points about the collective focus of this body of work.

The most common word stem in the titles is “genet**” indicating genetic or genetics (45% of the papers, n=12) – not including the one paper with the word epigenetics in its title. “Genom**” – indicating genome, genomic or genomics – is common, but less so (28%, n=8); gene or genes is equally frequent. It seems unsurprising that the authors tend to situate their papers clearly in the general area of genetics, but for the purposes of the current review it is noteworthy that the papers are not necessarily flagged as concerning genomics, and that a focus on difficulties with the gene concept is evident in the titles as well as the content.

School is referred to in 24% (n=7) of the titles, with understand or understanding(s) in 21% (n=6). The latter reflects the inclusion of several papers focused on the security of related learning outcomes.

More sparsely included words are: curricula or curriculum; teaching, learning and education; young people; textbook. These are each included in three or four titles (10-14%). This was unexpected given the focus on education.

Words in only one or two of the 29 titles include: child (or children) and teacher; standards and progression (as in ‘learning progression’); health; concept and misconception. This reflects the diversity of this body of literature as well as the dominance of some research foci, as discussed below.

Quantitative key word analysis of the titles is consistent with qualitative thematic inspection of the aims, objectives and research questions of the empirical studies reviewed, discussed below.

1.4.2 Qualitative analysis

Data were extracted from the 29 papers under the headings below:

- Definitions of genomics (implicit or explicit)
- Goals of genomics education (implicit or explicit)
- Aims, objectives and research questions
- Major findings and conclusions
- Main recommendations
- Key assumptions made by the authors (implicit or explicit)
- Prominent strengths and notable weaknesses
- Any further work proposed
- Other points.

Following a brief summary of the major empirical studies, in the in-depth review, Emergent themes under corresponding headings are discussed in the subsequent sections. In section 1.4.2, cited examples are from the 29 papers reviewed in depth, unless otherwise noted by *. Not all of the papers under review contribute to each of these headings – for example, many of the papers give no definition of genomics and/or no clear statement of the goals that they would propose for genomics education.

1.4.2.1 An overview of the major empirical studies in the in-depth review set

Investigating the state of English school leavers’ knowledge in the 1990s, Lewis and colleagues surveyed children at the end of KS4, revealing widespread misunderstanding of key ideas in genetics and inheritance (Lewis, Leach, & Wood-Robinson, 2000a, 2000b; Lewis & Wood-Robinson, 2000; Wood-Robinson, Lewis, & Leach, 2000). For example, 50% were unsure that all living things contain genetic information; there was confusion between terms such as gene, chromosome, and allele and between genetic code (universal) and genome (unique); they were largely unaware that genes can be switched on and off. Lewis and Kattman (2004)*, having investigated similar difficulties amongst German high school students, offer an explanation in terms of strongly-held naïve ideas and misconceptions.
Revisiting the English survey 16 years later, Lewis (2014) found that things seemed to have improved – but only partially. Explanations remained poor amongst this 2011 cohort and genetic code-genome confusion was still rife. The young people held a traditional view of genetics, with little awareness of genomics. Lewis asks to what extent this should cause concern, suggesting that it will take time for genomics to permeate through to the curriculum, and advocating consideration of pedagogy and assessment as well as curriculum content.

Smith and Williams, in Edinburgh, have demonstrated that children may acquire significant knowledge of genetics and inheritance by the age of 10 (Smith & Williams, 2007). The knowledge was acquired through their families and focused on kinship ideas within their families (including any pets!). These ‘naïve’ ideas proved very resistant to change in 10-14-year olds. The authors suggest that 7-10 may be a receptive age range for teaching about inheritance concepts. They also call for more evaluative intervention research on ways of shifting children’ s understanding of concepts in genetics/genomics.

The studies cited above (and included in the in-depth review set), from Lewis and her collaborators and from Smith and Williams, are part of a larger body of work from these researchers and others, characterising naïve ideas and misconceptions that young people may hold about matters relating to genetics and genomics. Such work includes papers from Donovan and Venville (2012)* and Witzig, Freyermuth, Siegel, Izci, and Pires (2013)* which indicate that children may, for example, have difficulty distinguishing genes from traits; regard genes as particles that carry traits; do not connect genes and DNA; and regard DNA as alive.

Addressing coverage of modern genetics content in required curricula, two reviewed studies have scrutinised high school standards from across the USA. Wefer and Sheppard (2008) used an unconventional definition of genomics in relation to bioinformatics and surveyed the standards for references to bioinformatics and component ideas. They found that the term was not directly used, but that related ideas were variously present – the least frequent being about the Human Genome Project/genomics. They go on to criticise “ambiguous and overgeneralised” standards, and recommend that aspects requiring moral and ethical consideration, in particular, should be more tightly prescribed (including in the manner in which students should explore them). Dougherty, Pleasants, Solow, Wong, and Zhang (2011) come to very similar conclusions about the failure of state standards to keep up with genomics. In particular they complain that the most weakly covered aspects are those which are becoming increasingly relevant in a post-HGP era and that instruction should be focused on complex (rather than single-gene) traits.

One piece of evaluative research in the USA, has concerned two independent ‘learning progressions’ that are in use in the USA in the domain of genetics. One concerns Mendelian inheritance; the other concerns the ‘central dogma’ of molecular genetics, i.e. the relationship between DNA, gene products (proteins), and function (phenotype). Duncan, Castro-Faix and Choi, working with 10–13-year olds, tested whether the order in which these were taught was associated with differences in learner outcomes. They found that teaching molecular genetics first may support or ‘bootstrap’ learning of Mendelian inheritance, but not the other way round. So, teaching sequence can make a difference (Duncan, Castro-Faix, & Choi, 2014).

Linking to the current report’s landscape review of resourcing, in a US study of high school biology textbooks (post-2005), the authors (Hicks, Cline, & Trepanier, 2014b) found that mentions of ‘classic’ chromosomal single-gene disorders were 2.5 times more frequent than mentions of five common adult-onset multifactorial disorders (cancer, hypertension, cardiovascular disease, diabetes and Alzheimer’s disease). Discussion of genetic/hereditary factors in the complex conditions was scant. These conditions are far more common than single-gene disorders and may be open to personalised medicine and environmental modulation of genetic risk – in such ways, they represent prime themes and contexts for genomics education, and illustrate why aspects of genomics education are considered desirable for all.

Also focusing on textbooks as key knowledge mediators that support teaching and learning, two research groups represented in the sample have examined textbooks’ use of gene concepts (dos Santos, Joaquim,
& El-Hani, 2012; Gericke & Hagberg, 2010b). Gericke and Hagberg found that textbooks for 16–19-year-old students (typically post-compulsory learners of biology) in Sweden, the UK, the USA, Australia and Canada use gene concepts in potentially unhelpful ways. There is, typically, fluidity between various different meanings of ‘gene’, ambiguity, contextually inappropriate usage and a paucity of more modern (process-orientated, less deterministic, less reductionist) concepts in the way genes are conceptualised. Dos Santos, Joaquim and El-Hani scrutinised Brazilian textbooks for a slightly different age range (14–18-year-olds) but found, similarly, that textbooks tend to confuse and ‘hybridise’ a variety of historical concepts of ‘gene’ that in fact serve different purposes. They posit that no single unitary gene concept exists that is adequate for all purposes.

1.4.2.2 Definitions of genomics

Haury & Nehm (2012) bemoan the opacity of the term genomics and the lack of clarity about what makes it a unique area of biology, and they cite the evidence of Duann & Nehm (2010)* that the content considered by various authors to be characteristic of genomics varies widely.

Dougherty, Pleasants, Solow, Wong, & Zhang (2011), in their paper related to the teaching of what they term modern genetics, claim that “genetics/genomics” has a set of fundamental concepts at its intellectual core. They describe these concepts in relation to goals of genomics education. This raises three issues that also permeate the other papers under review: distinguishing genomics from genetics; establishing what genomics does and does not encompass; understanding the relationships between definitions of genomics and the aims ascribed to genomics education.

Firstly, there is a general elision between genetics and genomics – indicating that genomics tends to be regarded as contiguous with genetics, perhaps either a development or extension of it, rather than as something distinct. Many of the reviewed papers are essentially critiques of genetics education in a post-genomic age.

Secondly, there are claims or at least intimations of some essential and perhaps distinctive ideas that collectively constitute genomics, without a clearly or consistently defined consensus as to what these ideas are. Where authors state, or imply, definitions of genomics these vary markedly, particularly in what they include or exclude. Some broadly common features are ideas of continuous rather than discontinuous variation; multiple influences on phenotypes, including environmental factors; differential gene expression; non-deterministic genetic influences; acknowledgement that not all DNA is ‘genes’. There is a general theme of complexity in relation to traditional focuses on discrete variation and Mendelian inheritance (single-gene; dominance model). Some of these common features are also less about new topics or additional content than they are about new ways of understanding or interpreting entities or phenomena – for example taking a more nuanced approach to the relationship between DNA and genes than a classical ‘beads on a string’ model.

A related observation across the papers is variation in what is included in (or excluded from) their consideration of biology related to genomics. In particular, technologies such as genetic engineering, cloning and gene therapy, and their associated socio-ethical issues, are included in some papers but not others, reflecting slightly different emphases. For example, Lewis (2014) includes stem cells in her paper concerning understandings of gene technology, whereas Mills Shaw, Van Horne, Zhang, & Boughman (2008) specifically exclude stem cells in their paper on misconceptions in genetics. Some technologies, such as direct to consumer genetic testing (DTCG testing), are more typically included than others, presumably either because of their potential to be directly relevant to learners’ own lives in a relatively short time-span and/or because of their close theoretical relationship to central ideas of what a genome is and how phenotypic variation occurs.

Similarly, van Eijck (2010), in arguing for the importance of learners gaining a process-based appreciation of the dynamics of science (where the distal goal is scientific literacy), discusses ‘spin-off disciplines’ from genomics such as bioinformatics, proteomics and systems biology. Verhoeff, Boerwinkel, & Waarlo (2009) regard bioinformatics, for
example, as a discipline within genomics.

Such contrasts reveal possibilities for taking relatively broad or narrow approaches to defining ‘genomics’ for curriculum purposes.

One might anticipate that the goals of genomics education would stem from an understanding of what genomics is, but this relationship can be more reflexive. The third issue exemplified in Dougherty et al. (2011) is that the nature of genomics and the goals of genomics education are intertwined. These authors exclude, for example, epigenetics, the regulatory roles of small RNAs, and chromatin remodelling. Their grounds are that these topics are too complex, too unfamiliar to teachers, too peripheral to what is needed for basic ‘genetic literacy’ and too uncommon in various secondary school curricula (this latter point would be a somewhat tautologous argument if one were looking towards a future, improved curriculum).

Such flexibility in what genomics is taken to be might be regarded as understandable in a relatively young field. However, it presents a difficulty when considering education for or about genomics, as it is not clear that different researchers, commentators and perhaps some stakeholders have a fully shared understanding of the matter under discussion.

The establishment of a shared understanding amongst stakeholders of what genomics is taken to encompass, in relation to school-age learners and in terms of content and perspective, could help to move practice in genomics education forwards. An alternative view could be that the crucial process is to establish shared goals for education that reflect contemporary science and/or technology relating, in some broad sense, to genomes.

1.4.2.3 Goals of genomics education

Intertwined with the matter of what genomics is understood to encompass is the question of what schoolchildren should learn about it. Several, but again not all, of the papers in the detailed review group address this, and some offer answers to the question.

The vagueness of some curricula or standards for science education in relation to learning about genes and genomes attracts some comment, for example in the USA (Mills Shaw et al., 2008) and in Sweden (Thörne & Gericke, 2014). The former complain that some standards do not ensure that students are taught “even the most basic concepts”, for example polygenic inheritance and environmental influence.

Where the reviewed papers include suggestions about what should be taught and why, the phrase ‘genetic literacy’ is used by several authors (for example Knippels, Waarlo, & Boersma, (2005a). The phrase is reminiscent of ‘scientific literacy’, seen by many as an important goal of school science (Millar, (1996)*; Millar & Osborne, (1998)*). Genetic literacy is not in itself a complete statement of the goals of education in the field, as its components are not consistently, or even necessarily clearly, set out. Thörne & Gericke (2014) note a need to educate people appropriately for the biotechnology age, but this again raises a question of what specific knowledge, skills and dispositions that need implies.

A tension exists around the dual-audience approach to science education, as set out for example by Millar & Osborne (1998)*, which notes twin imperatives of educating all young people for citizenship in increasingly technological societies, and educating a subset of these young people to take up careers that directly use and/or generate scientific knowledge. The latter group need all that the former need from their science education, plus more. Accordingly, discussions of goals in science education raise the question of whether the constituency served is seen as the larger group of future citizens, or its smaller sub-group of future ‘scientists’. The tension between these is apparent across the 29 papers, where there are implied or explicitly stated goals for genomics education that are related to one audience or the other.

A specific question arises about precisely what based scientific conceptual content is required to enable the achievement of aims related to citizenship, such as the ability to understand and make informed decisions about technologies that may have an impact on one’s life – personalised medicine, for example. It is not necessarily clear what it means to be ‘adequately prepared for decision-making’
Hicks, Cline, & Trepanier (2014b) treat active participation – here, in healthcare – as a goal and seek a public who are educated about related applications of genomics, implicitly prioritising utilitarian considerations over their theoretical bases. Verhoeff et al. (2009) advocate preparing both constituencies of young people for ‘social participative discourse’ about genomics and its personal and social impacts, noting that a ‘basic’ view of the genome is needed. But how basic is basic, and how is this to be reconciled with positions exemplified by Smith (2014) that a ‘good’ understanding of genetics will be needed to function in an era of personalised medicine? Lewis, in Boerwinkel and Waarlo (2009) (p48) notes that it would be very useful to have research data on the capacity of 11-16 students to understand the relevant content. Notwithstanding individual differences between students in their apparent capacities for learning, and Bruner’s (1960) notion of a spiral curriculum in which “any subject can be taught in some intellectually honest form to any child at any stage of development”, Lewis’ comment seems valid.

Various authors, notably Amber Todd, Michael Dougherty and their collaborators (for example, Dougherty, (2009); McElhinny, Dougherty, Bowling, & Libarkin, (2014)*; Todd & Kenyon, (2015)) set out learning progressions, which subsume sequenced steps in understanding. What is not always immediately evident is any underlying direction, aim or purpose in these progressions. There may be different interpretations of “learning progression” in use by different authors (as descriptive or predictive of how learners actually learn, or as statements of intent about how or what they should learn). Duncan and Hmelo-Silver (2009)* provide a useful explanation of the nature of learning progressions.

One repeated theme across the papers is that of challenging overly simplistic understandings of inheritance, the origins of variation and the nature of genetics as a discipline. In particular, there is a desire to guide learners away from the deterministic models of inheritance that may be reinforced by simplistic presentation of Mendelian models in textbooks and by teachers’ own conceptualisations (Forissier & Clément, 2003)*. Mills Shaw et al. (2008) note that deterministic thinking poses a risk to the achievement of public health goals. Less deterministic appreciation of multifactorial influences (polygenic inheritance, environmental influence and gene-environment interactions, possibly including epigenetics) is preferable. Examples of papers highlighting this goal are dos Santos, Joaquim, & El-Hani (2012) and Venville & Donovan (2005); the inclusion of epigenetics is proposed by Drits-Esser, Malone, Barber, and Stark (2014). Roseman et al. (2006)* and Dougherty (2009) advocate
an “inverted curriculum” in which more complex (polygenic) inheritance is encountered by learners before more simple models, as a means to achieve this goal. This idea can be traced through the work of Todd & Kenyon (2015) and Smith & Gericke (2015).

In her paper comparing two cohorts of young people 15 years apart, Jenny Lewis noted that trickle-down of new science into school curricula is a slow process, taking in the order of 15 years (Lewis, 2014). Lewis also observed that learning of less contemporary material has not generally become more secure with the passage of time. If one takes the view apparently espoused by Wefer & Sheppard (2008) that one goal is to represent rapidly proliferating new science to upper secondary students, such a slow trickle-down is likely to be unsatisfying. A debatable point, therefore, is whether stakeholders should be content with such gentle trickle-down, balancing a desire to meet anticipated needs of learners with recognition of the demands on teachers, teacher educators, and curriculum developers of more rapid change.

In parallel with a lack of shared understanding of what genomics constitutes, there is not yet a settled position in the biology education community of the purposes of genomics education. There are, however, some clear themes which appear to be emerging. Firstly, there is recognition that all young people should learn, in school, some aspects of modern genetic biology for the purpose of supporting their engagement in personal, social and civic decision-making about related issues, now and in the future. That does not preclude some young people also learning more than this, to support their future work as scientists (in a broad sense). Secondly, in the education of young citizens, matters related to knowledge and understanding about human health are prioritised. Thirdly, this is taken to include two broad areas of content: a non-deterministic appreciation of multi-factorial variation and inheritance; some consideration of ELSIs associated with the applications of genomics

Beyond this, there are few specific details on which the genetics/genomics education academic community appears to have settled. One such point is that the ‘gene concept’ is problematic in education and requires some re-evaluation. A second point is that there are sequencing issues in the traditional canon of genetics teaching – specifically that teaching first about discontinuous variation and single-gene (Mendelian) inheritance is not helpful for meeting the emerging goals of genomics education. These are both essentially pedagogical points, made to support teaching that could meet its intended outcomes.

It could be helpful, in order to continue improving school-level genomics education, to make efforts to achieve greater clarity about its purposes, aiming to confirm the positions which are emerging, to add detail to establish the necessary and sufficient knowledge, understanding and skills that are required to meet the participatory citizenship goals, and to consider what specific additional content is needed for the sub-set of young citizens who will go on to become scientists. To aid this, engagement with the wider evidence base regarding effective practice in education for personal and civic participation in socio-scientific issues is likely to be useful.

1.4.2.4 Aims, objectives and research questions of the empirical studies reviewed

Those studies in the sample under detailed review which set out to collect and analyse data did so for a variety of reasons. However, these rationales cluster into a smaller set of overlapping themes, which are:

• to evaluate curriculum requirements;
• to characterise learners’ alternative or misconceptions;
• more specifically, to explore difficulties with representation of gene concepts to learners;
• to evaluate the quality of resources for learning, specifically textbooks and teacher talk;
• to support the development of learning progressions.

Focusing on the extent to which curricular requirements promote learning of contemporary genetic/genomic content, studies by Wefer and Sheppard (2008) and Dougherty and colleagues (Dougherty et al., 2011) both scrutinised the science standards in States of the USA for
inclusion of bioinformatics and modern genetics, respectively. Wefer and Sheppard used a very broad definition of bioinformatics, which encompassed “Human Genome Project/genomics” (the least well represented aspect, in their findings).

Further framing genomics education as problematic, several studies in the reviewed set collectively set out to reveal ideas in genetics that learners find difficult, the impacts of this on learning outcomes, and some of the conceptual gaps and barriers. Lewis and colleagues (Lewis et al., 2000a, 2000b; Lewis & Wood-Robinson, 2000) did so in order to provide base-line data about cohort-level learner outcomes at the end of compulsory science education. In 2014, Lewis published a follow-up which set out to investigate any changes in this baseline over ten years (Lewis, 2014). In the intervening period, Duncan and Reiser explored reasons why molecular genetics appears to challenge learners, focusing on the difficulty of reasoning across ontological levels (physical and informational) (Duncan & Reiser, 2007). Marbach-Ad and Stavy had pursued a similar line of enquiry when critiquing the genetic explanations offered by learners at different stages (Marbach-Ad & Stavy, 2000); they cite Shayer’s claim (Shayer, 1974) that pre-16 students’ cognition is insufficiently mature to support understanding of complex genetic explanations. Knippels and her colleagues, in deriving design criteria for genetics learning progression, took learners’ and teachers’ views on the sources of difficulty (Knippels et al., 2005a), whereas Mills Shaw and colleagues characterised high school students’ genetic misconceptions by analysing their extended writing (Mills Shaw et al., 2008). Smith and Williams (2007) set out to track the quality and quantity of children’s genetic knowledge from early primary into secondary schools, focusing on the acquisition and marked persistence of naïve concepts.

