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Abstract

Developments in genomics have led educational researchers to advocate for more genomics in high school biology education and a concept context approach instead of more conventional teaching strategies. Besides changes in the curriculum this will require a change in the content knowledge of biology teachers. At this moment it is unclear whether biology teachers possess the required knowledge. Based on interviews with four experts, ten learning objectives have been developed. Starting from these learning objectives, a case based assessment has been developed that can be used to evaluate teachers' knowledge on genomics in relation to genetic testing. Besides producing learning objectives and a case this research has made a first assessment of the deficiencies in the knowledge of teachers. This knowledge can be used when designing new (post) teacher training activities. It will help teachers to be more confident when teaching about genetic testing.

Introduction

Developments in genomics research have led to an enormous increase in available genetic tests, since 2005 the number has been doubled to over 2500 ('GeneTests,' 2012). These tests are not solely offered by healthcare providers; more and more companies offer direct-to-consumer genetic tests (Borry, Howard, Sénécal, & Avard, 2010). Prospective genomics research will increase this number even more, thereby increasing the probability of citizens being confronted with them (Henneman, Timmermans, & Van der Wal, 2004). Although genetic testing by both healthcare providers and companies is expected to improve the quality of life, it will also bring personal and societal dilemmas. Scientific literacy is required for informed decision making on such dilemmas (Kolstø, 2001).

Science education is considered the way to contribute to scientific literacy or, in the case of genetics, genetic literacy taught in biology (Molster, Charles, Samanek, & O'Leary, 2009). Genetic literacy is defined as *'sufficient knowledge and appreciation of genetics principles to allow informed decision-making for personal well-being and effective participation in social decisions on genetic issues'* (Bowling et al., 2007; McInnerney, 2002). Thus, adequate genetic literacy should help to understand genomics and its applications in genetic testing. To ensure that the biology curriculum is effective in preparing students for future personal and societal dilemmas related to genetic testing, a change in the biology curriculum has been advised. An example of this is the advice to emphasize polygenetic disorders instead of single gene disorders (Dougherty, 2009). This change will also affect high school biology teachers (Van der Zande, Waarlo, Brekelmans, Akkerman, & Vermunt, 2010), because they will have to teach more complex concepts related to genomics.

Genomics research has also led to a change in the scientific knowledge on which genetic tests are based. The genome, for example, can no longer be described as an unchangeable chain of genes, but is considered a complex system that interacts in many different ways with the environment (Boerwinkel, Verhoeff, & Waarlo, 2008). Research has revealed that education often holds on to older conceptions than those used in scientific research. This might cause students trouble to understand genomics and its (future) applications, for example genetic testing (Gericke & Hagberg, 2007).

Although some suggestions have been made demonstrating the importance of genomics education and how genomics education should be designed (e.g. Boerwinkel et al., 2008; Dougherty, 2009), very little research has been done on biology teachers in relation with genomics. This research is necessary because the success of implementing genomics in biology education is largely dependent on biology teachers' capabilities and willingness (Hashweh, 1987). Also, there is a shift in teaching strategies to a concept-context approach, which will require additional (context) knowledge of teachers (Bennet, Lubben, & Hogharth, 2006; Sadler, 2009). Based on an exploration of the educational practice and the clinical genetic practices, Van der Zande et al. (2010) identified eleven concepts in the context of genetic testing that should ideally be in the biology curriculum. However, it remains unclear whether these concepts are a part of the teachers' content knowledge and whether possible shortcomings in this content knowledge can be solved by acquisition of new concepts, a conceptual change for some concepts or by restructuring of knowledge.

This study aims at the formulation of the necessary content knowledge and at the production of an adequate assessment tool for knowledge about the genome in relation to genetic testing. Using this assessment tool, a first assessment will be made of the content knowledge of biology teachers on genomics and genetic testing. Together these outcomes can be used to design new (post) teacher training activities that will enhance knowledge and confidence of biology teachers in teaching genomics in high school (Genomics in schools, 2012). Another point of relevance of this research is that the procedure followed in the research may provide a general applicable strategy to assess teacher content knowledge in context.

Theoretical background

Content knowledge

According to Shulman (1987) teachers should possess deep and flexible content knowledge about the concepts they teach. Content knowledge is defined as *'the amount and organization of knowledge per se in the mind of the teacher'* (Shulman, 1986, p. 9). This includes knowledge of concepts and various theories. Teachers that possess sufficient content knowledge are best fit to teach and help students in the most effective way. As Ball, Thames, and Phelps (2008) state: *'Teachers who do not themselves know a subject well are not likely to have the knowledge they need to help students learn this content.'* Next to content knowledge, teachers need pedagogical content knowledge (PCK); knowledge about how content and pedagogy should be combined effectively (Shulman, 1986). PCK includes

understanding why certain concepts are hard for students to learn or what the best way is to teach concepts.

The focus of this research is the content knowledge that teachers (should) possess about the genome in the context of genetic testing. To identify the concepts that should be part of this content knowledge, Van der Zande et al. (2010) conducted interviews with three groups of representatives of the clinical practices (patients, medical professionals and medical ethicists) and experienced teachers. This research revealed eleven important concepts that are not a part of the biology curriculum. These concepts include biological concepts (e.g. genetic polymorphism) as well as techniques (e.g. genome wide screening) and professional practices (e.g. genetic testing). Besides these eleven concepts, knowledge of ethical, legal, and social aspects (ELSA) are important as well as knowledge of the characteristics of the genetic test practice; uncertainty, complexity, probability, and morality (Van der Zande et al, 2010). These concepts, ELSA, and the characteristics will therefore be included in this research.

However, besides new concepts, new scientific knowledge might also have caused meanings of existing concepts to evolve. Therefore existing content knowledge of teachers could have become outdated. One of the concepts already present in the content knowledge of teachers, that illustrates these evolving concepts, is the concept of the gene. Over the past years, several authors have written about the historical development of this concept (e.g. Gericke & Hagberg, 2007; Portin, 2009). Gericke and Hagberg (2007) describe the development of the scientific conception of the gene from 1900 to 2007. In this period five distinct models of the gene are distinguished, from a Mendelian model to a modern model. For each model a clear overview of the discoveries that caused the scientific concept to evolve is provided. The changes in the scientific conception of the gene might also have caused a change in the scientific conception of the genome. Such changes in existing conceptions are defined as conceptual changes (Chi, Slotta, & De Leeuw, 1994). A conceptual change can only happen when a person already has conceptions about what is being taught (Posner, Strike, Hewson, & Gertzog, 1982).

According to Boerwinkel et al. (2008) changes in the scientific practice of genomics might also cause relationships between concepts to be altered or, in other words, lead to the restructuring of content knowledge. These changes have to do with new and more knowledge on complex relations between DNA, the environment, and traits. An example of changing relations is demonstrated by the shift from categorizing diseases in ‘non-genetic diseases’ versus diseases caused by a single gene mutation to describing most diseases as caused by multiple low-penetrance gene variants in combination with environmental influences (Balmain et al., 2003). This research will provide a tool for evaluating teachers’ content knowledge and might provide insight in the current content knowledge of teachers. This might reveal if content knowledge should be updated by teaching new concepts, a change in the meaning of concepts or restructuring of knowledge.

Concept-context approach

The concept-context approach, also called context-based approach (Bennet et al., 2006) or situated learning (Van der Zande et al., 2008), is an upcoming teaching strategy that is especially evident in secondary education. In this strategy the contexts and applications of science are used as a starting point for teaching about science instead of as an endpoint, which is a more conventional teaching strategy (Bennet et al., 2006). In science education the conventional teaching strategy is based on teaching students scientific knowledge that they should memorize for a test after which the knowledge is very often forgotten, while in professional science, the knowledge is used to reach a certain goal (Sadler, 2009). Students do not per se have to learn about professional science, they are confronted with various scientific contexts in their everyday life. These everyday scientific contexts could be used in science education. This will emphasize the relevance of taught scientific subjects and therefore students will become more motivated and interested. This is believed to improve students' affectivity with science as well as improve their learning outcomes. These benefits have been endorsed by the research of Bennet et al. (2006), in which the results of seventeen studies were compared.

The change of a conventional teaching approach to the concept-context approach does not only result in changes in textbooks and student learning activities. Teachers as well will have to adapt to this new approach. It is not sufficient to know the concepts and theories associated with a subject. Teachers, as well as the students, will have to be able to apply the concepts and theories in contexts: real world situations or science applications. Therefore, it is important also to have knowledge about the practices that use the specific knowledge. These practices are the contexts in which they have to apply the concepts and theories, for example the biological meaning of the concept gene in the context of genetic testing. This change in required knowledge also changes the way of assessing the knowledge of teachers about concepts and theories. This research will assess teachers' knowledge about the genome and genetic testing in a context presented as a case. This means that the teacher will be approached as a participant in an activity in which knowledge of genomics is involved: the teacher will be an expert on genetic testing who will give advice to entrepreneurs that are starting up a company that offers genetic tests.

Case-based teaching

Cases are stories about a specific subject that can be used as an illustration of more general theories; they add a context to the theory (Darling Hammond and Hammerness, 2002). Cases have proved to be an effective strategy for teaching purposes; and according to some researchers even better than conventional teaching methods like lectures and learning from books (Kim, Phillips, Pinsky, Brock, Phillips, and Keary, 2006). Therefore case-based teaching can today be found in many disciplines as for example medicine and law, but also in teacher education (Kim et al, 2006; Shulman, 1992). However, a variant of case-based teaching has been around much longer to educate people by story telling about moral or ethical principles, for example fables (Shulman, 1992). The benefit of using case-based teaching is that the theory comes alive and that they can add another layer to the theory. It could help learners to see the diversity, the difficulties, and prevents overgeneralization by only

learning theories (Shulman, 1992). At last, '*learners are challenged to analyse problems presented in cases, make inferences based on limited information, and make decisions on uncertain, ambiguous and conflicting issues that simulate a real-world, professional context*' (Kim et al. 2006, p867).