Amongst several researchers commenting on difficulties with the definition of ‘gene’ are Gericke (Gericke & Hagberg, 2010b) and dos Santos et al. (2012) who considered the impacts of this on school genetics teaching, as mediated by textbooks. In papers not in the detailed review set, Gericke and Hagberg (2010a)* describe the impact of textbook authors’ use of mixed models on learning outcomes as “conceptual incoherence”, and Gericke, Hagberg, dos Santos, Joaquim, and El-Hani (Gericke, Hagberg, dos Santos, Joaquim, & El-Hani, 2014)* collaborated to review gene ideas in textbooks across six countries. Venville & Donovan (2005) set out to inform their reflections on school curricula and pedagogy by collating expert views on the gene concept and how it should be taught.

Further to the critiques of textbooks’ use of gene concepts, Hicks et al. (2014b) set out to assess how well school textbooks addressed health-related genetics by contrasting the rarity of references to complex multi-factorial diseases to their focus on single-gene Mendelian disorders. Thörne and Gericke (2014) considered teacher talk as a potential barrier to learning, asking whether teachers’ language bridged the ontological levels between genes and traits.

A small number of papers in the set focus on the development of learning progressions for genetics/genomics. As noted above, Knippels and co-researchers set out to derive design criteria for a genetics learning progression (Knippels et al., 2005a). Duncan, Castro-Faix, and Choi (2014) studied the relative merits of two different sequences of learning (molecular genetics before or after Mendelian inheritance), building on the learning progression proposal from Duncan, Rogat, and Yarden (2009)*. Todd and Kenyon (2015) captured data to help validate and refine a related progression; Todd has gone on to publish further work on this (Todd & Romine, 2017; Todd, Romine, & Cook Whitt, 2017)*.

1.4.2.5 Major findings and conclusions of the reviewed work: a synthesis

a) Curricula

Concepts related to genomics and modern genetics, such as the interplay of genes and the environment in complex traits, are not well represented in curricular standards, at least in the USA and Sweden. Where they are represented, they tend to be loosely specified, without strong frameworks for consideration of socio-scientific issues and with little prescription regarding content or pedagogical approach. Curricular overcrowding impedes the introduction of new science.
There is no longer a good reason to maintain the historical sequence of topics in syllabuses. An overemphasis on Mendelian inheritance leads to poor learning outcomes including over-simplified deterministic thinking. The curriculum should be inverted to place molecular genetics first (which can support subsequent learning about inheritance), to emphasise multifactorial variation and to highlight the roles of gene regulation and of proteins in connecting genes to cells to traits at the organismal level.

These conclusions have been synthesised from Dougherty (2009); Dougherty et al. (2011); Duncan et al. (2014); Hicks, Cline, & Trepanier (2014a); Hicks et al. (2014b); Mills Shaw et al. (2008); Thörne & Gericke (2014); Verhoeff et al. (2009); Wefer & Sheppard (2008).

b) The gene concept as presented to learners is problematic

Different, historically appropriate concepts of ‘gene’ serve different purposes, as noted for example by dos Santos et al. (2012). No single unitary concept exists that encompasses all meanings and uses associated with the word and that is also appropriately simple for introductory courses. In schools, several different concepts of gene are regularly used with little evident reflection on the associated tensions – for example a chemical definition, a physical definition, a genetic information definition and a particulate-inheritance definition. Many school textbooks use gene definitions that are potentially confusing hybrids; more modern ideas about genes (process-oriented, less deterministic and reductionist) are only rarely invoked. Privileging of Mendelian gene concepts may lead to students holding exaggerated and deterministic beliefs about the roles of genes in variation and inheritance. Instruction about genomic and environmental co-construct of phenotype is necessary.

These conclusions have been synthesised from dos Santos et al. (2012); Gericke and Hagberg (2010c); Meyer, Bomfim, and El-Hani (2013); Smith (2014); Smith and Adkison (2010); Venville and Donovan (2005).

c) Other conceptual challenges

The cognitive demand of some ideas in genomics education may exceed the cognitive maturity of many pre-16 learners. The attainment of understanding of molecular genetics concepts is challenging for reasons including dynamic processes at different organisational levels (from macro to micro), abstraction, complex vocabulary and – specific to this topic – different ontological levels (informational and biophysical). The relationships between genes, proteins and traits are seldom clear to learners, a situation which can be exacerbated by teacher talk.

It is important to start with matters of personal and societal relevance to learners, which are concrete and motivating – such as their own families. However, many young people acquire naïve concepts about inheritance outside school, often from and about their own families, or from the media, and these can be very persistent.

These conclusions have been synthesised from Duncan and Reiser (2007); Knippels et al. (2005); Marbach-Ad and Stavy (2000); Smith and Williams (2007); Thörne and Gericke (2014).

d) Learning Outcomes

Many conceptual, curricular and pedagogical challenges act as barriers to successful learning outcomes in genomics education. The body of research, including that published by Lewis and colleagues, constitutes an on-going warning about the low security of learning outcomes regarding the understanding of fundamental genetics concepts. This includes ideas that are well enshrined in curricula, such as single-gene inheritance patterns. Familiarity with terms is not equivalent to correct understanding of their meaning, and some metaphorical terminology (such as ‘barcode’ and ‘fingerprint’) may be understood literally. There may be a tendency to exaggerate the benefits and risks of genomic technologies (‘genohype’).
Teaching may result in fragmented knowledge of ‘facts’ in the absence of explanatory conceptual frameworks that help learners make sense of counterintuitive ideas. Explicit teaching of such frameworks is required and could be informed by clear learning progressions.

These conclusions have been synthesised from Lewis (2014); Lewis et al. (2000a, 2000b); Lewis and Wood–Robinson (2000); Marbach–Ad and Stavy (2000); Mills Shaw et al. (2008).

e) Ways forward

There have been diverse and dispersed interventions aimed at enhancing teaching and learning about modern genetics/genomics. However, as yet there has been nothing systematic on a large scale. There is a need for partnerships between teachers, genome scientists and others. There appears to be some appetite for change; a clear vision and guidance could add potency.

The context and relevance of content to learners (personally and socially) are important, as they can motivate learning – families seem a good place to start. Interventions such as mobile DNA labs can have positive affective outcomes but typically lead to only modest cognitive outcomes with regards to concepts or socio-ethical discussion.

Learning progressions are emerging which have some empirical support for their validity and for their usefulness in helping to secure stronger learning outcomes. These vary in their breadth and in the target learner age range. There is increasingly clear empirical and theoretical support for an ‘inverted’ genetics curriculum, as noted previously.

There are some alternative or complementary views in the literature. Alignment of genomics education more closely to health education is advocated by some, as is the use of genomics as a context for the study of the dynamic nature of scientific activity.

These conclusions have been synthesised from Drits–Esser, Malone, Barber, and Stark (2014); Haury and Nehm (2012); Knippels et al. (2005); Todd and Kenyon (2015)*; Todd et al. (2017); van Eijck (2010); Verhoeff et al. (2009); Zusevics et al. (2014).

1.4.2.6 Main recommendations from the reviewed work

A number of recommendations for action feature prominently and in several cases repeatedly, though with some variations, in the work under review. These are synthesised below.

New frameworks for learning are needed as genomics becomes increasingly practically relevant to citizens. Standards need to specify, guide and exemplify good practice. Instruction should be refocused on multifactorial ("quantitative") traits, inverting the curriculum, and prioritising – perhaps not exclusively – health and disease. Some aspects of genomics could and, some would suggest, should be integrated into health education for all, including consideration of health-related ethical, legal and social implications (ELSIs). These recommendations arise from Dougherty (2009); Dougherty et al. (2011); Hicks et al. (2014); Wefer and Sheppard (2008); Zusevics et al. (2014).

At a large scale, rethinking of key concepts in school biology curricula is advocated by Verhoeff et al. (2009), to reduce cognitive load and generate space for genomics. Current standards, being too vague, do not generate enduring understanding (Mills Shaw et al., 2008). Several authors recommend ways in which the curriculum should be more specific about the content of a genetics/genomics curriculum. Lewis and colleagues (Lewis et al., 2000a, 2000b; Lewis & Wood–Robinson, 2000) recommend that a conceptual framework for understanding genetics and inheritance should identify ideas clearly and explicitly, delineating a basic core of knowledge, upon which future instruction could build. It should connect related ideas and entities that are often separated in the curriculum, such as DNA, genes and chromosomes. Duncan and Reiser (2007) advocate a focus on big ideas and conceptual underpinnings rather than on mechanistic details of ‘central dogma’ processes. Knippels et al. (2005) similarly require that curricular connections should be clear between related themes, such as sexual reproduction and inheritance. More recently, Lewis (2014) has recommended that any genomics
curriculum for scientific literacy should identify not only necessary content but also pedagogies (for reasoning, criticality, application) and assessment criteria.

Knippels et al. (2005) advocate beginning with more concrete observations, ideas, lines of reasoning and contexts (such as humans and other familiar organisms – see also Marbach-Ad and Stavy, 2000). Inversion of the traditional (historically-ordered) presentation of genetics to learners (which focuses first on discontinuous variation and Mendelian inheritance) is widely advocated, as noted previously. There are nuances of the inverted curriculum idea across the set of reviewed papers. One notion, which can be identified from many authors, is to present continuous variation and its multifactorial causes before Mendelian ideas. A second is to teach children aspects of molecular genetics before teaching them about the mechanisms of inheritance (tested in Duncan et al., 2014). These are, of course, not mutually exclusive.

An explanation for the “bootstrapping” effect of learning some molecular genetics before looking at Mendelian theory is partly to be found in the connecting role of proteins. The mechanisms by which genes exert their effects would seem to be a critical conceptual “hinge point” (Duncan et al., 2014), and as Lewis and Kattmann (2004)* note, “when gene and characteristic are seen as equivalent there is little intellectual need to consider how a gene might be transformed into the characteristic”. A repeated recommendation across studies both in and beyond the in-depth review set is that the role of proteins in connecting genetic information to traits should be given greater and earlier emphasis – see for example Duncan and Hmelo-Silver (2009)*; Duncan and Reiser (2007); Roseman et al. (2006)*; Todd and Kenyon (2015); Venville and Donovan (2005). Pavlova and Kreher (2013)* suggest that early establishment of this relationship may avoid students conflating genes with traits and help them to be more open to subsequent learning about the mechanisms of gene expression.

It would be helpful to be more explicit at school level about the diverse set of ideas that can be represented by the gene concept. Ways should be sought by which children can come to understand that ‘gene’ can mean different things in different contexts. Papers echoing this recommendation include Gericke and Hagberg (2010b); dos Santos et al. (2012); Meyer et al. (2013) and Smith and Adkison (2010).

Related to classroom practice, Gericke and Hagberg (2010b) recommend that textbook authors should support process-oriented integration of molecular and Mendelian aspects of genetics, alongside presenting school science as scientific knowledge about nature (rather than as nature itself). Graphic or advanced organisers could be useful to assist teachers in teaching genetics/genomics (Smith and Adkison, 2010) and to give cognitive support to learners (Verhoeff et al., 2009).

Lewis (2014) also highlights the need to provide science teachers with effective professional development to support curriculum innovations, in content and in pedagogy (particularly regarding socio-scientific issues). This echoes the reflection in Mills Shaw et al. (2008) that learner misconceptions may reflect teacher misconceptions. Thörne and Gericke (2014) recommend using specific courses and learning resources (such as textbooks) as key routes to influence teachers so that research recommendations become translated into classroom practice.

Assessment “backwash” is likely also to be a significant influence on teachers’ practice (Black, 1993). Haury and Nehm (2012) advocate the development of rigorous validated instruments for assessing genomics knowledge and understanding, and their use in evaluating the impact of interventions and curriculum developments. There is, in the literature, a series of studies which have pursued this (Tsui & Treagust, 2010; Tsui & Treagust, 2007)*, devising a two-tier diagnostic instrument to evaluate secondary students’ scientific reasoning in the context of a genetics teaching approach which utilises computer-based multimedia learning.

The upper primary age range, argue Smith and Williams (2007), is a sensitive period for the acquisition of ideas about genetics, in which there tends to be divergence between children’s understanding of inheritance as it relates to their own experiences and their grasp of decontextualized scientific concepts. Smith and Williams recommend that genomics educators should seek to capitalise on children’s receptivity at this age. They highlight a “significant research gap”
regarding the ways in which 7 to 12-year old children (in other words, upper primary and lower secondary pupils) understand genetics, and they also note that most studies have been cross-sectional rather than longitudinal. Their implied recommendations for future research are clear.

1.4.2.7 Notable assumptions

Across the in-depth review group, several assumptions are discernible in one or more papers. In identifying and critically considering these assumptions, it is not necessarily the intention to reject them – indeed, many of them seem very reasonable. Rather the intention is to make explicit some of the points on which important arguments and conclusions may rest and to discuss their validity. This section starts with more general points that appear to be widely assumed, before setting out ideas that can be detected in specific papers.

There is a general acceptance that – as put for example by Dougherty et al. (2011) – genetics is “structured round an intellectual core of fundamental concepts”. Reasonable though this seems, it is not clear that this core can be consistently identified or agreed upon.

The ‘dual audience’ argument about science education – Science for All but not all science for all – is widely accepted, where it is commented on at all. However, there is some scepticism (Lewis & Wood-Robinson, 2000) and it is also the case that there is diversity and some lack of clarity across the body of work about the intended audience, in terms of compulsion, universality and age range. There is also little discussion about differentiation in regard to ‘ability’ or prior attainment.

There are some common views in evidence about learners’ interests, including an assumption that science content pertinent to the anticipated future life experiences of learners’ is motivating. In the context of possible ‘genohype’ and media interest in emerging genomics and its applications, assumptions about young people’s ability to discern between science fact and science fiction should be questioned. The influence of out-of-school sources on learners’ beliefs can be strong, as noted by Smith and Williams, (2007) and, in a different contemporary science context (human spaceflight), by Dunlop, Airey, Turkenburg, and Bennett (2019)*. The media, especially television, can be a major source of information for primary school students about DNA, genes and heredity – and this can generate and reinforce misconceptions (Donovan, 2012; Donovan & Venville, 2012, 2014)*. Haury and Nehm (2012) discuss related matters in detail as part of their argument about the way forward in genomics education.

In considering textbooks’ portrayal of gene function, dos Santos et al. (2012) assume that sub-optimal text is associated with pupil (and perhaps teacher) misunderstanding and confusion. It might be expected that this would depend on the centrality of textbooks in pedagogy; there is some acceptance in the reviewed literature that textbooks are important mediators of knowledge in science classrooms, for example in Gericke and Hagberg (2010b). In the UK, for example, it seems reasonable to question whether textbooks are, in fact, so influential, in comparison – for example – to examinations and the specifications from which they are derived. It seems prudent to extend the assumption about textbooks to other learning resources to which pupils and teachers may be exposed. Thörne & Gericke (2014) posit that teachers’ own use of language in classrooms is an important influence on learners.

There is a thread of assumptions about the influence of state educational standards, linking the specified (or intended) curriculum closely to the realised (taught) curriculum. The role of the assessed curriculum, through washback effects on teaching, and dependent itself on state standards, is also a discernible assumption across this body of research. These do not seem to be controversial points. Verhoeff et al. (2009) explicitly state their view, again unlikely to be controversial, that science curricula are overcrowded with content.

The assumption that experts in genomics (researchers and professional organisations, for example) can and should contribute to discussion about the curriculum and to reforming genetics education (Dougherty, 2009; Dougherty et al., 2011) may seem attractive, but it is dependent on achieving clarity of purpose and on access to key policy makers.
With regard to learning outcomes, Mills Shaw et al. (2008) include some critical discussion of assumptions about the relationship between deficits in teaching (by absence or poor quality) and deficits in learning outcomes. There are various assumptions evident about what makes genetics difficult for learners, not all of which are tested empirically in the reviewed papers, including probabilistic reasoning, abstraction, and ideas at multiple levels of organisation (see for example Knippels et al., 2005). These seem reasonable, and are widely discussed in the literature as factors contributing generally to learning challenge in biology (see for example Knippels et al., 2005). However, unlike for example the multiple ontological levels of gene concepts, they are not unique to genetics/genomics.

Zusevics et al. (2014) question the orthodoxy, implicit in other papers, that genomics ideas (specifically health-related) that are appropriate for all young people to learn require in-depth understanding of biology and that they must be taught in science rather than in health education.

1.4.2.8 Prominent strengths and notable weaknesses of the body of work reviewed

Critical reading of each of the 29 studies selected for in-depth review leads to identification of strengths and relative weaknesses in their methodologies and conclusions. An important matter for consideration is whether the body of work as a whole suffers from shortcomings, or whether instead they complement each other (akin to triangulation), permitting confidence in their collective conclusions.

Strengths across this set of papers come from various qualities. Many of the papers are well grounded theoretically, with clear exposition of supporting frameworks. There is generally thorough cross-referencing to other literature. The set includes empirical studies alongside arguments from principle. There is statistically robust quantitative work and qualitative enquiry which gives rich data on, for example, the views of particular stakeholders (Knippels et al., 2005).

Some of the studies have developed and used bespoke instruments, for example diagnostic assessments of learners’ knowledge and understanding, which have good face validity. However, there seems to have been less work published on the detailed validation of these instruments – in some cases (see for example Lewis et al., 2000a) because the idiographic data required in the study are not contingent on such validation.

There is also strength in the intended audiences for the papers, to the extent that this can be inferred. The range of work, from highly theoretical to applied (e.g. intervention design and evaluation), includes knowledge transfer to practitioner audiences (see for example (Hicks et al., 2014b); Smith, 2014).

Just as the strengths noted above do not apply to every paper in the in-depth review set, the relative shortcomings noted below are by no means true of all 29 papers. This is also reflected in the range of ratings under Weight of Evidence that the papers scored in the quantitative phase of this landscape review.

There is a need to consider the potential for authorial bias in the body of work. The papers tend to be written, inevitably, by people who have strong interests in the field, including officers of professional bodies and those seeking to validate particular approaches, such as interventions. There may be scope for factors such as confirmation bias to have affected the work.

As has been raised in other sections, because genomics and genomics education are inconsistently defined (if at all), it is not always clear that the studies are referring to the same ontological entities. This extends to other criteria too – specifically the characteristics of the learners with whom the papers are concerned. The ages and stages of the relevant learners are not always clearly stated, and they are not consistent across the body of work. For example, it is often unclear whether the genomics content under discussion is intended for all learners or for some learners, as per the dual audience argument. Words such as ‘introductory’ are ambiguous and ill-defined. The learning needs of students are often not defined. There is little discussion of differentiation – or, indeed,
recognition of a need for differentiation to meet the needs of all learners based on characteristics such as prior attainment or general cognitive ability. Even where data were collected from children in different ‘ability bands’, in the studies by Lewis and colleagues (2000, 2000a, 2000b), no analysis is presented at that level.

Some of the work under review seems distant from the realities of teachers’ practice, such as the aforementioned need for differentiation. An example of this is in the extensive discussions about the confusion of varied concepts of ‘gene’ to which children are exposed, for example in dos Santos et al. (2012) and in Gericke et al. (2014). There is less consideration of what teachers should do about this in practice. In a spiral curriculum (Bruner, 2009)* where there is an attempt to introduce learners to models that are ‘good enough’ for their current learning needs, teachers are likely to favour a simple model unless it is clear to them why that model is inadequate at that stage. Stern & Kampourakis (2017)*, in discussing teaching for ‘genetics literacy’, suggest a strategy that may be useful here: that teachers should confront the issue by noting two broad categories of gene concept, the referentially definite (i.e. structural, in terms of nucleic acid content) and the indefinite (i.e. functional). However, it is not only with regard to the gene concept that the issue arises of models that are fit for particular purposes.

The 7-12 year-old age group is clearly understudied in this body of work, with one notable exception (Smith and Williams, 2007). Teacher behaviours and beliefs are also relatively understudied here (again with exceptions, such as Knippels et al., 2005; Thörne and Gericke, 2014).

Methodologically, there are some attributes of this body of research which are non-ideal. In design, most of the empirical studies are cross-sectional with few examples of follow-up (an exception being Lewis, 2014, and there are longitudinal elements to Smith and Williams, 2007). As noted previously, well-controlled experimental designs are very rare even amongst the long-listed papers in this review: there are some quasi-experimentally designed empirical studies, typically with weaknesses such as lack of control groups or counterbalancing. Sample sizes are small in some studies (the ‘Lewis papers’ being notable exceptions here – for example Lewis et al. 2000a). On the other hand, the data in the original studies by Lewis and colleagues were collected in 1994–1996 – approaching 25 years old – and similar is true of other empirical studies published early in our target range.