According to Shulman (1992), case construction is based on two steps. The first step is to determine the theoretical background of the subject; what is the theory that needs to be conveyed to the learner (the concepts). Only once the theoretical background has been determined the second step, selecting or developing an illustrative case (the context), can be carried out. Kim et al. (2006) have developed a conceptual framework specifically for developing teaching cases. By reviewing 100 studies they discovered seventeen strategies for making a good case. These seventeen strategies can be divided in five categories: relevant, realistic, engaging, challenging, and instructional. The relations between the categories and strategies are shown in figure 1. The above mentioned aspects will be taken into account during the development of the case.

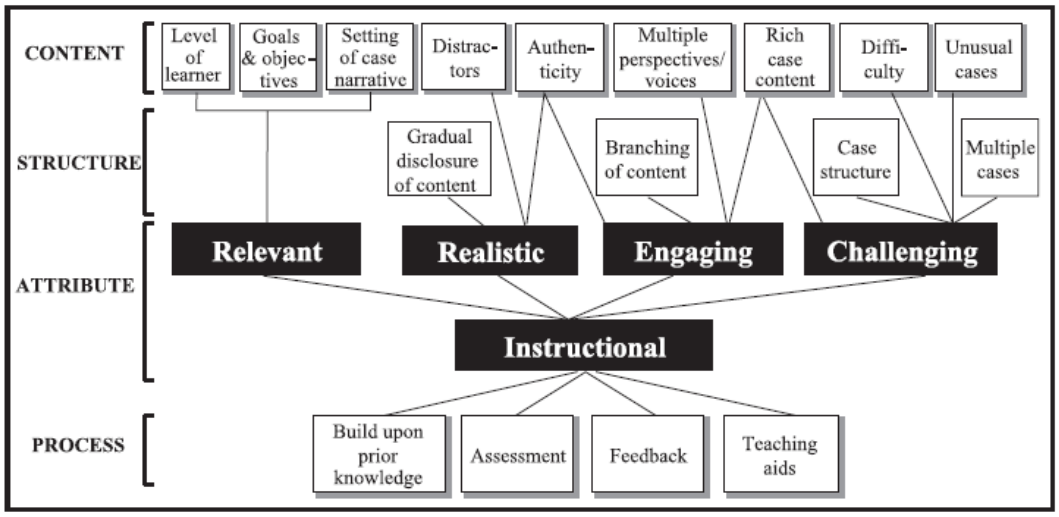

Figure 1. Conceptual framework of teaching case development (Kim et al., 2006)

Research aim and question

The aim of this research is to identify the deficiencies in biology teachers' content knowledge necessary to teach about genomics in relation to genetic testing by developing a tool that deals with the required content knowledge. The developed tool might later be used to evaluate teachers' prior knowledge before attending (post) teacher training and as a tool to teach about the content knowledge during training activities. Besides, knowledge about possible deficiencies identified by this research will provide information about concepts that should be part of (post) teacher training activities. This could be taken into account during designing these activities. Therefore the following main question is defined:

Which content knowledge related to the genome and genetic testing should be added or adjusted in (post) teacher training to create a better understanding of genetic testing in context?

To answer this question, the following sub questions are defined:

1. *Which conceptions of the genome and genetic testing are required to understand current and future practices in genetic testing?*
2. *Which conceptions of the genome and genetic testing do high school biology teachers use to understand genetic testing?*

The first sub question will lead to the development of a tool, which will be used to provide an answer to the second sub question. A comparison of the answers provided for both sub questions will be used to answer the main question.

Methods

To provide an answer to the main research question several steps where taken as described by the flowchart shown in figure 2. The framed steps indicate those steps that contain parts of the results. The various steps will be elucidated in the following section after discussing the participants.

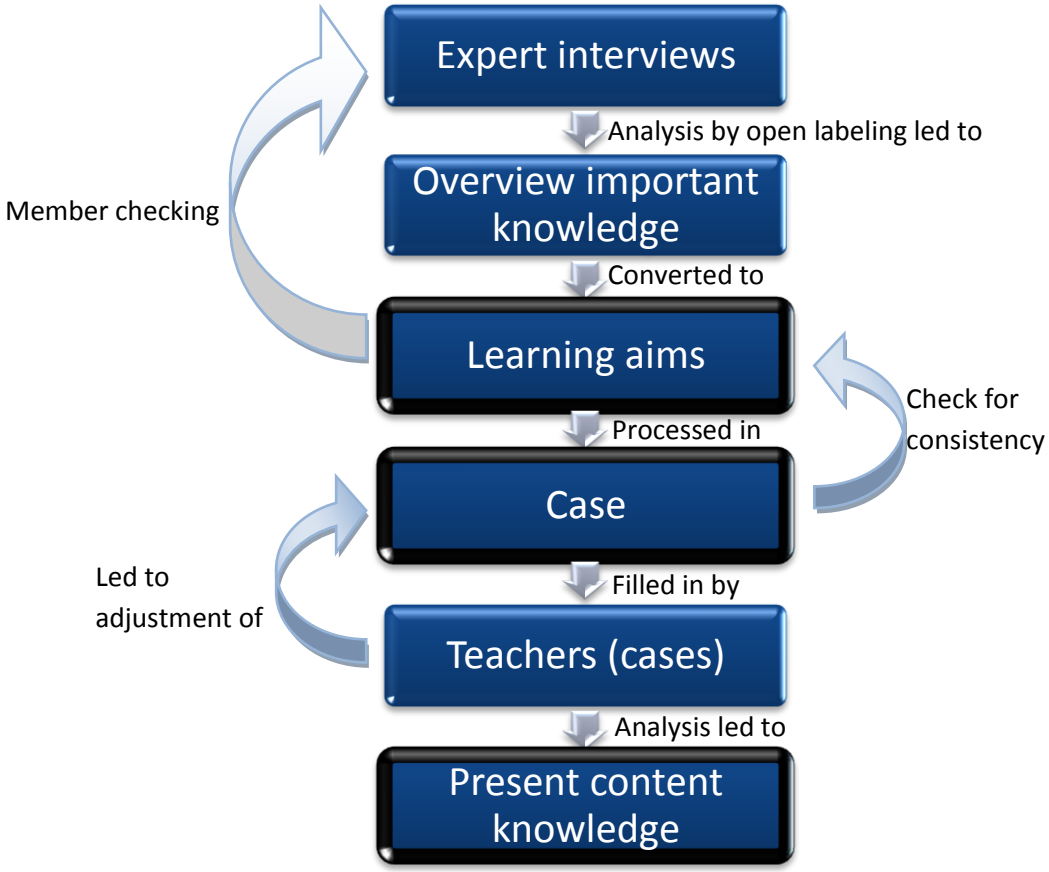

Figure 2. Flowchart of the used research

Participants

To determine the knowledge related to the genome important for understanding genetic testing, interviews were conducted with four experts. These experts were randomly selected from a list of experts that have connections with the genomics education research group in Utrecht. The sample included experts in various genetic testing fields: a researcher in occupational health & clinical genetics (section community genetics); a professor of translational epidemiology; a PhD researcher in genomics developments for primary care workers; and a professor of clinical genetics. All experts were female and associated to a different university in the Netherlands.

To make a first assessment of the content knowledge nine teachers were asked to fill in the case that was developed as a result of the expert interviews (as explained in the following section). The teachers were selected from several sources. These sources included both networks of teachers related to the university teacher training and teachers that reacted on an appeal placed on a website for biology teachers. All teachers have at least three years of experience, with an average teaching experience of 12 years. Three teachers had participated in additional courses about genomics besides biology lessons during university.

Instruments and data analysis

This section describes both the instruments used for the research as well as the analysis in a chronological order as described by the flowchart (figure 2). First, expert interviews were conducted to determine the required content knowledge of biology teachers. These interviews were used to develop the second research instrument, a case. The aim of the case was to evaluate the content knowledge of biology teachers.

Expert interviews

The experts in the field of genetic testing were interviewed using a semi-structured face-to-face interview. Three of four interviews were conducted in the office of the expert and one of the interviews was conducted in a public place, which caused some disturbance during the interview. The interviews lasted approximately one hour. During the first part of the interview experts were asked to adjust or complement a concept map about the genome, in a way that it would contain all the important concepts in relation to the genome for consumers to understand genetic testing. This concept map was a modified version from the neoclassical model of gene function provided by Gericke and Hagberg (2007). This specific model was chosen because it is the most commonly used model in high school biology textbooks. The modification included the addition of the concept genome and the concept phenotype (figure 3). Only one of the experts actually did make changes in the concept map, the other experts only talked about changes in the concept map. During the second part of the interview experts were interviewed on three topics:

- Changes in the field of genetic testing that they have experienced
- Their experiences with conceptual difficulties of people they have met (for example students during lessons or patients in practice)
- Their ideas about the importance of knowledge of the genome for understanding genetic testing.

The interviews were recorded and transcribed.