There are some issues with data collection, for example in one study children’s understanding being assessed regarding material they had not yet studied (Marbach-Ad and Stavy, 2000). Extended writing tasks may not be ideal instruments for assessing children’s knowledge and understanding, as noted by Todd and Kenyon (2015), though they are used in many of the studies reviewed. The interpretation of data, particularly qualitative data, is inevitably somewhat subjective, for example in the analysis or assessment of student responses to open questions such as scenario tasks and essays, raising doubts about reliability and validity.

Such methodological issues are not unusual, in our experience, in science education research and they reflect the significant challenges (practical and ethical) of conducting research in schools. Though they indicate a need for caution in interpreting in detail – at the level of individual studies, for example – the major findings and recommendations of the body of work seem reasonably robust, not least because they are well triangulated and oft-repeated.

**1.4.2.9 Further work advocated by the study authors**

There are many examples in the in-depth review set of authors specifically noting further research work that would help to move their fields forward. It is not always clear that the authors are proposing to do this research themselves. In this section, a subset of these suggestions is extracted, focusing on work which aligns with the major findings and recommendations across this body of work.

Todd and Kenyon (2015) note that learning progressions are models describing typical pathways of concept acquisition by learners, and that they remain hypothetical models until empirically validated. They advocate more empirical testing of proposed learning progressions in
Further pedagogical research is advocated to address a variety of questions. Smith and Williams (2007) suggest that research is needed into how learners’ ideas about variation and inheritance can be changed. In a notably constructivist take on this point, Mills Shaw et al. (2008) note that the ‘reconstruction’ of ideas by learners requires logical and critical thinking as well as recognition of the inadequacy of current ideas and awareness of alternative constructs. They propose that research is needed into which pedagogical methods can result in both acceptable content knowledge and deep conceptual understanding.

Duncan and Reiser (2007) identify a need for research into teachers’ instructional choices (for example the selection of learning activities) and how these choices impact on learning. Thörne and Gericke (2014) advocate more studies on verbal communication in science classrooms (and a focus on language in initial teacher education in science).

Smith and Adkinson (2010) recommend research on the extent to which teaching should overtly acknowledge the various meanings of the word ‘gene’. The complexities of the gene concept in relation to genomics education clearly exercise many academics in this field; Smith and Adkinson’s suggestion to focus on what schools should do about this seems reasonable.

1.4.2.10 Other points of note

There are numerous references to the desirability of children learning about the Nature of Science or, less frequently, the history and/or philosophy of science (for example, dos Santos et al., 2002). Learning about genetics could support this, as noted by van Eijck (2010), and vice versa. Gericke and Hagberg (2010b) note that it would be helpful to learners if school science was presented as scientific knowledge about nature rather than as nature itself.

However, Redfield (2012)* – in a position paper related to teaching at undergraduate level, but which draws implications for school-level genetics/genomics education – notes that a historical presentation of genetics (though it has a long-established rationale) does not lead to helpful learning progression in the subject. The canon is, in Redfield’s words, “past its sell-by-date”, with precious learning time “wasted” on Mendel’s Laws and Punnet squares. Redfield expresses the view that genetics/genomics education reinforces the “dominance problem”, which, she argues, is increasingly problematic as our understandings of phenotypic variation deepen. She regards direct-to-consumer genetic testing to be a catalyst for change in a curriculum that has failed to consider what learners really need to know or to prioritise how learners will use their knowledge. Redfield notes, however, that textbook publishers are “very conservative” and though the internet could fill the gap, locating and adapting online resources is an onerous task.

Dougherty et al. (2011), writing under the auspices of the American Society of Human Genetics (ASHG) encourage volunteers from the ASHG’s Genetics Education Outreach Network to get involved in the enhancement and evaluation of school standards for genetics/genomics. They suggest that these volunteers should combine content expertise with knowledgeability about education in schools.

Duncan and colleagues, in their work on learning progressions (for example Duncan et al., 2009), argue that a cognitive model of reasoning in genetics/genomics is needed, which focuses on big ideas, conceptual underpinnings and the connections between different aspects (reproduction and cell division; molecular genetics; inheritance).

Lewis et al. (2000a) note some evidence that young people may focus on social ‘uses’ of DNA, for example in genealogy or in forensics, rather than on biological functions, and they suggest some possible explanations (such as interest and relative familiarity). This may warrant closer attention and consideration in curriculum design. The ‘Nowgen manifesto’ (Finegold & Starling, 2012) cites similar comments made by Lewis and others.

There is recognition of the need to develop high-quality learning activities (e.g. van Eijck, 2010), to apply and test design principles for
teaching interventions (Knippels et al., 2005), and to evaluate curriculum interventions in trials (Dougherty, 2009).

1.4.2.11 Workshop proceedings

In the review group of papers there are two sets of workshop proceedings (Boerwinkel & Waarlo, 2009, 2011). These set out the proceedings of two invitational workshops on genomics education, held in the Netherlands in 2008 and 2010. Although each was relatively small, with about 20 participants, they included several researchers whose work is represented in other papers in the current landscape review. Examples are Knippels, Lewis, van Eijck, Verhoeff, and Yarden, as well as the proceedings editors themselves. There is an unsurprising bias towards European, particularly Dutch, researchers.

The first of the workshops was themed around the curriculum, particularly the extent to which it needed (and indeed to a large extent still needs) to be reconceived for the genomics era. Alongside this there was consideration of associated pedagogy and of how new approaches could be implemented.

In their introduction, Boerwinkel, Verhoeff and Waarlo frame the consideration of curricular change around questions of what, why and how – what has changed with the coming of genomics that is relevant and accessible to young people as learners; why curriculum space for genomics is justified; how genomics education could be provided such that desirable learning outcomes are achieved. In discussing these questions, the authors note the dual-audience arguments about scientific (and genomic) literacy, and they note that a number of different approaches to provision could be possible. For example, context-led and concept-led approaches could be quite different, and may be appropriate in different situations or for different audiences. The assumption that ‘genomics’ should be taught as one (or more) distinct topics in the science curriculum could be challenged by considering the possibility of embedding genomics ideas across several different units of provision – including some beyond the science curriculum. This is reminiscent of Zusevics et al. (2014) highlighting the potential for inclusion of genomics into health education.

The proceedings include a summary list of twelve suggestions from the participants which relate to their collective views about the goals of genomics education and (consequentially) the content of a genomics curriculum (p120–121). Some of the statements perhaps raise more questions than answers (such as “students must be prepared for lifelong learning”). However, in moving forward with genomics education a decade later, it would be worth revisiting these statements to consider their on-going relevance and validity.

Boerwinkel’s summing up of the discussions and conclusions of the first workshop (p122–133) raises many of the issues also highlighted in other studies published around this time. The workshop recognised that education needed to reflect the change in research from a focus on monogenic traits to the interpretation of genome-wide data relating to complex traits. The personal and societal issues and impacts of genomics also warrant change in educational curricula; the workshop also noted that these issues arise not only in the field of healthcare but also in activities such as agriculture. Some of the necessary educational outcomes of curriculum development for genomics education are at the level of those who aspire to careers in scientific research and its applications, including healthcare professionals. An advanced curriculum for such young people should build on or include the core curriculum for all learners.

Most students who have experienced contextualised curriculum innovations and enhancements related to modern genetics and genomics, the authors claim, have found these interesting. However, they point out, these have been young people who have chosen to continue their biology education at post-compulsory levels.

The report advocates further research to define the necessary to be able to engage with social and personal issues relating to genomics, including considering of students’ capacities to develop such knowledge. The development of abilities to cope with information about risk and uncertainty and to participate in moral and ethical discussions
are necessary, they argue, but insufficient: some non-deterministic understanding of nature of the genome is also required, as well as some degree of appreciation for how genomic information is collected and used.

In terms of the pedagogical implementation of a genomics curriculum, the workshop participants argued that new approaches should stimulate “systems thinking”, such as being able to move – conceptually – between different biological levels of organisation; linking concepts within a particular level of organisation; relating abstract models to concrete phenomena. This argument relates to the work of Knippels, Waarlo and colleagues (for example, Knippels et al., 2005). The intellectual challenges associated with these cognitive abilities could be significant.

Boerwinkel’s summing up also notes the overcrowding of the biology curriculum, and that the inclusion of new material on genomics implies potentially difficult decisions about what might be removed to make space. A coherent vision of the curriculum is necessary, he argues, and he suggests both some cooperation with social studies in schools and that learning outside the school (e.g. from the media) could be a useful channel. Little is stated in the proceedings about the gains that might be made by changing emphases and approaches, as opposed to adding additional curriculum load.

The workshop also advocated working with teachers in the development of new curricular approaches for genomics. The final draft of the report claims to provide the first draft of a proposed genomics education curriculum. However, it is cast in very broad terms, with many outcome-based aspirational statements; there is little ‘backwards design’ evident in the proposals, with the effect that instructional design is not prioritised. Nevertheless, the principles outlined could make a useful starting point for any new approach to design and implement a genomics education curriculum.

The second invitational two-day workshop was held approximately two years later, in 2010. The focus, genomics education for decision-making, centred on socio-scientific aspects of genomics education, particularly for active citizenship. The workshop involved a similar number and range of participants, again with substantial involvement of members of the Dutch biology education academic community.

The workshop proceedings include a key-note paper that was used as a stimulus for discussion about SSI-based science education through and about genomics. The paper takes the premise that the important question is not whether genomics education should have an intended outcome of learners being able to make informed decisions, but how this outcome should be achieved. It poses seven questions for discussion, ranging from deciding which genomics issues could help empower students’ decision-making, through identifying the necessary knowledge and the ways in which personal and societal decisions are made, to considering the competencies that teachers would need. Almost a decade later, it could be worthwhile to revisit these questions to determine whether they are still apposite, and if so, whether the wider genomics education community has reached any consensus about the answers to these questions.

There had, in recent years in the Netherlands, been substantial activity in genomics communication and education through sixteen genomics centres and with six mobile DNA laboratories visiting schools (the workshop itself also arose through this programme of activity). The mobile DNA laboratories had been intended to bridge the gap between scientific practice and school science and to link research in genomics, research in education and educational design. An evaluation of the mobile DNA lab provision has been published (van Mil, Boerwinkel, Buizer-Voskamp, Speksnijder, & Waarlo, 2010)*; in the workshop’s stimulus paper, Boerwinkel and Waarlo note that students and teachers had shown appreciation for the activities, but that the achieved learning outcomes (including those related to societal issues) had been disappointing (Boerwinkel & Waarlo, 2010). They caution that positive user evaluations do not necessarily equate with effectiveness – a reminder that is pertinent to the development of educational interventions in general.

The workshop itself, as recorded in the proceedings, explored themes
that led to three key discussions. Firstly, what kinds of decisions related to genomics do we need to prepare students to make? Secondly, what do (and don’t) we know from research about the way genomics education needs to be constructed, to achieve these purposes? Thirdly, what are the implications for instructional design and teacher preparation? The final reflections from the workshop summarise the conclusions, under the headings of why genomics education for decision-making is required; what encompass; and how it should be provided. In these conclusions, there is a marked emphasis on matters related to genetic testing. They note, for example, agricultural and biotechnological implications and applications of genomics research, but take the view that these are less relevant for personal decision-making. This might, in the current geopolitical climate, be regarded as debatable – for example, there may be important implications of genomics technologies for social decision making related to sustainability and food security.

A rationale for genomics education is articulated as helping students both to assess the influence of new technologies on people’s lives and on public morality, and to appreciate the ways in which technological innovation can itself be influenced. The authors note that both healthcare professionals and people being tested need to be able to use and interpret information from genome-wide testing in order to make informed decisions and choices. This claim raises, again, issues of deciding which aspects of genomics education are needed only by learners who will go on related employment, what is needed by all, and what might best be provided at the point of need. Furthermore, if genetic testing, in all its forms, is becoming more and more common, at what point does education for some effectively become education for all? The authors also report a workshop conclusion that citizens should learn about the nature and implications of storage of and access to genetic information.

Conclusions about what should be taught about genomics, for decision-making, the report suggests that examples should be provided of genomic technologies that influence views on morality and the quality of life, and how society and politics influence technology. However, no specific examples are identified. They also recommend that students should learn about the different ways in which they might encounter genetic testing, and the personal and societal choices these might raise. There is a detailed list of thirteen aspects of genomics knowledge which the authors claim is required in order correctly to interpret genomic information. However, there is very little discussion of the accessibility of these (to all or some learners), or of the pre-requisite knowledge and understanding that would be necessary in order to achieve this level of sophistication.

In discussing how to achieve genomics education for decision-making, the authors focus on educational strategies for the consideration of socio-scientific issues. They provide a useful summary of points related to successful pedagogy in this area (p136-137), and a table of necessary teacher expertise (p139). Two key points made are that argumentation skills are open to improvement with reasonable ease, given appropriate pedagogical approaches, and that adversarial approaches (such as debates) and highly value-laden contexts may be less productive than cooperative inquiry strategies and ‘destabilisation’ by presentation with narratives that give differing perspectives on particular issues.

Finally, three recommendations are made for continuing research. Misconceptions about the influence of technologies on personal decisions and vice versa can be blockages to the development of informed attitudes; research is advocated into these misconceptions. Research is needed, they conclude, into the knowledge and understandings that are needed in order to empower students in decision making, for both of the classic audience groups in science education. Thirdly, research is required into pedagogical strategies that could help to avoid or address misconceptions and to empower future decision-makers with the necessary knowledge, understanding and skills.

### 1.4.2.12 Ongoing research

Research in genomics education, of course, continues and there have been several papers published in academic journals since the present landscape review was undertaken. A watch has been kept, using JISC’s Zetoc database and through occasional searching, including citation...
searching, though this has not been undertaken systematically.

The papers which would have scored most highly on the criteria used in our landscape review, had they been published earlier, are noted and briefly considered here. Some of the more recent publications build on previous work and have been cited elsewhere in this landscape review. Examples include work on developing and validating learning progressions for modern genetics (Todd & Romine, 2017; Todd et al., 2017) and critiques of determinism in genetics curricula (Jamieson & Radick, 2017).

Stern and Kampourakis (2017) consider teaching for “genetics literacy” in their detailed and important contribution to the literature, which includes a useful literature review on the nature and origins of students’ misconceptions in genetics. They propose, once again, shifting the focus away from Mendelian genetics, and the teaching of genes as segments of chromosomes, preferring a ‘whole genome first’ approach and emphasising that genomics is science in the making. They prioritise student understanding of genetic testing and they also advocate addressing directly with learners the metaphors that are often used in explaining the functions of DNA and genes, and in doing so, challenging deterministic interpretations. Research on learners’ ideas and on learning progressions is, they note, aligned with their proposals. Stern and Kampourakis also speculate that biological development might prove to be a useful topic through which to introduce these ideas, though it would need testing empirically. They note the central role of teachers and the need to support them with training, professional development and pedagogical resources.

Nichols has investigated teachers’ use of multimodal representations when teaching about molecular genetics, and the impact on learners, concluding that it is open to enhancement through professional development and that greater multimodality is beneficial to learners (Nichols, 2018). The value of animations in explaining dynamic processes is likely to be a contributory factor. Also considering teachers, (Kampourakis, Silveira, & Strasser, 2016) and (Yakisan, 2016) have studied pre-service teachers’ ideas about, respectively, the origin of biological traits and the genetic differences between gametes and somatic cells.

In both cases, sub-optimal knowledge and understanding were revealed, with implications for teacher education and support.
1.5 DISCUSSION AND RECOMMENDATIONS

1.5.1 The landscape of research pertaining to genomics education

The set of academic papers that met the inclusion and exclusion criteria for this review (the ‘long-list’) raises some matters that suggest recommendations for the academic research community and their supporters.

In terms of weight of evidence, there is a roughly normal distribution of scores, which was not inevitable. It is reassuring that there is not a preponderance of papers where the weight of evidence was poorly rated. However, there are also few papers which were assessed to be of the highest weight of evidence. Of the three components of our weight of evidence rating (lack of bias in relation to the study questions; appropriateness of research design and analysis; relevance of the focus of study to this review), the first two are indicators of reliable and internally valid study designs.

Furthermore, approximately one third of the included studies are non-empirical – they do not present and analyse original data, but focus instead on theoretically-derived arguments and points of view. These may be very useful contributions to debate, but they lack the exploratory and hypothesis-testing powers of empirical research. It may be advantageous to seek a different balance between empirical and non-empirical work in the field, and to promote more robust study designs, such as well-controlled experimental methods and – over time – an increasing use of meta-analyses and reviews. This is not necessarily to privilege quantitative over qualitative approaches.

Although the work of researchers in many countries is represented in the long-list, academics from five countries dominate the field. There is evidence of some collaboration on specific projects. However, in the light of low levels of consensus in the in-depth subset of papers about fundamental matters such as what constitutes genomics and genomics education, it could be helpful to promote collaboration and consensus-building within the academic research community in the field.

There is very little attention paid in the surveyed landscape of genomics education research to work with upper primary, lower secondary or middle school learners. For example, in the long-list’s twenty-one studies classified as ‘intervention, design and evaluation’, only one is focused on primary school children and five on middle-schoolers or lower secondary age children. However, there are grounds to suggest that there are important opportunities to teach children in these younger age ranges genomics-related content (Duncan, Freidenreich, Chinn, & Bausch, 2011; Freidenreich, Duncan, & Shea, 2011; Smith & Williams, 2007; Williams, DeBarger, Montgomery, Zhou, & Tate, 2012a; Williams, Montgomery, & Manokore, 2012b). However, as noted by Lewis in Boerwinkel and Waarlo (2009) (p48), little is known about the capacities of learners to understand the relevant science content at lower secondary level – highlighting another important research gap and an imperative to match curricular content to the intellectual development of learners.

Similarly, studies focused on teachers (rather than on pupils) are only about 10% of the surveyed sample. It is overwhelmingly through teachers that curricular content reaches children. It could be helpful to know more about what teachers think and how they act, in order to work more effectively with them in influencing learning.

Problematisation is certainly a necessary element of transformation in education, but it is not sufficient to bring about change. Within the detailed review set, empirical studies focusing on understanding the challenges associated with genomics education (barriers to learning such as misconceptions, poor representation in curricula and resources, and models operating at different ontological levels) outnumber those which relate to the systematic development and validation of new approaches such as learning resources and progressions.
Recommendations relating to research on genomics education are for the research community, including funders, to

1. review the balance between empirical and non-empirical work in the field, to promote more robust study designs, be they qualitative or quantitative, and to promote further research towards solutions to problems;
2. facilitate collaboration and consensus-building in the genomics education research community;
3. address the relative lack of attention that has been given to research with primary and lower secondary age learners and with teachers.

1.5.2 The scope and purposes of genomics education

There is considerable variation in what, precisely, is considered by different authors to be ‘genomics education’ (and, indeed, genomics) and its aims and purposes. The lack of a settled consensus about what genomics is and what genomics education encompasses, including its aims and purposes, is unlikely to be supportive of curriculum development and enactment in the field.

This aligns with the authors’ anecdotal findings, from two informal group interviews with secondary science teachers in the UK. In these discussions, notable confusion emerged, even amongst biology specialists, about what could be said to constitute genomics education. More systematic research into teachers’ opinions, as well as their knowledge and understanding, could be revealing.

In terms of purposes, there is an emerging consensus discernible in the reviewed literature that genomics education needs to equip all young people with knowledge and understanding of contemporary genetic biology to support their active citizenship, focused on human health. This is predicated on general acknowledgement that modern genomic biology and its rapidly developing technological applications pervade society at all levels and have ever-increasing implications for and impacts on citizens, individually and collectively.

There is also implied recognition that a subset of young people requires more than this from their genomics education, to put them in a position to continue to scientific careers, in a broad sense. The lack of consensus in the surveyed research literature is at the level of defining that necessary core material and the extra material required by some; both the circumscription of the field as a whole and the positioning of the boundary between “for all” and “for some” are inconsistent across the reviewed papers. There is a need, also, to understand how much and what knowledge of genomics is necessary to support the engagement of all citizens in personal and democratised societal decision making about genomics. There is academic literature in broad area of knowledge for discussion of socio-scientific issues, to which two authors known for their contributions to genomics education have contributed (Lewis & Leach, 2006). Lewis and Leach (2006) claim that the scientific knowledge required to explain and understand socio-scientific issues is not necessarily extensive, and that concise contextualised examples can often provide it.

Regarding more detailed scope – what is to be included – there are several apparent content domains, not mutually exclusive, including:

- multi-factorial variation and inheritance
- applications of genomics research and their ethical, legal and social implications
- structure and function of genomes
- structure and function of components of genomes (for example DNA and genes)
- techniques for studying genomes and their components, including sequencing and bioinformatics.

The first two aspects above are, in the reviewed work, central to the most securely articulated purposes for young citizens’ learning about genomics. It is not clear how much consideration of the other
three content domains is necessary or sufficient to support learners’ understanding of the first two. It is also not clear from the reviewed work what other learning (for example critical analysis of science in the media) may also be required to support young people’s genomics learning and their abilities to act on it. Other related topics are also considered in some of the reviewed work, and could potentially also be regarded as part of genomics education:

- related biological ideas, including proteomics and systems biology.

This set – and detailed work on genomic structures, functions and analyses – is likely to be encountered only by learners who may choose or have chosen to study biology beyond compulsory curricula.

In Figure 2.5 in this report (page 64), we offer a model for classifying the broad types of material that together could constitute the substantive content of genomics education.

**Recommendations relating to the nature and purpose of genomics education are for stakeholders to**

1. make efforts to achieve greater clarity about the purposes of genomics education (individually and collectively), aiming if possible to confirm the positions which are emerging; to add detail to establish the necessary and sufficient knowledge, understanding and skills that are required to meet participatory citizenship goals; and to agree what specific additional content is needed for the sub-set of young citizens who will go on to become scientists;

2. as part of the above, seek to establish and articulate a shared understanding amongst stakeholders of what genomics is taken to encompass, in relation to different groups of school age learners and in terms of content and perspective.

1.5.3 Supporting curriculum development for genomics education

Building on analysis of the reviewed research papers’ contribution to debate about the scope and purpose of genomics education, some more specific points can be synthesised from this body of work concerning related curriculum and pedagogy. Curriculum can be understood as what is required or intended to be taught, what is taught and what is assessed. Pedagogy refers to how the material is taught. Two key arguments can be drawn from the surveyed literature. Firstly, that there are some traditional practices in genetics education which are sub-optimal for genomics education. Secondly, that there is a challenge in the contemporary and rapidly changing nature of genomics.

Duncan, Castro-Faix and Choi (2014) have presented evidence that ‘inverting’ the curriculum to teach children about molecular genetics before (rather than after) introducing Mendelian inheritance is advantageous. Traditionally, many children would have learned about Mendelian (single gene) inheritance before finding out about the functions of genes and the mechanisms of their relationships to phenotypic traits, if indeed they learned about the latter at all. Another case of outdated educational practice widely criticised in the reviewed literature (see for example Gericke et al., 2014) is the potentially confusing use of the gene concept to mean different things. Molecular genetics can help to reconcile these different concepts, for example helping to explain how dominance relationships between gene variants can occur, emphasising the roles of proteins. The conceptual value of understanding how proteins link genes to traits is a recurrent idea in these papers, and suggest that genomics education might cast a spotlight on the proteome as well as focusing on the genome.

Learning about Mendelian inheritance is likely, for many students, to follow on from observation of differences between individuals that emphasised discontinuous variation; it is this discontinuous variation that a study of Mendel’s ideas is intended to explain. This contrasts with the emerging goals of genomics education, which prioritise the understanding of continuous variation and its causes. As explained by Moore (2008), convincing students to give up simple Mendelian
determinism in favour of more complex multifactorial ideas is unlikely
to be easy. Starting with the complex realities of variation and explicitly
treating Mendelian inheritance as an incomplete part of an explanatory
model may be helpful.

Genomics technologies – for studying the genome or for applying our
understanding of it (in healthcare, for example) – continue to develop
rapidly and so are also prone to outdated representation in school curricula. For example, many learning resources on genome sequencing
focus on Sanger sequencing, rather than on more modern methods. Such anachronisms are not only misrepresentations but also risk
confusing learners if and when they encounter more contemporary
material later.

Rapid developments in genomics pose a challenge to the education
system if it is to present learners with material that is contemporary
and relevant to their current and anticipated lived experiences. The fast
pace of change in genomics contrasts with what may seem to be the
slow pace of curriculum reform; Lewis (2014) bemoans the slow trickle-
down of new science into school curricula. The many influences on this
are beyond the scope of the present review, but the implications are
pertinent.

Those who drive and support curricular change in science education
will need to make effective use of the mechanisms available to them.
In educational systems involving high-stakes assessments, such as
the UK, exam specifications can offer useful leverage, as a result of a
strong ‘backwash’ effect of assessment on what is taught. Genomics
now features in the statutory national curriculum for 14-16 year-olds
in England and Wales. However, as noted by Mills Shaw et al. (2008)
and demonstrated by, for example, Lewis (2014), inclusion in curricular
standards is unlikely to be sufficient to secure desired learning outcomes.
In striving to define necessary and sufficient content for school curricula,
some reflection on three points is likely to be helpful:

a) Can a set of content be identified for a core curriculum that is likely
to be reasonably stable over the lifetime of the curriculum? This might
include a requirement for unspecified exemplification which could be
used by stakeholders as an opportunity to present at least some
learners with very contemporary material.
b) How secure are the distinctions between what all young citizens
should learn at school (underpinning knowledge and skills, for
example), what is best provided to citizens at their point of need,
and what is needed only by a subset of young people for their further
education and employment? What knowledge is necessary to support
personal and societal decision-making?
c) What is necessary and sufficient additional material for the subset of
young citizens who may go on to scientific careers, and what makes
this material distinct (for example, does it focus more on mechanisms
than impacts, or on technologies for studying genomics)?

Recommendations relating to curriculum development are for
stakeholders to

1. delineate the knowledge, understanding and skills that are
necessary and sufficient to meet participatory citizenship
goals for all, to fulfil the requirements of those young citizens
who will go on to become scientists, and to allow citizens to
engage with further information at the point of need;
2. consider the implications of different paces of change in
genomics and in education for a core curriculum requirement
that is reasonably stable over time;
3. use the channels open to them, including those which
could influence specified and assessed curricula, to promote
educational adoption of evidence-informed learning
progressions, appropriate terminology and contemporary
examples of genomics technologies.
1.5.4 Supporting pre- and pro-genomics education – early foundations

In addition to the content domains that seem to delineate the scope of genomics education, an underlying group of ideas is considered in some of the papers reviewed. These could be considered ‘pre-genomic’ ideas, for example notions of variation and inheritance that might be encountered at an early age and which may influence children’s later learning of more overtly genomic concepts. Children acquire naïve theories of kinship and inheritance when they are pre-schoolers (Solomon, 2002; Springer, 1996).

Ideally, early, pre-genomic learning in schools would be pro-genomic. If appropriately conceived and well taught, these ideas could potentially support later learning by helping children to reconstruct naïve conceptions and to construct instead more helpful understandings. For example, it could be supportive for children to come to understand that practically every observable feature of humans (and indeed other living things) varies continuously to some extent – the basis of a ‘Weldonian’ idea (Jamieson & Radick, 2017) that could steer learners away from genetic determinism. Jamieson and Radick (2013) suggest that ‘Weldon’s dissent from Mendelism could well serve to inspire those attempting now to cast Mendelian tradition aside in order to reshape genetics teaching for a genomic age’.

Weldon’s dissent from Mendelism could well serve to inspire those attempting now to cast Mendelian tradition aside in order to reshape genetics teaching for a genomic age.’

(Jamieson & Radick, 2013)

However, it seems likely that in practice, much pre-genomic learning – in or out of school – is not so supportive of future learning. The work of Smith and Williams (2007) relates directly to this aspect. Although this is the only study in the in-depth review group that has a principal focus on primary-age learners, it is part of a significant body of work on the ideas of children up to age 11 about variation and inheritance. Collectively, these studies are of considerable interest in relation to pre- and pro-genomics education. Set in a broader context of work on children’s ideas in science (Allen, 2014; Driver, 1985; Driver, Rushworth, Squires, & Wood-Robinson, 2005), numerous common misunderstandings about variation and inheritance have been identified amongst primary and middle school children (Cisterna, Williams, & Merritt, 2013; Smith & Williams, 2007; Williams, 2012; Williams et al., 2012a; Williams et al., 2012b). These include - but are not limited to - uni-parental inheritance, cis-parental inheritance, inheritance of acquired characteristics, misattribution to “nature” or “nurture”, and difficulties with probabilistic reasoning.

Pro-genomic language should be encouraged amongst teachers, for example distinguishing carefully in their use of terms (such as ‘DNA’, ‘gene’, and ‘genome’) and avoiding deterministic implications that an organism’s characteristics are influenced solely by its genome. This is not to suggest that teacher talk with younger learners should be over-complicated or laboured, but to acknowledge the risk of reinforcing naïve ideas if better models are poorly expressed. The literature includes studies which have suggested introducing explanations of heredity to young children, focused on kinship and based on a very simplified notion of genetic material (Ergazaki, Alexaki, Papadopoulou, & Kalpakiori, 2014; Ergazaki, Valanidou, Kasimati, & Kalantzi, 2015; Solomon & Johnson, 2000). This notion can serve as a conceptual ‘peg’ or ‘placeholder’ on which children can hang a rudimentary scientific explanation – a ‘precursor’ model that is fit for purpose at that age and will support the later construction of more detailed concepts of what is inherited and how.
Recommendations relating to pre- and pro-genomics education are for researchers, developers and teacher educators to

1. review the research evidence base regarding naïve concepts about genetics and inheritance, with the aim of assessing their impact on genomics learning and thereby considering actions that need to be taken;
2. consider how to ensure that pre-genomics learning in primary and lower secondary schools is pro-genomic, through curriculum and resource development and through teacher professional development for the enhancement of pedagogical content knowledge.

1.5.5 Supporting pedagogy for genomics and pre-genomics education

Pedagogically, genomics education could be considered as a perspective from which biological ideas are presented to learners within a pervasive genome paradigm, from the earliest explorations with learners of variation and inheritance. This would represent a paradigm shift for many teachers. If some teachers are reluctant to adopt this approach, or do so sub-optimally, there may be several contributory factors, including some which are rooted in teacher characteristics, for example:

- insecure or outdated (traditional) subject knowledge and pedagogical content knowledge regarding topics of variation and inheritance
- insecure pedagogical content knowledge and/or skills regarding effective practice in education for personal and civic engagement with socio-scientific issues
- lack of recognition of a need for change
- lack of awareness about high-quality supporting resources.

Within the surveyed literature, several of these points are highlighted. They are potential targets for intervention to support genomics education. In 1999, Munn and colleagues noted that, in their experience, “genome education programs require the collaborative efforts of science teachers, genome researchers, ethicists, genetic counsellors, and business partners”. (Munn, Skinner, Conn, Horsma, & Gregory, 1999). This underscores the need for partnerships between the genome science community, particularly public engagement specialists, and the education community (teachers, and others, for example examiners and resource producers) to secure high-quality classroom practice in genomics education.

Teachers’ subject knowledge and pedagogical content knowledge are potentially open to adjustment through professional development and training (Van Driel & Berry, 2012). This can take many forms, of variable reach and impact. Engagement with high-quality teaching resources has the potential for positive impact on many teachers’ own knowledge and practice. For example, high-quality resources could influence the way teachers talk about molecular genetics ideas. They could help teachers to develop and use ‘scripts’ that, for example, bridge more carefully the ontological levels between genes as units of inheritance and observable phenotypic traits – addressing concerns expressed by Thörne and Gericke (2014).

There are numerous examples in the research literature of small-scale interventions in genetics/genomics pedagogy (small scale in terms of curriculum coverage and/or the number of students involved), typically with some degree of evaluation (often post-hoc). Knippels, Waarlo, and Boersma (2005b) have proposed design criteria for teaching and learning activities in genetics. Only a smaller number of interventions have reached stages of development where they have been subjected to more robust evaluation. Examples include those reported by Duncan and colleagues (Duncan et al., 2014; Duncan et al., 2009; Duncan & Tseng, 2011) and, for younger children, Ergazaki et al. (2015).

Robust evaluation of the impacts of pedagogical interventions requires not only good study designs but also valid approaches to assessing key learning outcomes. Here too, there are some models reported in the literature of assessment tools that are at least partially validated including those from Lewis and her collaborators, for example in Lewis et al.
(2000a), from Tsui and Treagust (2010), and from Todd et al. (2017).

The University of York Science Education Group’s BEST project (UYSEG, 2018) is providing teachers with succinct summaries of relevant published research alongside evidence-informed items for administering and responding to diagnostic and formative assessment. The aim of this project is to promote the adoption of evidence-informed pedagogical practice; amongst the “key concepts” addressed are heredity and genetic information, and the structure and function of the genome. Section 2 of the current report addresses the provision of resources to support genomics education, including the quality of textbooks directed towards English curricula, and the signposting of these resources towards teachers.

Although it is disappointing that textbooks are not serving genomics education well, as concluded by, for example, Gericke et al. (2014) and Hicks et al. (2014b), it seems important to note that – as is the case with teachers – textbook authors and publishers may be substantially influenced by the curriculum and its assessment. Such appears to be the situation in the UK, for example. The findings on textbooks are perhaps best regarded as a barometer indicating the pressure on educators to cover the examined curriculum efficiently. This also highlights the dependence of pedagogical reform on curriculum reform.

Stereotypically, as with curriculum change, pedagogical change is slow and difficult to effect. However, the burden of educational change is felt keenly by schoolteachers (Hargreaves, 2005), which elevates the importance of understanding teachers’ opinions and needs and of working in partnership with teachers to effect change.

Recommendations are for those who develop and support school-level genomics education to

1. engage with the evidence base regarding effective practice in science education for personal and civic engagement with socio-scientific issues;
2. facilitate research to
   • establish the range of ideas that schoolteachers associate with ‘genomics’ and ‘genomics education’ and compare them with those of specialist stakeholders in the field
   • explore the factors influencing teachers’ approaches to teaching genomics, including subject specialism, time elapsed since training and engagement with professional development activities
   • strengthen the evidence base regarding effective pedagogical practice for genomics education, including the development of validated assessment tools;
3. facilitate access to teaching resources that are evidence informed, congruent with curriculum and assessment models, and that meet teachers’ perceived needs;
4. facilitate access to professional development that supports teacher acquisition and application of subject knowledge and pedagogical content knowledge for genomics education, including approaches to teaching about socio-scientific issues.
1.6 REFERENCES


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SECTION 2: RESOURCES TO SUPPORT TEACHING AND LEARNING
2.1 BACKGROUND

Notwithstanding previous discussion of curricula for genomics education (see Section 1.5), we could regard as a desirable outcome of school science education that all school leavers should have some basic understanding of what a genome is and of some of the impacts of genome-related science and technology. The advantages of this would be two-fold:

1. It would support school leavers to engage with socio-scientific issues (SSIs) pertaining to genomics as informed citizens.
2. It would enable students to decide whether genomics is an area they wish to pursue in further study or employment, and equip those who choose to do so with knowledge and understanding that is sufficient to be a foundation for progression.

What is taught is defined broadly by the curriculum; Schmidt and Prawat (2006), Oates (2010) and others have noted dependencies on a number of mediating instruments, which in England include:

- policy instruments, namely the science programmes of study for ages 5–16 which are set out in the National Curriculum issued by the Department for Education;
- at ages 13–18 the GCSE and AS/A level specifications and associated high-stakes assessments issued by the Awarding Organisations;
- textbooks and other teaching and learning resources.

This section of the report surveys the landscape of instruments that serve both to drive and to support teaching of ideas related to genomics in school science. Its scope is limited to the school science curriculum in England, but is not limited to teaching resources. We consider how the content of the current National Curriculum and GCSE and AS/A level specifications will drive teaching of genomics-related ideas in schools in England, and what exists in terms of textbooks and other resources to support this teaching.

2.1.1 Overview of the school science curriculum in England

Maintained schools in England must follow the statutory National Curriculum issued by the Department for Education, which comprises programmes of study for each subject in four key stages (Department for Education, 2013a, 2013b, 2014b). Science is compulsory for all learners up to the end of Key Stage 4 (KS4). The National Curriculum defines a minimum entitlement.

For most students, the end of KS4 marks the end of their formal science education. Academies (including free schools) and independent schools are not required to follow the National Curriculum, though we presume that many of them still use it as the basis for what is taught in preparation for public examinations.

KS4 of the National Curriculum (typically studied by learners aged 13–16) leads to qualifications at Levels 1 and 2 of the National Qualifications Framework, including GCSE qualifications. GCSE science students follow either a ‘combined science’ route (leading to two GCSEs in science) or a ‘separate science’ route (leading to GCSEs in biology, chemistry and physics). Schools in England teaching GCSE science select a specification from one of the Awarding Organisations (typically the ‘big three’: AQA, Edexcel and OCR). The content of these specifications is determined by subject content criteria issued by the Department for Education, which cover the KS4 programme of study for science.

Post-KS4, learners may choose to continue into further education in science leading to qualifications at Level 3, including AS and A level qualifications (typically studied at age 16–18). Schools choose an AS/A level specification from one of the Awarding Organisations. Approximately 60% of the content of each A level specification is determined by subject content criteria issued by the Department for Education, and is therefore common between the specifications (Department for Education, 2014a); the remaining content of each specification is determined by the Awarding Organisation and there are differences between specifications.
2.2 SURVEY METHODS

The survey of teaching and learning resources encompassed three strands of work:

1. surveying genomics-related content in the current National Curriculum in England (covering ages 5–16) and in the AS/A level specifications (16–18);
2. surveying genomics-related content in biology textbooks associated with the current GCSE and AS/A level biology courses in England;
3. compiling a catalogue of free resources that could be used in teaching and learning about genomics in schools.

2.2.1 Survey of genomics-related content in the school biology curriculum in England

The current science National Curriculum programmes of study for ages 5–14, forming Key Stages 1 to 3, were downloaded from the Department for Education (2013a, 2013b).

The subject content criteria for GCSE Biology and GCSE Combined Science, covering Key Stage 4 of the National Curriculum in England and covering ages 14–16, were also downloaded from the Department for Education (2015a, 2015b). The science programme of study document for Key Stage 4 (Department for Education, 2014b) covers the same content as the GCSE science subject content criteria but is expressed in less detail, and was therefore omitted from the survey. The GCSE Biology specifications were downloaded from the Awarding Organisations (AQA, 2016; Edexcel, 2016a; OCR, 2016a, 2016b).

The subject content criteria for AS/A level Biology, covering ages 16–18, were downloaded from the Department for Education (2014a). The AS/A level Biology specifications were downloaded from the Awarding Organisations (AQA, 2017; Edexcel, 2016b, 2016c; OCR, 2014a, 2014b).

2.2.2 Survey of genomics-related content in GCSE and AS/A level biology textbooks

The textbooks included in the survey were books endorsed by the Awarding Organisations for use with their GCSE and AS/A level courses (see Appendix A5). These books are not free resources; they are purchasable, either by the school (typically for class sets) or by parents or students (for personal copies).

Although other textbooks are available to support teaching and learning of GCSE and AS/A level biology, a recent survey of the use of textbooks in schools in a number of countries noted that the use of Awarding Organisation-endorsed textbooks is compelling for schools in England (Oates, 2014). This is ascribed to teachers’ desire for their students to do well in the high-stakes GCSE and AS/A level examinations administered by the Awarding Organisations, as they are linked to national performance measures for schools.

A set of five questions was used as the framework for analysis of each textbook, as follows:

Does the book...

1. ...explain the difference between genes and the genome (for example, by explaining the presence and functions of non-coding DNA)?
2. ...make clear that most phenotypic features are affected by multiple regions of the genome, not just single genes?
3. ...make consistent use of modern terminology such as variant?
4. ...discuss genome sequencing, or the field of genomics?
5. ...discuss the social, technological and ethical impacts of genomics?