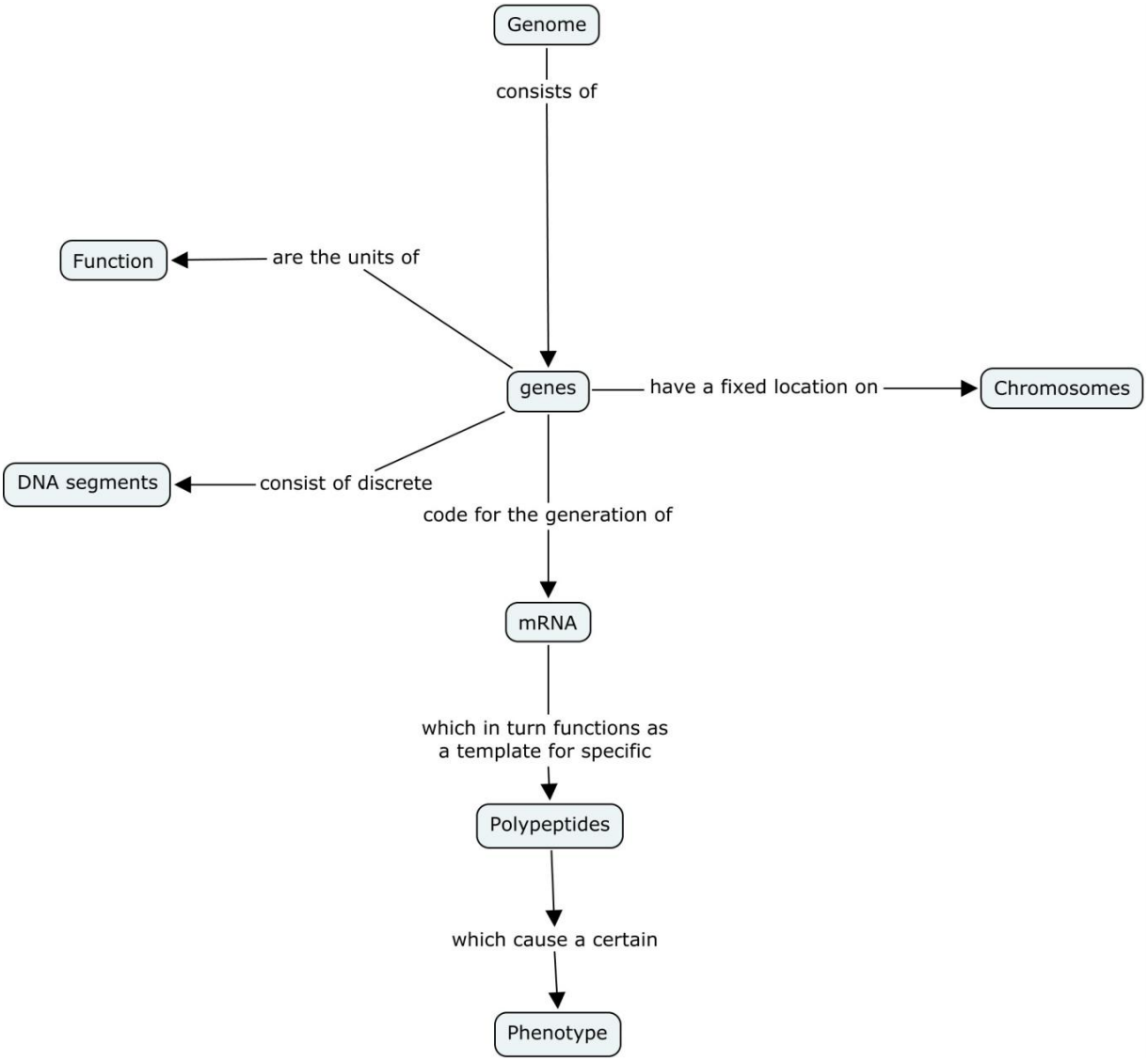

Figure 3. The modified neoclassical genetic model of Gericke and Hagberg (2007). The concepts genome and phenotype have been added.

After transcription the interviews have been analyzed by open labelling according to Baarda, de Goede, & Teunissen (2005) and Corbin & Strauss (1990). To check for inter-rater reliability another researcher labelled eleven interview fragments according to the designed labelling system (appendices). This led to the conclusion that the labelling of both researchers corresponded well with one another. Open labelling resulted in an overview of important content knowledge in relation to genetic testing, including: concepts and relations between concepts that experts directly mentioned as important; concepts and relations that were indirectly mentioned by the experts; the difficulties that the experts have faced during lectures or consultations; and the developments they have noticed in the field of genetic testing. By axial coding, relating codes to each other, the various labels were ordered and grouped into themes that were then grouped in five core themes (Baarda, de Goede, & Teunissen, 2005; Corbin & Strauss, 1990), namely:

- Who qualifies for a genetic test
- What is being tested
- The utility of a genetic test
- The outcome of a genetic test
- Inheritance

The themes in each core theme were then translated into learning objectives for biology teachers. To make sure that these learning objectives were consistent with the ideas of the interviewed experts, a member check was performed. This was done by sending the list of learning objectives to the experts. They were asked to appoint for each learning objective if they thought it was important, partially important, or unimportant. Besides they could add additional comments for each learning objective. The member check led to the adaptation of some learning objectives and the removal of learning objectives that were appointed as important by less than two experts. This resulted in the removal of two learning objectives. The residual learning objectives were mentioned to be important by at least two experts (an average of approximately three for each learning objective).

Case

To identify the deficiencies of biology teachers a case was designed. A case as tool has been chosen because cases can comprise all kinds of aspects related to one subject; their motivating nature; and their ability to prevent overgeneralization (Kim et al., 2006 and Shulman, 1992). The case is developed based on the two steps described by Shulman (1992). First, the required content knowledge, the concepts and relations important for understanding genetic testing, revealed by the expert interviews and the research of Van der Zande et al. (2010) were determined (the learning objectives). Second, based on the determined required content knowledge the case has been developed. The context of the case, giving advice to a company that is planning on offering genetic health screenings to consumers, has been derived from one of the interviews in which the interviewee used this true story as an example. This matches with the criterion of a case being realistic set by Kim et al. (2006). The relevance of the case is stressed in the introduction of the case in which the rapid development of genomics with the associated consequences is sketched (Kim et al.

2006). In the case the required content knowledge is projected in case related open questions. Every question can directly be related to a specific learning objective. Before the case was send to the teachers, it was send to two biology teachers as a pilot to verify the clarity of the questions and to make sure that the questions were proposed in such a way that they yield the correct response. Their reaction revealed that the case was challenging, mainly because of the difficulty of the questions (Kim et al. 2006). In addition to the pilot with teachers, the researcher went through the case with another researcher for an additional check. The final case can be found in the results (box 1).

The final case was sent to the nine teachers in the form of an online survey. The teachers were asked to fill in the case without using any additional information (e.g. internet or text books). In addition to the case, the teachers were asked to indicate for each learning objective if they thought they could manage, partly manage or not manage the specific learning objective. In general it took teachers about half an hour to fill in the online survey.

To determine the efficiency of the designed tool as well as to make a first assessment of biology teachers' knowledge of the genome and genetic testing, the teachers' answers to the case were collected and analyzed. Analysis was performed by determining for each question the aspects of the answer that were in common with the model answer as well as answer aspects that were not a part of the model answer. This led to a list of answers for each question. In this list was tallied how many times each aspect was mentioned by the teachers. This provided information about the parts of learning objectives that did not led to difficulties as well as to aspects that were not mentioned or were wrong, indicating that teachers have difficulties with these parts. The difficulties that are identified in this way provide an answer to the main research question. Besides information about the concepts teachers have difficulties with, the answers of the teachers provided information about the clarity of the case and its questions. This has led to the adaptation of a few questions.

Results

Results from expert interviews

The expert interviews were used to provide an answer to the first sub question: *'Which conceptions of the genome and genetic testing are required to understand current and future practices in genetic testing'*. As a result of the interviews a list of concepts and relations between concepts was constructed. The list consisted of concepts that belonged to genetic testing and concepts related to the genome. The latter were usually mentioned as essential concepts for understanding (a specific subject of) genetic testing.

'I think in a simple way you should know what genes are, as we sometimes explain: you have a cookbook that contains letters and those letters together form a gene and when one letter is not good, then you will get the disease in case of monogenic diseases.' (E4)

One expert illustrated this with being able to read a book. In order to read a book you first should be able to read at all. The same is true for understanding genetic testing; in order to understand genetic testing, you should first have some basic knowledge of the genome.

The concepts were either mentioned directly as being important or indirectly by using them to explain other important aspects. These concepts in combination with the concepts mentioned by Van der Zande et al. (2010) were used to create a concept map that provides an overview of the required content knowledge, containing concepts as well as relations (figure 4). Besides, the required content knowledge has been converted to learning objectives for biology teachers as described in the methods section (table 1). It should be kept in mind that some concepts mentioned in both the learning objectives and in the concept map are quite difficult, for example pleiotropy. Researchers did not mention that people should know this kind of concepts by heart, but that they should be aware of the existence of such difficulties. The learning objectives were converted into a case, which can be found in box 1.