Taken together, the five questions attempt to probe the issue of whether the textbooks go beyond traditional genetics teaching in schools.
2.2.3 Survey of free resources that could be used in teaching and learning about genomics

2.2.3.1 Strategy for locating free resources

Teachers in general seem enthusiastic and keen to make use of high-quality resources in their teaching, but are also short of the time needed to comprehensively search for such resources. We take the view that if a resource is to be useful to teachers in practice, a primary characteristic of that resource is that it must be easily found. Therefore, the search strategy that was adopted in this survey deliberately emulated the type of online search strategy a teacher in England might use when searching for teaching and learning resources related to genomics.

Genetics and genomics-related resources were located in two stages of online searching:

Stage 1: a trawl of websites that teachers commonly visit for biology teaching resources, namely:

- the websites of the Awarding Organisations
- the National STEM Centre e-library (www.stem.org.uk/resources)
- the Practical Biology website from the Nuffield Foundation (www.nuffieldfoundation.org/practical-biology)
- the National Centre for Biotechnology Education (NCBE) website from the University of Reading (www.ncbe.reading.ac.uk)
- the Science and Plants for Schools (SAPS) website (www.saps.org.uk).

Stage 2: a trawl of three websites developed in England specifically to support genomics education:

- the Your Genome website from the Wellcome Genome Campus Public Engagement team (www.yourgenome.org)
- the Genomics for Schools website from Nowgen (www.genomicsforschools.org)
- the Virtual Genetics Education Centre website from the University of Leicester (www2.le.ac.uk/departments/genetics/vgec)

The TES Resources website (www.tes.com/teaching-resources) was not included in the search strategy. This website hosts a very large bank of resources created by teachers, rather than by dedicated resource providers or science specialists. Although teachers may use this website when looking for genetics and genomics-related resources, the quality of the resources is highly variable and there are issues of provenance (the uploader of a resource is not necessarily the owner or author of all the material included in it); for these reasons, it was omitted from the search.

The searches were conducted between March and September 2016, and were updated in March 2017.

2.2.3.2 Cataloguing free resources

Resources found by the searches were added to a resources catalogue if they met the following inclusion criteria:

- the resource is written in English
- the resource can be accessed, downloaded and used in schools without any cost to the school, teacher or student (excluding printing and photocopying costs incurred at the point of use)
- the resource was published or last updated in the year 2000 or later
- the resource is suitable for use with students between the ages of 5 and 18.

The following attributes were recorded for each resource added to the catalogue:

- resource title
- URL
- year of publication or of last update
- author
- author type (teacher/science specialist/dedicated resource provider)
- target age
presentation / role-play or debate / scheme of work / video)
• classification according to the Biological Sciences Curriculum Study (BSCS) 5Es instructional model (Bybee et al., 2006) – see note 1
• classification of practical activities according to Millar’s description of the three possible purposes of a practical activity (Millar & Abrahams, 2009) – see note 2.

In addition, the following attributes were recorded:

• whether the resource facilitates the development of
  – data analysis/numeracy skills
  – understanding about the nature of science (NoS) – i.e. the processes of scientific enquiry, how scientific explanations are developed, and the role of the scientific community
  – understanding about ideas concerning risk, ethics or decision making
• whether the resource goes ‘beyond genetics’ to introduce modern ideas about the genome and genomics, going beyond traditional school topics about inheritance and the structure and function of DNA
• notes briefly summarising the resource and any points about which teachers should be aware.

Note 1: The BSCS 5Es instructional model was developed by Bybee et al. (2006) to describe five phases of teaching and learning that facilitate conceptual change. The five phases have been described in detail by Bybee et al. elsewhere, but for the purpose of this survey they were applied to resources in the catalogue as follows:

• engagement – resources that could be used to engage learners at the start of an episode of teaching related to genomics
• exploration – resources that enable learners to develop their own explanations, usually through enquiry
• explanation – instructional resources that transmit information directly to learners, and that facilitate teacher-led development of explanations
• elaboration – resources that challenge learners to apply key ideas in new contexts, and that provide extension by introducing ideas that are beyond the scope of the curriculum
• evaluation – resources that enable teachers (and learners themselves) to evaluate what the learners have learnt, by providing evidence of students’ knowledge and understanding (usually through a series of questions related to the topic); these resources can be used as formative assessments.

Note 2: Millar and Abrahams (2009) proposed a system for analysing practical activities in order to determine which of three purposes they fulfil. Thus, practical resources were classified in the catalogue according to which of the three purposes they appeared to satisfy, from:

Purpose 1: developing learners’ scientific knowledge and understanding
Purpose 2: developing learners’ ability to use scientific equipment or to follow a standard procedure
Purpose 3: developing learners’ understanding of the scientific approach to enquiry.
2.3 RESULTS AND ANALYSIS

2.3.1 Results of the survey of genomics-related content in the school science curriculum in England

Genetics teaching in schools has traditionally focused on the molecular structure of genetic material, how information is transferred from gene to protein, how genes affect our features in combination with the environment, and models of Mendelian inheritance. More recently, ideas about applications of gene technology in healthcare and agriculture have been included, such as genetic engineering and the testing for alleles associated with disease. In the real world, the importance of studying whole genomes is now clear, and genomics is at the forefront of modern biology. Genome sequencing involves new technologies, bioinformatics and ‘big data’, and the evidence it provides is becoming increasingly important in fields across biological science.

Ideally, to better prepare school children for citizenship and for science careers, school curriculum developers, resource writers and teachers should reflect real-world advances. This implies widening the focus beyond the functions and inheritance of single genes (‘genetics’), to a more sophisticated consideration of genomes and the processes and applications of genome sequencing (‘genomics’).

Some steps have already been taken in this direction. For example, ideas about genomes were added to the statutory national curriculum for 14-16-year olds in England in 2014. This resulted from a deliberate attempt to update the genetics-related content of the curriculum by those tasked with rewriting the biology content for 14-16 (personal communication, 2016). However, there does not appear to have been a coordinated effort to update the biology content in this way across the entire 5-18 age range; although all the key stages have been reformed since 2013, the stages were redeveloped more-or-less concurrently with different working groups responsible for the different stages (rather than sequentially and/or by the same group, both of which may have been more conducive to a coordinated approach).

An overview of the ideas related to the genome and genomics in the current school science curriculum in England follows.

KS1-2 (primary school; ages 5-11):
There are no references to the structure and function of the genome in Key Stages 1 and 2 of the National Curriculum in England. However, a number of ideas are introduced that we could consider to be ‘pre-genomics learning’, because they prepare students to encounter genome-related ideas later; they could be regarded as the first learning outcomes in one or more learning progressions concerned with genomics. These ideas relate to reproduction, variation, classification and evolution, and are presented in Table 1 in Appendix A4.

KS3 (lower secondary school; ages 11-14):
At Key Stage 3, students are introduced to ideas about heredity and a simple model of chromosomes, genes and DNA; these ideas can be grouped broadly as relating to the structure and function of the genome. These are supported by some pre-genomics, and perhaps ‘pro-genomics’: ideas related to cell structure, reproduction, variation and evolution, which – if appropriately taught – could facilitate subsequent genomics learning. In addition, this stage of the curriculum introduces an application of genome-related science that has real-world impacts: the use of gene banks to preserve biodiversity at the genetic level. The relevant ideas from the National Curriculum are presented in Table 2 in Appendix A4.

KS4 (GCSE: upper secondary school; ages 14-16):
Pre-reform GCSE Biology and combined science courses (which began teaching in September 2012) continued to be taught until summer 2017. These specifications were based on subject content criteria (Ofqual, 2011) that required students to understand:

- variation within species including the effects of genotype and environment
- how genes determine the structure and function of organisms
- the structure and function of DNA and its role in protein synthesis.
Reformed GCSE Biology and Combined Science specifications feature genomics-related ideas more prominently and in more detail than the previous courses. First teaching of these reformed GCSE Biology and GCSE Combined Science courses officially began in September 2016. However, many schools started teaching the new specifications to their Year 9 (13- and 14-year-old) students during the 2015-2016 academic year, reflecting an apparent trend towards schools teaching GCSE science as a 2.5 or 3-year course starting in Year 9, rather than as a 2-year course starting in Year 10.

The learning outcomes related to genomics at this stage of the curriculum are presented in Table 3 in Appendix A4. As already noted, the recent round of reform of the KS4 (14-16) biology curriculum attempted to update the ideas being conveyed about genes and genomes, as well as the terminology used. Students are required to learn about the structure and function of the genome in some detail; the term ‘genome’ and the idea of the genome as “the entire genetic material of an organism” are introduced for the first time. The curriculum includes ideas related to the structure and function of the genome, specifically the molecular structure of genetic material, the genetic code, protein synthesis, mutations, how the interaction of genome and environment determines phenotype, and the principles of single gene inheritance.

KS4 students should now learn about genetic variants, including that they arise from mutations and the effects of variants in coding and non-coding DNA. However, in this particular reform, an inconsistency has arisen. The criteria require students to be able to “explain the terms... allele/variant”, leaving it up to specification writers, examiners, textbook authors and teachers to decide whether allele and variant are synonyms or discrete terms, and how, precisely, to define them. All of the GCSE Biology specifications from the Awarding Organisations AQA, Edexcel and OCR use the term ‘variant’ in learning outcomes copied from the subject content criteria (AQA, 2016; Edexcel, 2016a; OCR, 2016a, 2016b) but only one specification – OCR GCSE Biology B (Twenty First Century Science) – attempts to explain the terms allele and variant in a ‘Teaching and learning narrative’ that accompanies the learning outcomes, as follows: “The two chromosomes in a pair each carry the same genes. The two versions of each gene in the pair are called alleles, and [they] can be the same or different. A different version of a gene is a genetic variant”. Hence, there is likely to be variation in how the terms are explained and used in classrooms; in practice, the term ‘variant’ may be mostly omitted in favour of the more familiar (to teachers) ‘allele’.

In another sign of modernisation students are explicitly required to know that “most phenotypic features are the result of multiple genes rather than single gene inheritance”, but are still required to rehearse and predict the outcomes of single gene crosses (in part as a vehicle for the development and assessment of mathematics skills within biology, including ratios and probabilities).

In addition to details of the structure and function of the genome, this stage of the curriculum, as with earlier key stages, includes ideas that could be considered pre- or pro-genomics learning. These relate to the function of the nucleus, mitosis and meiosis, and ideas about cell differentiation. Students are also required to consider social, technological and ethical impacts of genome-related technology, in the contexts of medicine and agriculture.

Finally, unlike at earlier key stages, students are also required to learn about how our understanding of genetics and the genome has developed. At this stage, the curriculum only explicitly requires students to be familiar with the early work of Mendel and does not specify other developments to be studied. The Edexcel specification requires students to “discuss the outcomes of the Human Genome Project”. The OCR specification for Gateway Science Biology A offers no further guidance, whereas the OCR Twenty First Century Science Biology B specification provides guidance in its teaching and learning narrative, stating “today, scientists sequence whole genomes to investigate how genetic variants influence an organism’s characteristics”. The AQA specification goes furthest, including the late 19th century observation of the behaviour of chromosomes during cell division, the early 20th century idea that Mendel’s ‘units of inheritance’ are genes located on chromosomes, and the mid-20th century elucidations of the structure of DNA and gene function; it stops short of genome sequencing, but does mention
elsewhere in teacher guidance that “the whole genome has now been studied”.

AS and A Level (16–18):
Teaching of the current AS and A level Biology specifications began in September 2015. The specifications are based on subject content criteria issued by the Department for Education (2014a). The learning outcomes related to genomics at this stage of the curriculum are presented in Table 4 in Appendix A4.

It is worth noting one particular implication of the reform timetable. Students commencing the reformed AS and A Level Biology courses in September 2015, 2016 and 2017 will have studied the pre-reform GCSE Biology courses, and will therefore not have benefited from the more prominent consideration of genomics-related ideas in the reformed GCSE courses. The first cohort of students to have studied the reformed GCSE courses will start AS/A Level courses in September 2018, complete them in summer 2020, and enter Higher Education in autumn 2020. The AS/A level criteria require students to explore some ideas that are not featured explicitly in earlier stages of the curriculum, including regulation of the genome, ideas about genome sequencing projects, and alteration of gene function for research. Otherwise, many of the ideas at this level appear, prima facie, to have been covered at earlier stages of the curriculum, for example most of the content that could be considered pre- and pro-genomics, the idea that nucleic acids have important structure-function relationships, and the idea that the sequence of bases in DNA determines the structure of proteins. The AS/A level criteria are expressed in much less details than the GCSE criteria, and define only 60% of the content of each specification; therefore, specification writers have leeway to add breadth and depth to the required learning to help ensure progression in learning from GCSE level.

This leeway has led to some variation between the specifications. All of the specifications (AQA, 2017; Edexcel, 2016b, 2016c; OCR, 2014a, 2014b) add to GCSE-level understanding of the structure and function of the genome with ideas about semi-conservative replication. They also expand understanding of the link between DNA and proteins with ideas about how gene expression and protein synthesis are regulated: all of the specifications except OCR B consider transcription factors; AQA includes regulatory genes; AQA, Edexcel A (Salters-Nuffield) and OCR A include epistasis; all of the specifications consider post-transcriptional changes to mRNA such as splicing, while Edexcel A (Salters-Nuffield), Edexcel B and OCR B explicitly include the idea that this means one gene can give rise to more than one polypeptide.

The existence of non-coding DNA is explicitly acknowledged by AQA; AQA and the two OCR specifications mention introns, and Edexcel B states that “not all the genome codes for proteins”. However, in the AQA specification the genome is defined as “the complete set of genes in a cell” (our emphasis, seemingly overlooking non-coding DNA). The other specifications use the term genome without defining it. The term ‘variant’ is not used in the AQA and Edexcel specifications, but ‘allele’ is used frequently; AQA holds that “random mutations can result in new alleles of a gene”; the term variant appears fleetingly in the OCR specifications, wherein “gene variants (alleles)” is used in the context of genetic biodiversity, and “allele (gene variant)” in the content of inheritance. Considering inheritance, in all of the specifications, students are required to understand monohybrid and dihybrid crosses, and all except OCR A introduce ideas about epigenetics. Evolution is considered consistently across the specifications as the natural selection of alleles (rather than variants, as in the GCSE criteria).

2.3.2 Results of the survey of GCSE and AS/A level biology textbooks

GCSE biology textbooks:
Notes from the analysis of the endorsed GCSE Biology textbooks are presented in Table 1 in Appendix A5.

The content of the endorsed GCSE textbooks relates very closely to the specifications they are written to support; indeed, close coverage of the specification is one of the criteria for the granting of an endorsement by the Awarding Organisation. The specifications, in turn, relate very closely to the subject content criteria; close adherence to the criteria is a
condition of specification accreditation by Ofqual (the qualifications and examinations regulator).

Hence, all of the endorsed GCSE textbooks discuss social and ethical impacts of our increasing understanding of the genome in the contexts of health and disease, agriculture and classification, because this is specifically required by the subject content criteria. They also follow the criteria in introducing modern terminology, such as variant, and new ideas, such as the ideas that most features are affected by more than one gene and how non-coding DNA can affect phenotype, but there are differences in the extent to which the new terminology and ideas have been incorporated.

- Question 1: Do the GCSE textbooks explain the difference between genes and the genome?

The AQA textbook (Fullick, 2016) does not include the term ‘genome’ in its glossary, while the glossaries of the Edexcel, OCR A (Gateway Science) and OCR B (Twenty First Century Science) books present variations of the definition given in the subject content criteria (Levesley & Kearsey, 2016; Locke, 2016; Ingram, Moore, Skinner, & Winterbottom, 2016). Conversely, the AQA book is unique in stating explicitly (in the main text) that the genome includes mitochondrial DNA as well as chromosomal.

In explaining the difference between genes and the genome, all of the books make reference to non-coding DNA, linking to its role in controlling gene expression (as stated in the criteria); only the OCR B book defines ‘non-coding DNA’ in its glossary, and goes furthest in its discussion of the prevalence of non-coding DNA in the genome:

“Genes are very important, but they only make up about 1.5% of your genome. The remaining 98.5% of your DNA is more mysterious, and for a time scientists described it as ‘junk’. Scientists think that up to 80% of this DNA is important in controlling gene expression. This means it controls when the information in genes is used to make proteins.” (Ingram et al., 2016)

All of the books refer to the inheritance of genetic information, genetic material, chromosomes, genes and alleles, but only the OCR B book states explicitly that the genome is inherited (noting that half is inherited from each parent).

- Question 2: Do the GCSE textbooks make clear that most phenotypic features are affected by multiple regions of the genome, not just single genes?

Most of the books introduce the idea that most characteristics are affected by more than one gene before they explore the use of single gene crosses; the Edexcel book notes this after exploring single gene crosses. The OCR B book highlights dimples and the ability to roll one’s tongue as examples of human features regarding which scientists used to think they are controlled by a single gene but now think otherwise; these features are presented as single-gene characteristics in the other books. The AQA book includes the idea that mutations in non-coding DNA can have a big effect on phenotype, but does not give an example; the Edexcel book gives β-thalassaemia as an example, and the OCR B book cites the evolution of the opposable thumb. The OCR A book explains that a mutation in a non-coding sequence could prevent a gene being transcribed into mRNA and thus protein will not be produced, but stops short of linking this to effects on phenotype.

- Question 3: Do the GCSE textbooks make consistent use of modern terminology such as variant?

The term variant appears in the glossary of only the OCR B book, defined as “A different version of a gene, caused by a change (mutation) in the DNA”, while in the glossary of the AQA book the term alleles is defined as “Different forms of the same gene sometimes referred to as variants”. There is evidence of the term variant, where it occurs, being used interchangeably with allele across all of the books. The term is used very sparingly in the main texts of the AQA, Edexcel and OCR A books; for example, in the AQA book there is one reference to “alleles (variants)”, one to “variants (alleles)”, and a paragraph explaining natural selection that begins by stating that “New variants arise from a mutation” and
concludes that because of natural selection “the new allele will become common” (our emphases). In the Edexcel book a double page spread entitled “Genetic variants and phenotypes” does not use the term variant other than in the title, and includes the statement “A change in a gene that creates a new allele is called a mutation” (our emphasis). The OCR B book goes furthest, using the term “genetic variant” throughout the main text, often apparently in place of “allele” (for example: “The genetic variants an organism has make up its genotype”).

• Question 4: Do the GCSE textbooks discuss genome sequencing, or the field of genomics?

All of the books with the exception of the OCR B book include some information about the Human Genome Project; the AQA and OCR B books also mention ongoing efforts to sequence the genomes of humans (e.g. the 100,000 Genomes Project) and other organisms. The term ‘genomics’ could only be found in the OCR B book, which states that “The study of the structure and function of genomes is called genomics. This is an exciting and fast-moving area of science”; elsewhere it explains that “the science of genomics includes genome sequencing and bioinformatics”, and considers progression of the field from Mendel to genomics in a case study called “The grandfather of genomics”.

• Question 5: Do the GCSE textbooks discuss the social, technological and ethical impacts of genomics?

As noted previously, all of the endorsed GCSE textbooks discuss social and ethical impacts of our increasing understanding of the genome in the contexts of health and disease, agriculture and classification. All of the books consider how DNA sequencing could help us manage health and disease, with the AQA and OCR B books also mentioning testing for specific alleles. The OCR A book also includes basic ideas about human gene therapy.

In addition to considering the five questions, the sequence of genome-related ideas in each textbook was also analysed. The AQA, Edexcel and OCR B books all start with ideas about reproduction and variation, often using similarities and differences within families as a familiar, real-world context in which to explore scientific explanations; they then move on to explore the structure of DNA, protein synthesis, mutations, and explanations of inheritance. OCR A takes a different approach, starting with the structure of DNA and protein synthesis, before later considering variation, reproduction, inheritance and mutations; this approach follows the organisation of the content of the OCR A specification from cell-level systems, through ideas about ‘scaling up’, to organism-level systems. All of the textbooks present ideas about social, technological and ethical impacts of genome-related science after the sections on molecular genetics and inheritance.

AS/A level biology textbooks:
Notes from the analysis of the endorsed AS/A level Biology textbooks are presented in Table 2 in Appendix A5.

• Question 1: Do the AS/A level textbooks explain the difference between genes and the genome?