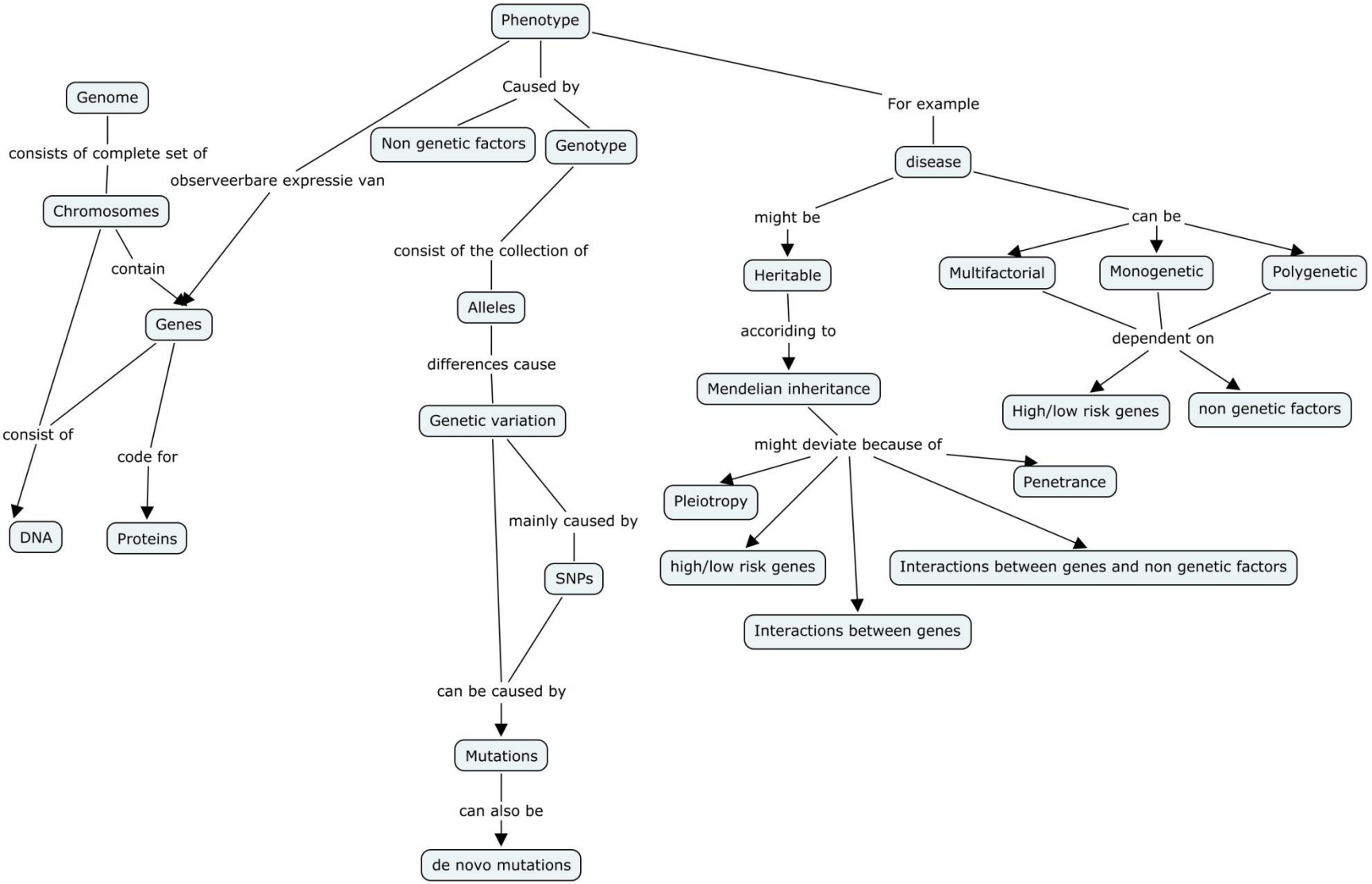

Figure 4. Concept map of the concepts and relations between concepts as mentioned during the expert interviews.

Table 1. The learning objectives derived from the expert interviews.

Subject	The teacher is able to:	Quotes from interviews
1. Who qualifies for a genetic test	1.1 Indicate that genetic tests can be carried out in persons with a disease history or a disease running in the family (genetic test), as well as in healthy persons (genetic screening).	*'What screening concerns, I think it is important that they (citizens) will notice the differences between a genetic screening and a genetic test, so I think that all teachers should be able to see the differences; that a screening is faster, cheaper, and less definitive and that a genetic test in a diagnostic way is more definitive.'* (E3)
	1.2 Indicate that genetic tests can take place in various life stages (preconception, pre-implantation, prenatal, and after birth/adults) and that, in the case of complex disorders, the timing of the test might be critical for the meaning of an outcome.	*'Yes, carrier testing, presymptomatic testing or predictive testing, and predisposition testing.'* (E3) *'And that (*100% penetrance*) happens often for rare syndromes and before a pregnancy you can test whether or not the both of you are a carrier. I think that is a kind of important because you might test everything preconception, but there is always a chance for de novo mutations. Those make that there still is a possibility that a child with a disorder will be born.'* (E2).
2. What is being tested	2.1 Indicate that genetic tests include all tests that can indicate genetic characteristics, thus the analysis of DNA, RNA, polypeptides, metabolites, phenotype, and family history.	*'Yes, those techniques are important. So that you explain that it is possible to look at metabolites, certain blood values as I already said, you can look at the phenotype, genotype, DNA, and at DNA level you can observe only one mutation, but you can also observe the whole genome.'* (E3) *'What I think is important teachers will see; I see genetic testing as something with a broad range, a very broad definition. Research of the phenotype for me is a genetic test, for example a nuchal scan for down syndrome.'* (E3)
	2.2 Indicate that disorders can be monogenic, polygenic or multifactorial and name the implications that this might have for a genetic test and a potential treatment for the disorder.	*'There exists something like high risk genes and low risk genes and ideas about a polygenetic model in which several low risk genes together with high risk genes* (function)*, that is something that I believe is even more important.'* (E3) *'If you can predict something very good or very bad depends on the heritability, so I think that is an important concept as well as the genetic complexity. When something is very hereditary and genetic simple because there is only one variant (…) than you will be able to predict with high certainty. When it has a low heritability than it will never*