The term genome is defined in the glossaries of the Edexcel B books (Fullick, 2015a, 2015b) and the OCR A book (Fullick, Locke, & Bircher, 2015). It is also included in the glossary of the OCR B book (Fisher, Parker, & Wakefield-Warren, 2015), though the definition – “all the DNA that makes up the organism” (our emphasis) – is potentially problematic. Very few uses of the term genome were found in the text of the OCR B book, with “a mutation is a change of the nucleotide sequence of the genome” being a rare example. The Edexcel B, OCR A and OCR B books explicitly refer to mitochondrial DNA as part of the genome.

The Edexcel A (Salters-Nuffield) books do not have a glossary (Anderson, Hickman, et al., 2015; Anderson, Owens, et al., 2015). The first instance of the term genome found in the text states that “Together, all the genes in an individual (or species) are known as the genome” (our emphasis). However, later it is stated that “the genome… is all the DNA containing a full set of genes” and elsewhere that “a genome is all the DNA of an organism”.

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The term genome does not appear in the glossary of the AQA book (Toole & Toole, 2015), and the term could not be found until late in the text (chapter 8 of 9) where genome projects are discussed; the first occurrence found was “a complete map of all the genetic material in an organism (the genome) is obtained”.

None of the books define ‘non-coding DNA’ in their glossaries, but they all discuss it and the concepts of exons and introns in their texts. The AQA, Edexcel A, Edexcel B and OCR A books refer to the very low proportion (1.5-2%) of the human genome that is thought to be coding DNA.

- Question 2: Do the AS/A level textbooks make clear that most phenotypic features are affected by multiple regions of the genome, not just single genes?

All of the books explore models of monohybrid and dihybrid inheritance, though explicit acknowledgement that most human characteristics are affected by more than one gene could only be found in the AQA and Edexcel books.

In accordance with the subject content criteria, all of the books include ideas about how the genome and gene expression are regulated, including transcription factors and promoters, and they describe RNA splicing. Only the Edexcel A and both OCR books also discuss the concept of an operon, and mentions of epistasis were only found in the AQA and OCR A books. However, all the books develop ideas about epigenetics.

- Question 3: Do the AS/A level textbooks make consistent use of modern terminology such as variant?

The term variant could not be found at all in the AQA and Edexcel A books.

The term was not defined in the glossaries of any of the other three books, although the Edexcel B glossary defines an allele as “a version of a gene, a variant”, and the OCR B glossary defines an allele as “a gene variant”. There are only a few instances of the term being used in the texts of these three books, almost always together with (and as an alternative to) allele – e.g. “each gene exists in slightly different versions called alleles (variants)” in the Edexcel B book, and “For most genes there are a number of different possible alleles or variants” in the OCR A book. All of the books include discussion of population genetic diversity and selection in terms of alleles, with only the OCR B book also referring to variants in this context (it states both that “variants of genes that benefit organisms are selected and their frequency in a population increases” and that “natural selection acts to increase the frequency of beneficial alleles”).

- Question 4: Do the AS/A level textbooks discuss genome sequencing, or the field of genomics?

The term genomics could not be found in the AQA and OCR B books, though the AQA book refers to “genome projects” and the OCR B book to “genome studies”. All of the books discuss DNA sequencing, with the Edexcel books and the OCR A book referring to the Human Genome Project and one or more of the 1000, 10000 and 100000 Genomes projects. The AQA book does not name any specific sequencing projects but mentions that mapping the human genome took 13 years to complete. The Edexcel B book goes furthest with a double-page spread entitled ‘Timeline of Genomics’, adapted from material on the Your Genome website.

The AQA, Edexcel B and OCR A books refer to bioinformatics in supporting DNA sequencing, and the AQA, Edexcel A and OCR A books also mention proteomics. All of the books describe PCR and DNA profiling (though it is referred to as DNA fingerprinting in the AQA and OCR B books). References to DNA barcodes were found in all of the books except AQA.
Question 5: Do the AS/A level textbooks discuss the social, technological and ethical impacts of genomics?

In accordance with the subject content criteria, all of the books consider how genome-related technologies can be used to develop new medical processes (including testing for sequences associated with disease, personalised medicine, and human gene therapy), the use of transgenic organisms in industry and agriculture, and the use of DNA analysis to work out evolutionary relationships and classify organisms. They all also consider applications in forensics and paternity testing. Material about the use of gene knockout organisms in research and the use of genetically modified organisms for bioremediation was found in some, but not all, of the books.

In addition to considering the five questions, the sequence of genome-related ideas in each textbook was also considered. All of the books begin with molecular biology, considering the structure of nucleic acids amongst other biological molecules, with the Edexcel A book (for the context-led Salters-Nuffield Advanced Biology course) doing so in the context of cystic fibrosis. After that, the sequence of ideas is different in each book. It is not, however, necessarily the case that teachers’ schemes of learning follow the appropriate textbook’s structures.

Results of the survey of free resources that could be used in teaching and learning about genomics

As described in Section 2.2, the survey sample is effectively an opportunity/convenience sample of free resources available at the time of surveying in widely used and well-established sources that could be useful in teaching and learning about genomics in school. It is not an exhaustive list of everything that is available, but rather gives an overall impression of the landscape.

The first stage of the survey involved cataloguing genetics and genomics-related resources from websites that teachers commonly visit for biology teaching resources. No genetics or genomics-related resources were found on the websites of the Awarding Organisations AQA and Edexcel, but seven free resources were available on the OCR website (labelled as ‘Topic exploration packs’, ‘lesson elements’, and ‘Checkpoint tasks’). The trawl of the National STEM Centre e-library, the Nuffield Foundation’s Practical Biology website, the National Centre for Biotechnology Education (NCBE) website, and the Science and Plants for Schools (SAPS) website yielded a further 97 resources that were added into the catalogue.

The second stage involved cataloguing resources from three websites developed in England specifically to support genomics education. The ‘Your Genome’ website developed by the Wellcome Genome Campus Public Engagement team is the most up-to-date and well provisioned of the three websites examined, yielding 159 resources that were added to the catalogue. Nowgen’s ‘Genomics for Schools’ website hosts only a small selection of resources, and most of them can also be found in the National STEM Centre e-library; three resources were added to the catalogue from the ‘Genomics for Schools’ website. The ‘Genetics for Schools and Colleges’ section of the University of Leicester’s Virtual Genetics Education Centre (VGEC) website presents a series of ‘Topic’ articles, each with an associated list of links to relevant resources. Most of the links are to external websites and the VGEC website hosts only a small number of its own resources – 19 were added to the catalogue. Unfortunately, a number of the links are now out of date and point to web pages that are no longer available; of the links that remain active, a number point to whole websites (rather than individual resources) and it would require a significant additional investment of teachers’ time to locate specific resources within the linked site that relate to a particular learning outcome.

In total 285 resources were added to the catalogue.

Analysis of resource attributes

Date and target age

Two attributes were very inconsistently reported by the web pages hosting the resources in the catalogue: the year of publication or last update, and the target age range. Yet these attributes are immediately
useful to a teacher in deciding whether or not a resource is likely to be suitable for use in an episode of teaching. The year of publication or last update indicates whether the resource is likely to reflect not only the latest changes in the curriculum but also recent ideas and terminology in genomics. This is particularly problematic where a resource sets out to consider the latest developments or issues in this fast-moving field, but gives no indication of when it was produced.

Some but not all of the resources on the Your Genome website state a target age; all of the resources hosted on the National STEM Centre e-library state a target age range, although a number that are part of the Centre’s “Post 16 genetics and genomics” collection could be used with students from age 14 or even 11. The target ages stated in the catalogue represent our own judgements, based on the current versions of the English curriculum. Most of the resources in the catalogue (60%) would be suitable for use from age 14 (KS4/GCSE); fewer (31.9%) would only be suitable for use from age 16 (AS/A level), while only a small proportion would be suitable from age 11 (KS3) (Figure 2.1).

Resource type and purpose

Just over half (50.5%) of the 285 resources in the catalogue are articles (comprising text and any associated images). The next most prominent resource type was animations and videos, which represented almost a quarter (24.9%) of the catalogue. ‘Mixed mode’ resources made up 7.7% of the catalogue; these resources comprised bundles of resource types – for example an animation or presentation accompanied by an explanatory article and sometimes a student worksheet. Other resource types appearing in the catalogue were practical activities (5.3%), modelling activities (such as building models of DNA; 3.5%), diagrams and photographs (lacking any accompanying information or activity; 2.5%), role-play and debate activities (2.1%), pencil and paper activities (1.8%), games (1.1%), and one example each of a free downloadable book (entitled Chemistry and the Human Genome from the Royal Society of Chemistry), a presentation (lacking any accompanying activity) from the National STEM Centre, and a scheme of work entitled Introducing genomics from Nowgen. The numbers of each resource type recorded in the catalogue are shown in Figure 2.2 on the next page.

Figure 2.1. Distribution of catalogued resources across target age ranges

3 https://www.stem.org.uk/elibrary/collection/120066
The prevalence of articles, animations and videos without any supporting activity correlates with the results of the classification of resources according to the Biological Sciences Curriculum Study (BSCS) 5Es instructional model. Most of the catalogued resources were classified as Explanation (48.8%), meaning that they are instructional resources that transmit information directly to learners and facilitate teacher-led development of explanations; they do not facilitate enquiry, and do not provide evidence of learning. The next most common classification was Elaboration (34.7%), which comprises resources that challenge learners to apply key ideas in new contexts and/or that provide extension by introducing ideas that are beyond the scope of the curriculum. Exploration resources, which enable learners to develop their own explanations usually through enquiry, made up 12.6% of the catalogue. Engagement type items, which could be used to engage learners with an interesting – often real-world – context at the start of an episode of teaching related to genomics, represented 5.6% of the catalogued resources. Finally, only 4.9% of the resources were classified as Evaluation, meaning that they provide evidence of students’ learning (usually through a series of questions that assess knowledge and understanding of the ideas developed in the resource). The distribution of resources across the 5Es is shown in Figure 2.3.
Biological Sciences Curriculum Study (BSCS) 5Es instructional model. Of the remaining attributes recorded in the catalogue (see Figure 2.4), 16 of the resources (5.6%) were recorded as involving practical work (15 logged as practical activities and one logged as 'mixed mode' in the analysis of resource types). Of these 16 resources, seven developed the 1st of Millar and Abrahams’ purposes (developing learners’ scientific knowledge and understanding), while the remaining nine developed the 2nd purpose (developing learners’ ability to use scientific equipment or follow a standard procedure).

Two of the catalogued resources (0.7%) were recorded as developing numeracy or data analysis skills; these were the BRAF and KRAS activities from YourGenome.org, in which students analyse real sequence data. Five (1.8%) of the resources developed ideas about the nature of science (i.e. the processes of scientific enquiry, how scientific explanations are developed, and the role of the scientific community); all of these were from YourGenome.org and were articles exploring the Human Genome Project and contemporary examples of genome sequencing work. Ideas about risk, ethics and decision-making were developed in 34 (11.9%) of the resources.

Finally, 120 (42.1%) of the catalogued resources were recorded as going ‘beyond genetics’, indicating that they introduce modern ideas about the genome and genomics, going beyond what has traditionally been taught in schools about genetics and the structure and function of DNA. Of these, 83 originated on YourGenome.org.

Figure 2.4. Numbers of catalogued resources that include practical work, development of numeracy or data analysis skills, ideas about the nature of science (i.e. the processes of scientific enquiry, how scientific explanations are developed, and the role of the scientific community), consideration of risk, ethics or decision making, and ideas beyond what has traditionally been taught as part of genetics at school.
2.4 DISCUSSION AND RECOMMENDATIONS

2.4.1 What do we mean by ‘genomics education’?

It is clear from the review of the research literature presented in section 1 of this report that there is considerable variation in how different authors define ‘genomics education’ and, more fundamentally, ‘genomics’.

We have adopted a broad definition of genomics education in school, comprising conceptual development in four domains:

- ‘pre-genomics’ and ‘pro-genomics’ learning
- learning about the structure and function of the genome
- learning about methods of studying the genome
- learning about social, technological and ethical impacts of genomics.

We suggest neither that these four domains should be afforded equal amounts of teaching time, nor that each of them should be developed at every stage of the curriculum. But each domain contributes, at various points in the curriculum, to students’ developing understandings of what a genome is and of the place of genomics in modern science and society. A particular episode of teaching, or even a particular resource designed to support teaching, could contribute to learning in one or more of these domains (Figure 2.5).

Some ideas are not directly related to the genome at the time they are encountered (such as recognising during primary school that normally offspring vary and are similar but not identical to their parents), but they lay the foundations for the development of genomic explanations later. These ideas can be classed as pre-genomic learning.

Other ideas lie outside the field of genomics but may prompt students to think about and explain phenomena at the genomic level (such as explaining what happens during cell division). These ideas can be classed as pro-genomic learning.

Learning about the structure and function of the genome includes ideas of molecular biology, information flow, and inheritance, and could therefore be said to encompass all that has been traditionally taught as ‘genetics’ in school. It also includes ideas that go beyond traditional school genetics, for example those concerning the structure, functions and importance of non-gene (“non-coding”) regions of the genome. Learning about methods of studying the genome, and about social, technological and ethical impacts of genomics, provides fertile ground for the contextualised exploration of how scientific explanations are developed and how science and society interact. Many curricula require students to explore the nature of science, including the reformed...
National Curriculum in England through its ‘Working Scientifically’ strand, and genomics education provides historical and contemporary contexts in which they can be developed.

2.4.2 The principle of curriculum coherence

Numerous studies have considered the issue of ‘curriculum coherence’ and its role in reducing variability in what is taught in the classroom. Schmidt and Prawat (2006) define two aspects of curriculum coherence:

• the sequencing of curriculum content in an understandable and logical way;
• the correspondence of content in the various mediating instruments.

The first aspect of curriculum coherence could be thought of as vertical alignment. In the context of genomics education, it necessitates the presentation of a coherent and appropriate learning progression (or a series of learning progressions) related to genomics from the early years of the school biology curriculum to the end.

The second aspect of curriculum coherence could be thought of as horizontal alignment. In the context of genomics education, it would necessitate at each stage of the curriculum a coherent presentation of genomics-related ideas across the various mediating instruments, including policy documents (such as the National Curriculum), Awarding Organisation specifications, textbooks and other teaching resources. Those seeking to promote and modernise genomics education in school science must consider both aspects of curriculum coherence.

With regard to the vertical aspect, efforts focussed only on GCSE and A level will be undermined if the lower end of the curriculum does not provide a solid foundation upon which to build; for example, numerous studies have shown that misconceptions established at early stages are persistent and difficult to change (for example, directly related to genomics education: Lewis, Leach and Wood-Robinson, 2000; Williams, 2012). Similarly, with regard to the horizontal aspect: efforts focussed only on particular instructional instruments, such as resources, will be of limited value unless the curriculum, specifications and textbooks are all delivering the same message.

However, different instruments drive change in classroom practice to different degrees. All maintained schools in England must follow the National Curriculum, and while academies (including free schools) and independent schools are not required to follow the National Curriculum it appears to be common to use it as the basis for their curriculum. Martin, Mullis, Foy, and Stanco (2012) report that just under half (49%) of sampled science teachers in England use textbooks either as a basis for instruction or as a supplement. It is difficult to predict how many teachers would make use of any particular resource or set of resources distributed online via a website such as YourGenome.org, but it is likely to be a small percentage. At GCSE and A level, the content-heavy curriculum and limited teaching time result in teachers tending to look for resources that tightly align with the specification they are teaching. Textbooks are endorsed by the Awarding Organisation if they closely match the specification, and specifications are only accredited by the regulator Ofqual if they closely match the curriculum. Therefore, working with policymakers to ensure appropriate curriculum content may be the most powerful way to drive change in the other instruments and in what is taught in classrooms.

2.4.3 Vertical alignment of genomics-related content in the curriculum

Appendix 4 presents the results of a simple analysis of the vertical alignment of genome-related content in the current school biology curriculum in England, wherein the genomics-related content of the curriculum documents has been arranged according to our four domains of genomics education. Only one of the domains, ‘pre- and pro-genomics learning’, is represented in the primary school curriculum (Key Stages 1 and 2, age 5–11). Three of the domains are represented in lower secondary school (Key Stage 3, ages 11–14), with only ‘Methods of studying the genome’ absent.

All four domains are represented at GCSE level (Key Stage 4, age 14–16) and AS/A level (age 16–18). Thus, the current curriculum introduces the
domains of genomics education in a phased and broadly age-appropriate way, though the detail of what is included in each domain at each stage requires further scrutiny.

The ideas within each domain can be seen as forming a learning progression, and thus the four domains represent four learning progressions in genomics education. Further work should consider issues including the best sequence for the ideas within each learning progression; whether individual ideas within each learning progression are introduced at the appropriate age; whether there is unnecessary repetition of ideas; whether any ideas need to be added to fill gaps in the learning progressions; whether any of the ideas need to be updated in keeping with advances in genomics; and whether any ideas could be removed to lighten the content load and give students and teachers more time to develop core concepts.

Further research of the sort recommended in section 1 of this report will add to the existing evidence base related to the effectiveness and age-appropriateness of particular teaching sequences. Useful insights may also be drawn from comparisons with international curricula and from major curriculum development work undertaken in other countries such as the USA (AAAS Benchmarks for Science Literacy [Project 2061]) and the Australian state of Victoria (Department of Education and Training Science, 2014).

The reformed GCSE biology subject content criteria introduce the term ‘variant’ into the curriculum for the first time, an indication of attempts to modernise the genetics content. However, the term was not included in the reformed AS/A level biology content criteria, and so there is some lack of vertical coherence. The omission of the word variant may be due to the fact that the AS/A level criteria are expressed in much less detail than the GCSE criteria, but its omission has meant that it is largely absent from the Awarding Organisations’ AS/A level biology specifications, though the term allele is still used frequently.

In all of the AS/A level specifications, evolution is considered as the natural selection of alleles (rather than variants as in the GCSE criteria).

Had the term been used in the AS/A level criteria, it may have been adopted more consistently into the specifications.

We recommend that those seeking to promote genomics education in school collaborate with science education researchers and curriculum developers to develop evidence-based guidance for improvement of the genomics-related content of the curriculum. The vertical aspect of curriculum coherence will be a key concern. The aim should be to influence policymakers and curriculum developers who will redevelop the curriculum content at some (as yet unknown) future date.

Recommendation 1: Develop evidence-informed guidance on genomics-related content in preparation for future reform of the school science curriculum, including guidance on sequencing and age-appropriateness of ideas, with a view to influencing policy.

During the most recent round of curriculum reform in England (2013-2016), the tight development timescale was criticised for not being conducive to a thorough consideration of curriculum coherence or to evidence-based decision making, and for precluding the piloting of any aspects of the new curriculum in schools. Although there were windows of public consultation, organisations such as the Royal Society of Biology have reported that they were given very little time to prepare thorough, evidence-informed responses (personal communication, 2016).

In response, the Royal Society of Biology and the corresponding learned societies for chemistry and physics have set up Curriculum Committees to begin formulating guidance on the societies’ positions regarding the content of the school curriculum, such that they can be well prepared in advance of any future announcement of curriculum reform. Those seeking to influence the genomics content of the curriculum should consider working with the Curriculum Committee of the Royal Society of
Biology to present a strong, unified voice. We would also advise that they should, in any case, undertake or support the work necessary to produce thorough, evidence-informed guidance sooner rather than later.

2.4.4 Horizontal alignment of genomics-related content in the curriculum

Appendix 5 presents the results of an analysis of the genome-related content in the Awarding Organisation-endorsed textbooks at GCSE and AS/A level. Taken together with Tables 3 and 4 in Appendix 4, it outlines the horizontal alignment of genomics ideas in the curriculum in England and in some of the associated textbooks.

Two aspects of control in England have resulted in close horizontal alignment between the science curriculum and textbooks at GCSE and AS/A level. The first is the controlling influence of Ofqual, the qualifications and examinations regulator, which will only accredit Awarding Organisation specifications that closely cover the programmes of study set out in the National Curriculum (and only accredited specifications can be assessed for the awarding of GCSE and AS/A level qualifications). The second is the controlling influence of national accountability measures (such as league tables) based on GCSE and AS/A level examination results, which motivate schools to choose textbooks endorsed by the Awarding Organisations because they offer close coverage of the specifications and examination-specific preparation for students.

Thus, the endorsed textbooks surveyed in this report closely match the Awarding Organisation specifications and, in turn, the content specified in the curriculum. The textbooks follow the curriculum criteria in introducing modern terminology and ideas about the genome. Nevertheless, there are differences in the extent to which modern terminology and genomic ideas have been incorporated.

At GCSE level, all of the textbooks in the survey discuss the concept of the genome, making clear that it is more than just genes - they refer to the existence and importance of non-coding DNA. They make use of the term variant, but only inconsistently and very sparingly across the GCSE textbooks. It may or may not be considered important that most school leavers should be familiar with the term, or understand it, and therefore that the word variant should be included in textbooks and in teaching; it is highlighted here as an example of problematic attempts to modernise the content and language of genetics in the curriculum.

The presentation of the term in the criteria as an alternative to allele with a solidus between the two terms (the criteria require students to “explain the terms... allele/variant”) gives specification writers, textbook authors and teachers the opportunity to overlook ‘variant’ in favour of the more familiar ‘allele’. Use of the term allele is entrenched in school biology, and behaviour is unlikely to change without stronger pressure from the curriculum and associated mediating instruments.

In our experience of producing textbooks for GCSE and AS/A level science courses, new editions that are produced during periods of curriculum reform tend to reuse material from previous editions, updating it where necessary. The tight timescale in which the most recent round of curriculum reform was completed resulted in updates that may not have been as thorough, consistent and radical as could otherwise have been the case.

Another well-intentioned change in the reformed GCSE criteria is the requirement for students to “describe the development of our understanding of genetics including the work of Mendel”. Accordingly, all of the specifications and textbooks include the work of Mendel, but they differ in the extents to which they cover contemporary genetics/genomics and the progression of the field from Mendel to today. The merits of highly detailed versus generic curriculum statements have been debated, but in general when the curriculum is less specific the importance of textbooks and other mediating instruments in defining what is taught increases (Oates, 2014).

At GCSE level, the endorsed textbook for OCR Biology B (Twenty First Century Science) goes furthest in the use of terminology such as variant and discussion of the prominence and importance of non-coding DNA;
the genetics content of the book was written by Ingram and edited by Moore. At AS/A level, the books written by Fullick include various double-page features such as a ‘Timeline of Genomics’ and ‘Think Bigger’ articles on contemporary issues is genetics and genomics adapted from materials developed for the YourGenome.org website.

It is notable that Ingram, Moore and Fullick visited the Wellcome Genome Campus with the Biology Education Research Group (BERG) during the curriculum reform period, and sought advice from Wellcome Genome Campus Public Engagement staff while developing content for the books. We recommend that those seeking to promote genomics education in school should collaborate with publishers and textbook authors during future periods of curriculum reform to develop appropriate genomics-related content in textbooks. The horizontal aspect of curriculum coherence will be a key concern, in that there should be strong alignment between the intentions of the curriculum and the development of ideas in the textbooks. Textbook authors (typically good general biologists but not specialists in any particular field) will benefit from guidance on the appropriate use of modern genomics terminology, insights into the latest thinking in the field, recommendations on appropriate sequencing of ideas, and access to case studies of research and real-world applications that can be used to develop ideas in context.

Recommendation 2:
Work with publishers and textbook authors to develop appropriate and up-to-date content in textbooks that is aligned with (and helps to define) the intended learning outcomes in the curriculum.

2.4.5 Provision of supplementary learning and teaching resources

Other teaching and learning resources of the sort made available via websites such as YourGenome.org are likely to be less widely used than the endorsed textbooks, but they have an important role to play in supporting genomics learning. They can be used to develop ideas in more depth, facilitate enquiry-based learning, develop practical skills, generate evidence of learning and reveal student misconceptions. Importantly for a fast-moving field such as genomics, they can also be updated more easily and frequently than textbooks, and with no cost implication for schools.

Work to develop research-informed guidance on sequencing and age-appropriateness of ideas in genomics education could and perhaps should include the development and trialling of resources as part of interventions in schools. Feedback collected from trials of these resources in school should be used to refine and improve them, as part of an iterative process, with the end result being high-quality evidence-informed resources suitable for wider use. We recommend that those seeking to promote genomics education in school work with science education researchers to develop new resources in this way.

The catalogue of resources compiled by this survey is dominated by ‘Explain’ type resources, in particular articles, animations and videos. These are instructional resources that transmit information directly to learners and facilitate teacher-led development of explanations. They are undoubtedly valuable, but do not facilitate enquiry and do not provide evidence of learning. The latter point could be remedied by the inclusion of a set of questions or a student worksheet with each resource; students’ answers to these questions will provide feedback to both the teacher and the student about what the student has learned from the resource, and will inform what happens next. This will help optimise the usefulness of each resource.

Recommendation 3:
Work with science education researchers to develop and trial new resources for genomics education to support teaching of the recommended learning progressions.
It should be noted that the 5Es model was intended to act as a guide for the design of teaching strategies, and was not necessarily intended to be applied retrospectively to teaching and learning resources after they have been written. Thus, resources written without the 5Es model in mind do not always fit neatly into one of the Es. Nevertheless, it can be insightful to classify existing resources according to the phase of the 5Es instructional model in which they could be usefully used, as the results offer a form of gap analysis to suggest where future resource development efforts could be focussed.

Efforts to develop new resources should take into consideration the relative lack of resources aimed at the Engagement, Exploration and Evaluation phases of conceptual change, and various types of resources that might be developed beyond articles, animations and videos. One would not necessarily expect that all of the different phases and item types should be represented equally, but there is a need to expand current provision in certain areas.

**Recommendation 4:**
Work to make existing resources more useful and diverse in type, for example by adding questions to ‘Explain’ resources to facilitate the collection of evidence of learning, and by clearly labelling the resource with information about the target age range and date of last update.

It was apparent from the searches conducted for this study that there is an enormous number of genetics and genomics resources freely available on the internet, of various ages, origins and quality. It is entirely possible that a teacher searching for genomics teaching resources would feel overwhelmed by the choice, and inevitable that they would not have sufficient time to survey all of the resources to find out which ones are the most up-to-date, which are of the highest quality, and which develop genomics ideas beyond what has traditionally been taught in school genetics. Indeed, we stopped adding resources to the catalogue after the first two stages of searching; these two stages alone, which focussed on websites of British origin, yielded 285 resources – many more than a teacher would have time to peruse when planning a course of genomics teaching. We did not expand the search to include websites developed in the USA to support genomics education, such as the National Human Genome Research Institute website (www.genome.gov), the BioInteractive website from the Howard Hughes Medical Institute (www.hhmi.org/biointeractive), the Learn Genetics website from the University of Utah (learn.genetics.utah.edu), the Dolan DNA Learning Centre website from Cold Spring Harbor Laboratory (www.dnalc.org), or the GeneEd website from the US National Library of Medicine (geneed.nlm.nih.gov).

Publication of the catalogue in some form may help teachers to locate resources to fulfil particular classroom needs. Resources from American websites could be added into the catalogue if it was felt useful to expand it further.

**Recommendation 5:**
Consider the publication of a catalogue of available resources for genomics education, alongside an analysis, in a form that may help teachers to locate resources to fulfil particular classroom needs.

**2.4.6 Specific comments about resources on YourGenome.org**

As described in Section 2.5, the survey of teaching resources undertaken in this review catalogued genetics and genomics-related resources from websites that teachers may visit for biology teaching resources, including three websites developed in England specifically to support genomics education: the ‘Your Genome’ website developed by the Wellcome Genome Campus Public Engagement team, Nowgen’s ‘Genomics for Schools’ website, and the ‘Genetics for Schools and Colleges’ section of
the University of Leicester’s Virtual Genetics Education Centre (VGEC) website. The YourGenome.org website (hereafter, YG) was found to be the most up-to-date and well provisioned of the three genetics and genomics-specific websites examined, and yielded 159 resources for the catalogue (making up 56% of all resources catalogued).

A teacher arriving at YG is likely to be looking for resources for a specific episode of teaching, such as a particular topic or even a particular lesson within a topic, and is also likely to have very limited time in which to locate suitable resources. Teachers of GCSE and A level courses in England may have a particular learning outcome or section from the Awarding Organisation specification in mind, and may be disappointed to find that the resources on YG are not tagged with specification references. However, we would advise against tagging the YG resources in such a way; doing so would only be of use in England, and YG must usefully remain an international platform; and doing so would also create a significant burden to update the references whenever the specifications are rewritten. Currently, YG resources can be browsed in five broad categories (“In the cell”, “Methods and technology”, “Targeting disease”, “Society and behaviour” and “Animals and plants”), and can be searched using key words or concepts; this organisation of the resources according to key concepts, rather than according to any particular version of any particular national curriculum or course, is internationally applicable and resilient to curriculum churn.

One attribute of a resource which is of immediate concern to a teacher in deciding whether or not it is likely to be suitable for use in an episode of teaching is the target age range. Most of the activities, videos and interactives on YG are labelled with a target age range, however the articles are not. In some cases, the target age range stated for a resource seems to be based on the nature of the activity rather than on the age-appropriateness of the key concept being developed by the resource and where this key concept might fit in a learning progression. For example, in the activity ‘Sequence Bracelets’ students are given a printed DNA sequence made up of the coloured letters A, T, C and G; they assemble one chain of a bracelet using coloured beads corresponding to the DNA sequence they have been given, and then have to assemble a complimentary chain using base-pairing rules. The stated age range for this activity is 10 years+ (KS2+). Building a bracelet from beads may be an appropriate activity for a 10-year-old. However, the key concepts of which this activity aims to build understanding (that information is encoded in DNA using four bases A, T, C and G, and that the bases of the two strands in DNA are arranged according to complimentary base-pairing rules) are not likely to be formally taught until age 14+ (as is the case in the English national curriculum). At age 10, primary school students will be exploring basic explanations for family resemblance and formalising the idea that some but not all features can be inherited; this is pre-genomic learning. They will be formally introduced to cells, the nucleus, and DNA as genetic material from age 11; so an activity that develops understanding of the structure of the genome is unlikely to be appropriate for age 10. The ‘Sequence Bracelets’ activity may be better labelled as 14+, where it would correspond to the understanding that students are expected to develop at that age.

Recommendation 1 for YourGenome.org:
Add a target age range to resources currently lacking this information, e.g. articles, and check that the target age ranges stated on existing resources are appropriate for students’ conceptual development.

Our age-range recommendations for all YG resources (including articles) are given in the catalogue that accompanies this report; they do not always agree with the age-range recommendations stated on the YG website. On the basis of our analysis, 8.8% of the YG resources would be suitable for use from age 11, 61.9% from age 14, and 29.4% from age 16. We could not find any resources that could support pre-genomic learning before age 11.
Recommendation 2 for YourGenome.org:
Develop new resources targeted at young students up to age 11, and suitable for use by non-science-specialist teachers in primary schools, with the specific aim of supporting pre-genomic learning.

Another attribute of teaching resources that is likely to affect a teacher’s decision about whether it is suitable for use is the date it was published or last updated. Genetics and genomics are fast-moving fields, and it is useful for teachers to be able to check at a glance whether a resource is up to date. Helpfully, the YG webpages state a “last updated” date for all resources.

Most of the YG resources are articles (123; 43.2%); next most common are animations and videos (16; 5.6%). There are 11 ‘mixed mode’ resources (3.9%); these resources comprised bundles of resource types – for example an animation or presentation accompanied by an explanatory article and sometimes a student worksheet. There are three role-play/debate activities, three hands-on modelling activities, two pencil-and-paper activities, one game, only two resources that develop numeracy or quantitative data analysis skills (“BRAF: from gene to cancer therapy” and “KRAS: Cancer Mutation Activity”), and only one activity that involves practical/experimental work (“Extracting DNA from fruit”). Practical and experimental work can be used to support the development of students’ understanding of scientific phenomena, their understanding of scientific methods and the empirical nature of science, and their ability to use apparatus and follow practical procedures (Millar & Abrahams, 2009), and it has been argued that they increase students’ motivation and interest in science (Holman, 2017).

The majority of YG resources do not state what key concept they are intended to develop or test understanding of, or what key competency they are intended to develop or test. Put more formally, they lack an explicitly stated learning objective. Teachers are likely to have a clear learning objective in mind for each activity they use in a lesson, it may help their planning if each YG resource included a learning objective – so that they can understand why it was developed and, thus, how it might best be used.

Recommendation 3 for YourGenome.org:
Develop new resources to expand the variety of activity types, including activities that involve practical or experimental work and activities that develop students’ numeracy or quantitative data analysis skills.

The YG resources were classified according to the Biological Sciences Curriculum Study (BSCS) 5Es instructional model. According to our analysis, 7.5% of the resources are Engagement type items, which could be used to engage learners with an interesting – often real-world – context at the start of an episode of teaching related to genomics. 6.3% could be used for Exploration, to enable learners to develop their own explanations (often through enquiry). 42.5% were classified as Explanation, meaning that they are instructional resources that transmit information directly to learners and facilitate teacher-led development of explanations; they do not facilitate enquiry (in which students build their own explanations), and do not provide evidence of learning; the prevalence of articles, animations and videos without any supporting activity correlates with this finding.

Recommendation 4 for YourGenome.org:
State the learning objective of each resource – i.e. what key concept they are intended to develop or test understanding of, or what key competency they are intended to develop or test.
The most common classification was Elaboration (43.8%), which comprises resources that challenge learners to apply key ideas in new contexts and/or that provide extension by introducing ideas that are beyond the scope of the curriculum; the vast majority of these were articles, but the three role-play/debate activities were also included in this category. Finally, only three YG resources (1.9%) were classified as Evaluation, meaning that they provide evidence of students’ learning (often from questions that test understanding of the key concept, or tasks that test a key competency). This kind of evidence enables both students and teachers to check what the students have gained from the activity and whether they have met the learning objective, and can be used formatively to decide what happens next.

Recommendation 5 for YourGenome.org:
Ensure that new and existing resources:
• promote ‘active learning’ (in which students do more than simply receive information);
• include, for example, questions that test understanding of the key concept, or tasks that test a key competency, so as to provide evidence of what students have gained from the activity and whether they have met the learning objective.
2.5 REFERENCES


national control of education: issue or non-issue? Journal of
Curriculum Studies, 38(6), 641–658. doi:10.1080/00220270600682804
A1 KEYWORDS FOR THE SEARCH STRATEGY

Scientific
Genome
Genomic
Genetic
Epigenetics
Gene
Genes
Genetic code
DNA
DNA sequence
DNA sequencing
Gene technology
Cancer
Organism
Inheritance
Mendelian
Human Genome Project
Human genome
Bioinformatics
Genome informatics
Complex disease
Central Dogma
Chromosome
Non-Coding
Untranslated Region
Regulatory (region)
Systems Biology
Gene Mapping
Genetic Engineering
Genotyping
Intron
Exon

Educational/pedagogical
Teaching
Secondary School
High School
College
Pedagogical
Pupil
Misconception
Understanding
Concept
Resource
Authentic science

Primary School
Elementary School
Sixth Form
Pedagogy
Student
Learner
Misunderstanding
Assessment
Activity
Inquiry based learning
A2 LONG LIST OF 112 IDENTIFIED RESEARCH STUDIES (THE RESEARCH ‘LANDSCAPE’)


Dagher, Z. (2014). The relevance of history of biology to teaching and


Gericke, N., & Hagberg, M. (2010a). Conceptual incoherence as a result of


of Science Education, 26(2), 195-206.


McQueen, J., Wright, J. J., & Fox, J. A. (2012). Design and implementation of a genomics field trip program aimed at secondary school students. Plos Computational Biology, 8(8), 6. doi:10.1371/journal.pcbi.1002636


A3 SHORT LIST OF 29 STUDIES FOR DETAILED REVIEW


Smith, M. U., & Adkison, L. R. (2010). Updating the model definition of
### Learning outcomes related to genomics in Key Stages 1 and 2 of the National Curriculum in England (Department for Education, 2013b)

<table>
<thead>
<tr>
<th>Stage</th>
<th>Pre- and pro-genomics learning</th>
</tr>
</thead>
</table>
| **KS1** | notice that animals, including humans, have offspring which grow into adults  
identify and name a variety of plants and animals in their habitats |
| **KS2** | describe the life process of reproduction in some plants and animals  
recognise that living things produce offspring of the same kind, but normally offspring vary and are not identical to their parents  
recognise that living things can be grouped in a variety of ways  
describe how living things are classified into broad groups according to common observable characteristics and based on similarities and differences, including micro-organisms, plants and animals  
identify how animals and plants are adapted to suit their environment in different ways and that adaptation may lead to evolution |
## Table 2: Learning outcomes related to genomics in Key Stage 3 of the National Curriculum in England (Department for Education, 2013a).

<table>
<thead>
<tr>
<th>Stage</th>
<th>Pre- and pro-genomics learning</th>
<th>Structure and function of the genome</th>
<th>Social, technological and ethical impacts of genomics</th>
</tr>
</thead>
<tbody>
<tr>
<td>KS3</td>
<td>cells as the fundamental unit of living organisms&lt;br&gt;the function of the nucleus&lt;br&gt;reproduction in humans (as an example of a mammal), including the structure and function of gametes, and fertilisation&lt;br&gt;reproduction in plants, including wind and insect pollination, fertilisation&lt;br&gt;differences between species&lt;br&gt;continuous and discontinuous variation between individuals within a species&lt;br&gt;variation between species and between individuals of the same species means some organisms compete more successfully, which can drive natural selection</td>
<td>heredity as the process by which genetic information is transmitted from one generation to the next&lt;br&gt;a simple model of chromosomes, genes and DNA in heredity, including the part played by Watson, Crick, Wilkins and Franklin in the development of the DNA model</td>
<td>the importance of maintaining biodiversity and the use of gene banks to preserve hereditary material</td>
</tr>
</tbody>
</table>
Table 3: Learning outcomes related to genomics in Key Stage 4, as specified in the GCSE Biology and GCSE Combined Science subject content criteria (Department for Education, 2015a, 2015b). Italicised statements are not assessed in Combined Science examinations.

<table>
<thead>
<tr>
<th>Stage</th>
<th>Pre- and pro-genomics learning</th>
<th>Structure and function of the genome</th>
<th>Methods of studying the genome</th>
<th>Social, technological and ethical impacts of genomics</th>
</tr>
</thead>
<tbody>
<tr>
<td>KS4</td>
<td>explain how the main sub-cellular structures of eukaryotic cells (plants and animals) and prokaryotic cells are related to their functions, including the nucleus</td>
<td>explain how the genetic material and plasmids of eukaryotic cells (plants and animals) and prokaryotic cells are related to their functions</td>
<td>describe the development of our understanding of genetics including the work of Mendel</td>
<td>discuss the potential importance for medicine of our increasing understanding of the human genome</td>
</tr>
<tr>
<td></td>
<td>describe the process of mitosis in growth, including the cell cycle</td>
<td>describe the genome as the entire genetic material of an organism</td>
<td></td>
<td>describe genetic engineering as a process which involves modifying the genome of an organism to introduce desirable characteristics</td>
</tr>
<tr>
<td></td>
<td>describe cancer as the result of changes in cells that lead to uncontrolled growth and division</td>
<td>describe DNA as a polymer made up of two strands forming a double helix</td>
<td></td>
<td>describe the main steps in the process of genetic engineering</td>
</tr>
<tr>
<td></td>
<td>explain the importance of cell differentiation</td>
<td>describe DNA as a polymer made from four different nucleotides; each nucleotide consisting of a common sugar and phosphate group with one of four different bases attached to the sugar</td>
<td></td>
<td>describe and explain some possible biotechnological and agricultural solutions, including genetic modification, to the demands of the growing human population</td>
</tr>
<tr>
<td></td>
<td>explain the role of meiotic cell division in halving the chromosome number to form gametes</td>
<td>describe simply how the genome, and its interaction with the environment, influence phenotype</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>state that there is usually extensive genetic variation within a population of a species</td>
<td>explain the importance of amino acids in the synthesis of proteins</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>describe evolution as a change in the inherited characteristics of a population over time through a process of natural selection which may result in the formation of new species</td>
<td>recall a simple description of protein synthesis</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>explain simply how the structure of DNA affects the proteins made</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Table 3 (continued): Learning outcomes related to genomics in Key Stage 4, as specified in the GCSE Biology and GCSE Combined Science subject content criteria (Department for Education, 2015a, 2015b). Italicised statements are not assessed in Combined Science examinations.

<table>
<thead>
<tr>
<th>Stage</th>
<th>Pre- and pro-genomics learning</th>
<th>Structure and function of the genome</th>
<th>Methods of studying the genome</th>
<th>Social, technological and ethical impacts of genomics</th>
</tr>
</thead>
<tbody>
<tr>
<td>KS4</td>
<td>explain how evolution occurs through natural selection of variants that give rise to phenotypes best suited to their environment</td>
<td>recall that all variants arise from mutations, and that most have no effect on the phenotype, some influence phenotype and a very few determine phenotype</td>
<td>describe how genetic variants may influence phenotype; in coding DNA by altering the activity of a protein; in non-coding DNA by altering how genes are expressed</td>
<td>explain some of the possible benefits and risks, including practical and ethical considerations, of using gene technology in modern agriculture and medicine</td>
</tr>
<tr>
<td></td>
<td></td>
<td>describe how genetic variants may influence phenotype; in coding DNA by altering the activity of a protein; in non-coding DNA by altering how genes are expressed</td>
<td>explain the following terms: gamete, chromosome, gene, allele/variant, dominant, recessive, homozygous, heterozygous, genotype and phenotype</td>
<td>describe the impact of developments in biology on classification systems</td>
</tr>
<tr>
<td></td>
<td></td>
<td>explain single gene inheritance</td>
<td>predict the results of single gene crosses</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>recall that most phenotypic features are the result of multiple genes rather than single gene inheritance</td>
<td>describe sex determination in humans</td>
<td></td>
</tr>
</tbody>
</table>
Table 4: Learning outcomes related to genomics in AS/A level, as specified in the GCE Biology subject content criteria (Department for Education, 2014a).

<table>
<thead>
<tr>
<th>Stage</th>
<th>Pre- and pro-genomics learning</th>
<th>Structure and function of the genome</th>
<th>Methods of studying the genome</th>
<th>Social, technological and ethical impacts of genomics</th>
</tr>
</thead>
<tbody>
<tr>
<td>AS/A level</td>
<td>proteins have important roles and functions related to their properties during the cell cycle genetic information is copied and passed to daughter cells daughter cells formed during mitosis have identical copies of genes while cells formed during meiosis are not genetically identical the variety of life, both past and present, is extensive, but the biochemical basis of life is similar for all living things biodiversity refers to the variety and complexity of life and may be considered at different levels biodiversity can be measured, for example within a habitat or at the genetic level transfer of genetic information from one generation to the next can ensure continuity of species or lead to variation within a species and possible formation of new species adaptation and selection are major factors in evolution and make a significant contribution to the diversity of living organisms adaptations of organisms to their environments can be behavioural, physiological and anatomical reproductive isolation can lead to accumulation of different genetic information in populations potentially leading to formation of new species</td>
<td>nucleic acids (DNA and RNA) have important roles and functions related to their properties the sequence of bases in the DNA molecule determines the structure of proteins, including enzymes the genome is regulated by a number of factors</td>
<td>sequencing projects have read the genomes of organisms ranging from microbes and plants to humans; this allows the sequences of the proteins that derive from the genetic code to be predicted gene technologies allow study and alteration of gene function in order to design new industrial and medical processes originally classification systems were based on observable features but more recent approaches draw on a wider range of evidence to clarify relationships between organisms</td>
<td>gene technologies allow study and alteration of gene function in order to better understand organism function</td>
</tr>
</tbody>
</table>
Does the book... | GCSE textbook |
---|---|
...explain the difference between genes and the genome? | for AQA GCSE Biology (Fullick, 2016) | genome and non-coding DNA not in glossary  
main text explains: "Genes are small sections of DNA" and the genome is “the entire genetic material of the organism” including chromosomes and mitochondrial DNA  
non-coding DNA affects gene expression ("switching genes, or parts of genes, on and off")  
can make different proteins from the same gene  
references to inheritance of genetic information, chromosomes, genes, alleles, and mitochondrial DNA, but not inheritance of the genome |
| for Edexcel GCSE Biology (Levesley & Kearsey, 2016) | genome in glossary ("All of the DNA in an organism. Each body cell contains a copy of the genome.")  
non-coding DNA not in glossary  
main text explains: "Along the length of a DNA molecule are sections that each contain a code for making a protein. These DNA sections are genes.”  
during transcription, RNA polymerase attaches to non-coding region in front of a gene  
references to inheritance of genetic material, genes and alleles, but not the genome |
| for OCR GCSE Biology A (Gateway Science) (Locke, 2016) | genome in glossary ("All the genetic material present in an organism.")  
non-coding DNA not in glossary  
“There are specific sequences of DNA bases found before a gene, which trigger the process of transcription. These are located within the non-coding sections of DNA”  
references to inheritance of genetic material, genes and alleles, but not the genome |
| for OCR GCSE Biology B (Twenty First Century Science) (Ingram, Moore, Skinner, & Winterbottom, 2016) | genome in glossary ("The entire genetic material of an organism")  
non-coding DNA in glossary ("Regions in the genome that do not store code for making proteins, but that can affect gene expression.")  
main text explains: “A gene is a region of DNA”  
“Genes are very important, but they only make up about 1.5% of your genome. The remaining 98.5% of your DNA is more mysterious, and for a time scientists described it as ‘junk’. Scientists think that up to 80% of this DNA is important in controlling gene expression. This means it controls when the information in genes is used to make proteins.”  
references to inheriting genetic information, genetic material, the genome (half of it from each parent), chromosomes, alleles and genetic variants
Table 1 (continued): GCSE textbooks

<table>
<thead>
<tr>
<th>Does the book...</th>
<th>GCSE textbook</th>
</tr>
</thead>
</table>
| ...make clear that most phenotypic features are affected by multiple regions of the genome, not just single genes? | for AQA GCSE Biology (Fullick, 2016)  
notes that “most characteristics are controlled by several genes interacting” – appears before consideration of the use of single gene crosses  
changes in gene expression can affect phenotype  
mutations in non-coding DNA can have big effect on phenotype, but no example given  
for Edexcel GCSE Biology (Levesley & Kearsey, 2016)  
“Most human characteristics are controlled by many genes, not just one” – after single-gene crosses  
ideas about gene expression described but the term not used  
β-thalassaemia as example of effect of mutation in non-coding DNA  
for OCR GCSE Biology A (Gateway Science) (Locke, 2016)  
“most features are caused by multiple genes” – before single gene crosses  
ideas about gene expression described but the term not used  
idea that a mutation in a non-coding sequence could prevent a gene being transcribed into mRNA, and protein will not be produced; stops of short of linking this to effects on phenotype  
for OCR GCSE Biology B (Twenty First Century Science) (Ingram, Moore, Skinner, & Winterbottom, 2016)  
“A very small number of your features are controlled by a single gene” – before single gene crosses  
“Scientists have changed their thinking about a number of features, such as dimples, curved thumbs, and the ability to roll your tongue. Scientists used to think these features were controlled by a single gene. Now we know the story is more complicated. Most of your features depend on multiple genes and on other regions of the genome.” – before  
evolution of the opposable thumb as an example of the effect of the effect of mutation in non-coding DNA |
Table 1 (continued): GCSE textbooks

<table>
<thead>
<tr>
<th>Does the book...</th>
<th>for AQA GCSE Biology (Fullick, 2016)</th>
<th>for Edexcel GCSE Biology (Levesley &amp; Kearsey, 2016)</th>
<th>for OCR GCSE Biology A (Gateway Science) (Locke, 2016)</th>
<th>for OCR GCSE Biology B (Twenty First Century Science) (Ingram, Moore, Skinner, &amp; Winterbottom, 2016)</th>
</tr>
</thead>
<tbody>
<tr>
<td>...make consistent use of modern terminology such as variant?</td>
<td>variant not in glossary (alleles defined as “different forms of the same gene sometimes referred to as variants”) two references in main text to “alleles (variants)” one paragraph explaining natural selection begins by stating that “new variants arise from a mutation” and concludes “the new allele will become common”</td>
<td>variant not in glossary (alleles defined as “most genes come in different versions called alleles”) “A change in the bases of a gene creates a genetic variant or mutation”; elsewhere, a spread entitled “Genetic variants and phenotypes” does not use the term variant other than in the title, includes e.g. “A change in a gene that creates a new allele is called a mutation”</td>
<td>variant not in glossary (alleles defined as “Different versions of the same gene”) “Different forms of a gene are called alleles (or variants)”, and “A genetic variant is a different version of an allele, which is caused by a change in the DNA”</td>
<td>genetic variant defined in glossary (“A different version of a gene, caused by a change (mutation) in the DNA”) uses “genetic variants” throughout the main text, often in place of “alleles” (e.g.: “The genetic variants an organism has make up its genotype”)</td>
</tr>
<tr>
<td>...discuss genome sequencing, or the field of genomics?</td>
<td>references to the Human Genome Project and ongoing projects to sequence human and other genomes ‘genomics’ not found in text</td>
<td>reference to the Human Genome Project ‘genomics’ not found in text</td>
<td>reference to the Human Genome Project ‘genomics’ not found in text</td>
<td>references ongoing projects to sequence human and other genomes “The study of the structure and function of genomes is called genomics. This is an exciting and fast-moving area of science.” “the science of genomics includes genome sequencing and bioinformatics” Links Mendel to genomics in a case study called “The grandfather of genomics”</td>
</tr>
</tbody>
</table>
Table 1 (continued): GCSE textbooks

<table>
<thead>
<tr>
<th>Does the book...</th>
<th>GCSE textbook</th>
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</thead>
<tbody>
<tr>
<td>...discuss the social, technological and ethical impacts of genomics?</td>
<td>genetic testing and DNA sequencing in the contexts of health and disease, DNA analysis to work out evolutionary relationships/classification, GM for industry and agriculture</td>
</tr>
<tr>
<td></td>
<td>DNA sequencing in the context of health and disease, DNA analysis to work out evolutionary relationships/classification, GM for industry and agriculture</td>
</tr>
<tr>
<td></td>
<td>human gene therapy, DNA sequencing in the context of health and disease, DNA analysis to work out evolutionary relationships/classification, GM for industry and agriculture</td>
</tr>
<tr>
<td></td>
<td>genetic testing and DNA sequencing in the contexts of health and disease, DNA analysis to work out evolutionary relationships/classification, GM for industry and agriculture</td>
</tr>
</tbody>
</table>
Does the book...

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</thead>
<tbody>
<tr>
<td>explain the difference between genes and the genome?</td>
<td>genome not in glossary</td>
<td>no glossary present</td>
<td>glossary and main text: genome &quot;is the entire genetic material of an organism&quot;</td>
<td>genome defined in glossary as &quot;all of the DNA that makes up the organism&quot; (our emphasis)</td>
<td>genome defined in glossary as &quot;all the DNA that makes up the organism&quot; (our emphasis)</td>
</tr>
<tr>
<td></td>
<td>the term genome not found until late in the text (chapter 8 of 9) where genome projects are discussed: the first occurrence found was &quot;a complete map of all the genetic material in an organism (the genome) is obtained&quot;</td>
<td>&quot;Together, all the genes in an individual (or species) are known as the genome.&quot; (our emphasis)</td>
<td>main text: genome comprises DNA in chromosome and plasmids (prokaryotes), chromosomes and mitochondria (animals) and chloroplasts (plants)</td>
<td>reference to eukaryotic genome including the DNA in the nucleus and the mitochondria</td>
<td>very few uses of the term genome in the main text (&quot;a mutation is a change of the nucleotide sequence of the genome&quot; is a rare example)</td>
</tr>
<tr>
<td></td>
<td>non-coding DNA not in glossary (introns “portions of DNA within a gene that do not code for a polypeptide. The introns are removed from pre-messenger RNA after transcription.”)</td>
<td>elsewhere: “the genome... is all the DNA containing a full set of genes” and elsewhere: “a genome is all the DNA of an organism”</td>
<td>non-coding DNA not in glossary (introns “large, non-coding regions of DNA that are removed before RNA is translated”)</td>
<td>non-coding DNA not in glossary (introns “regions of non-coding DNA or RNA”)</td>
<td>“mitochondrial DNA could be thought of as an extra chromosome in the human genome”</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>silent mutations “can occur in the non-coding regions of DNA (introns)”</td>
<td>non-coding DNA not in glossary (introns “portions of DNA within a gene that do not code for a sequence of amino acids within a polypeptide chain. These are removed from the mRNA after transcription”)</td>
</tr>
</tbody>
</table>
Table 2 (continued): AS/A level textbooks

<table>
<thead>
<tr>
<th>Does the book...</th>
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<th>AS/A level textbook</th>
<th>AS/A level textbook</th>
<th>AS/A level textbook</th>
</tr>
</thead>
<tbody>
<tr>
<td>...explain the difference between genes and the genome?</td>
<td>“Much of the DNA in eukaryotes does not code for polypeptides. For example, between genes there are non-coding sequences made up of multiple repeats of base sequences.”&lt;br&gt;“In humans it is thought that as few as 1.5% of genes may code for proteins”&lt;br&gt;exons and introns explained</td>
<td>“coding DNA probably makes up less than 2% of the human genome and non-coding DNA about 98%”&lt;br&gt;exons and introns explained</td>
<td>“Large parts of the DNA do not code for proteins. Scientists think the non-coding DNA sequences are very important – 98% of the human DNA is non-coding. They know they are involved in regulating the protein-coding sequences”&lt;br&gt;exons and introns explained</td>
<td>“your genes only make up about 2% of your total DNA. The large non-coding regions of DNA are removed from mRNA before it is translated”&lt;br&gt;exons and introns explained</td>
</tr>
</tbody>
</table>
Table 2 (continued): AS/A level textbooks

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</thead>
<tbody>
<tr>
<td>...make clear that most phenotypic features are affected by multiple regions of the genome, not just single genes?</td>
<td>“Most characteristics are influenced by more than one gene (polygenes)” regulations of the genome: gene expression, transcriptional factors (our emphasis), RNA splicing, the concept of the epigenome and epigenetics, epistasis</td>
<td>“Most human characteristics are inherited in a much more complex way [than monohybrid inheritance]... regulation of the genome: transcription factors, promoters, concept of the epigenome and epigenetics, the concept of an operon, RNA splicing</td>
<td>“most human traits are polygenic” regulation of the genome: gene expression, transcription factors, promoters and enhancers, RNA splicing, epigenetics, non-coding RNAs</td>
<td>regulation of the genome: transcription factors, promoters, the concept of an operon, epigenetics, RNA splicing</td>
<td>“Although the promoter is usually found next to the protein coding part of the DNA, there may be other sections of DNA that also help control when the gene is expressed that may be located thousands of nucleotides away from it.”</td>
</tr>
</tbody>
</table>

Table 2 (continued): AS/A level textbooks
A5 NOTES FROM REVIEW OF ENDORSED GCSE AND AS/A LEVEL TEXTBOOKS

Table 2 (continued): AS/A level textbooks

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</thead>
<tbody>
<tr>
<td>...make consistent use of modern terminology such as variant?</td>
<td>variant not in glossary (allele defined as “one of a number of alternative forms of a gene”) the term variant could not be found in the text “Any changes in the base sequence of a gene produces a new allele of that gene (=mutation)” discussion of population genetic diversity and selection in terms of alleles (not variants)</td>
<td>the term variant could not be found in the text discussion of population genetic diversity and selection in terms of alleles (not variants)</td>
<td>variant not in glossary (allele defined as “a version of a gene, a variant”) reference to identifying “alleles or gene variants” associated with disease “each gene exists in slightly different versions called alleles (variants)” discussion of population genetic diversity and selection in terms of alleles (not variants)</td>
<td>variant not in glossary (allele defined as “version of a gene”) main text: “For most genes there are a number of different possible alleles or variants” discussion of population genetic diversity and selection in terms of alleles (not variants)</td>
<td>variant not in glossary (allele defined as “a gene variant”) “Gene variants are a result of DNA mutations” “Variants of genes that benefit organisms are selected and their frequency in a population increases”, and “Natural selection acts to increase the frequency of beneficial alleles”</td>
</tr>
</tbody>
</table>
Table 2 (continued): AS/A level textbooks

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</thead>
<tbody>
<tr>
<td>...discuss genome sequencing, or the field of genomics?</td>
<td>just over two pages on 'genome projects'; ref. to HGP</td>
<td>references to the HGP, 1000 Genomes Project and HapMap</td>
<td>references to the HGP, 1000 and 10,000 Genomes Projects</td>
<td>references to the HGP, 10,000 and 100,000 Genomes Projects</td>
<td>brief mentions of sequencing and &quot;genome studies&quot;, but not explained</td>
</tr>
<tr>
<td></td>
<td>reference to bioinformatics as supporting DNA sequencing</td>
<td>DNA barcodes, DNA profiling, PCR</td>
<td>PCR, DNA sequencing, DNA profiling, DNA barcodes, and a double-page spread ‘Timeline of Genomics’</td>
<td>PCR, history of DNA sequencing, DNA profiling, DNA barcodes</td>
<td>DNA barcodes, PCR, DNA fingerprinting</td>
</tr>
<tr>
<td></td>
<td>ref to the proteome and “there is a human proteome project currently underway”</td>
<td>ref to “the studies of DNA (genomics) and proteins (proteomics)”</td>
<td>reference to bioinformatics as supporting DNA sequencing</td>
<td>reference to bioinformatics as supporting DNA sequencing</td>
<td>the term ‘genomics’ could not be found in the text</td>
</tr>
<tr>
<td></td>
<td>PCR, DNA fingerprinting</td>
<td>“the field of genetics that applies DNA sequencing methods and computational biology to analyse the structure and function of genomes is called genomics”</td>
<td>“the field of genetics that applies DNA sequencing methods and computational biology to analyse the structure and function of genomes is called genomics”</td>
<td>reference to proteomics</td>
<td></td>
</tr>
</tbody>
</table>
Table 2 (continued): AS/A level textbooks

<table>
<thead>
<tr>
<th>Does the book...</th>
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<th>AS/A level textbook</th>
<th>AS/A level textbook</th>
</tr>
</thead>
<tbody>
<tr>
<td>...discuss the social, technological and ethical impacts of genomics?</td>
<td>health &amp; disease management/personalised medicine, genetic testing, paternity testing, human gene therapy, forensics, transgenic organisms in industry and agriculture, bioremediation, DNA analysis to work out evolutionary relationships/classification</td>
<td>health &amp; disease management/personalised medicine, genetic testing, human gene therapy, forensics, paternity testing, transgenic organisms in industry and agriculture, DNA analysis to work out evolutionary relationships/classification</td>
<td>health &amp; disease management/personalised medicine, human gene therapy, forensics, paternity testing, transgenic organisms in industry and agriculture, DNA analysis to work out evolutionary relationships/classification</td>
<td>health &amp; disease management/personalised medicine, human gene therapy, forensics, paternity testing, transgenic organisms in industry and agriculture, DNA analysis to work out evolutionary relationships/classification</td>
</tr>
<tr>
<td>Resource title</td>
<td>Last updated</td>
<td>Author</td>
<td>Target age</td>
<td></td>
</tr>
<tr>
<td>----------------------------------------------------------</td>
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<td>---------------------------------------------</td>
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<td></td>
</tr>
<tr>
<td>Adverse drug reactions</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>BRAF: from gene to cancer therapy</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Build a bug</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Defeating the little dragon</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Direct-to-consumer genetic testing</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Direct-to-consumer testing</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>DNA Libraries: Subcloning</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>16+</td>
<td></td>
</tr>
<tr>
<td>DNA replication</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>16+</td>
<td></td>
</tr>
<tr>
<td>DNA sequencing</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Evolution of modern humans</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Evolution of the human brain</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>16+</td>
<td></td>
</tr>
<tr>
<td>Extracting DNA from fruit</td>
<td>2019</td>
<td>Wellcome Trust Sanger Institute</td>
<td>11+</td>
<td></td>
</tr>
<tr>
<td>From DNA to protein</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>From DNA to Protein (flash)</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Fruit flies in the laboratory</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Function Finders</td>
<td>2015</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Function Finders: BLAST!</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Genetic counselling</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Genome Generation</td>
<td>2016</td>
<td>Wellcome Trust Sanger Institute</td>
<td>14+</td>
<td></td>
</tr>
<tr>
<td>Genome-wide association studies</td>
<td>2016</td>
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