		become a good test because all the other things (genes and non genetic factors) play a major role. However, even when it is very hereditary, but has so many genetic factors, than you will have very bad predictions because you will never be able to model it in such a way that you will get a good test.' (E2) *'The difference between complex and monogenic, although it runs into each other, is very important. On the moment clinical genetics is focused on monogenic disorders and than it is doable. However, general practitioners are being confronted with the direct to consumer tests. Those might tell someone that he has a higher risk for developing heart diseases.'* (E4)
	2.3 Indicate that a genetic screening (depending on the type of screening) in addition to the risks for certain disorders can inform persons about carrier status (and thus risks for newborns), the risk for relatives, and (positive or negative) reactions on medication.	*'Pharmacogenomics is something they should know about. That you could have different sensitivities for medicines.'* (E3)
3. The utility of a genetic test	3.1 Indicate that the usefulness of a test is determined by the technical quality, the clinical relevance, and the validity (sensitivity/false positive and specificity/false negative).	*'Those tests have advantages and disadvantages, specificity, and sensitivity. These are terms they should know as well.'* (E4) *'(*Shows a figure about the quality of a test*) This means that if you want to use it (a certain test) for the identification of people with an increased risk then it probably will be a very good test. But when you want to adjust a treatment that is very expensive or painful, than the test is not good enough.'* (E2) *'If it is about the development of a test, if you want to link the biology to practice and what you can do with it in practice, then there is one aspect that is always important to considerate, namely what are the action options.'* (E2)
4. The outcome of a genetic test	4.1 Indicate that the outcome of a genetic test for complex disorders will never be definitive because: - New research might reveal new insights about risk increasing or decreasing alleles; - Not all alleles are taken into consideration during the test (they may vary in different ethnical groups or might be protected by	*'(In response to a conversation of the expert with an entrepreneur who wants to start a genetic test company) That is what the man said; he came up with five genes for prostate cancer. But I said: do you know if there still is an association? He said why? I said well, science, knowledge changes. Something that was once a risk factor can later turn out to be nothing.'(*E2)

	patents); - Non-genetic factors and family history might influence the outcome.	*'What she (other researcher) probably said is that that kind of things (risks) may vary every day. If you do such a test today at 23andme, than you will get a certain profile and the day after tomorrow you will get a message that the risk of heart diseases has increased or decreased. That is hard to understand for people. Does the teacher need to be able to explain this? Yes, I think he does.'* (E4) *'For CF (Cystic Fibrosis) there are, I think, around 1600 known mutations and if you say that for example during the screening of a population only 32 mutations are under investigation, or 36, it is hard to explain. But that has to do with the contribution of certain mutations within a population, where you from. No, what I think is important, is that you can explain that certain diseases are here important while hemoglobinopathy is important in other … (countries).'* (E3)
	4.2 Indicate that the outcome of a genetic test has positive effects as early identification as well as potential negative effects as false concerns or problems for access to certain professions or insurances and that this might raise ethical, legal, and social questions.	*'I myself think that teachers should be capable of answering questions about the nature of a test, but if you really look at what the consumer should know than a lot of other aspects become important. (…) Uncertainty of information, the consequences that a test might have for the family, and risk communication.'* (E3) Interviewee: *'What I think is important is that also the ethics that is involved and what it means for a patient when your whole exome has been mapped and what it means for the family…'* Interviewer*: 'so the ethical, legal….'* Interviewee: *'the legal, yes the ELSA (ethical, legal, and social aspects).'* (E3) *'The recommendation of the referents to include the specific ELSA of genetic testing in biology education and their descriptions is helpful for describing the knowledge base for teachers who want to prepare students for the complexity of the testing situation.'* (Van der Zande et al., 2008, p. 24)
5. Inheritance	5.1 Indicate how Mendelian heredity works and explain that heredity may differ caused by the complexity of interactions of the	*'You have genes that have a role in gaining certain phenotypes, but not all genes have an equally strong effect. That part is missing completely in the scheme (concept map)*

	genome (incomplete penetrance, pleiotropy, influence of high/low risk genes, and interactions between genes and between genes and non genetic factors).	*and that is essential for if something can be a test yes or no.'* (E2) *'So if you would extend this to the consumers in general, than I can imagine that such a family tree is important for them to understand and therefore they need a basic understanding of inheritance.'* (E1) *'I think in a simple way you should know what genes are, as we sometimes explain: you have a cookbook that contains letters and those letters together form a gene and when one letter is not good, then you will get the disease in case of monogenic diseases.'* (E4)
	5.2 Name the differences between congenital, heritable, and genetic, give accompanying examples and indicate the implications for inheritance.	*'... heritable versus genetics. Well yes, that is all mixed up.'* (E2) *'People and doctors have difficulties with the difference between congenital disorders and heritable disorders and the fact that you can have things in your DNA at birth without them being expressed at birth.' (*E4)

Box 1. The developed case

'Genetics for You'

We live in an era of great developments in the field of genomics. These developments are reflected by the number of disorders that can be identified by genetic tests, more than 2500 (Genetests, 2012). At the same time people get more curious about what their genome might tell them about their (future) health. Various entrepreneurs respond to this trend by starting up companies that offer genetic tests or genetic screenings.

One of these companies is 'Genetics for You'. The founders of the company have little knowledge of genetic tests and are willing to learn more about the process and everything that comes with it. Therefore they asked you, as an expert on the field of genetic testing, to advice them. As an expert, you do not have to take into account things like feasibility and costs of the project.

General

The founders of the company have read that VU medical centre has started to offer genetic screenings to identify carriers of a Cystic Fibrosis allele. They are wondering whether genetic screening means the same as genetic testing.

1. a. How would you explain this to the founders?

Learning aim: 1.1
Model answer
A genetic test means to look at genetic markers that might be indication for a certain heritable disorder. Genetic testing includes genetic screening as well as diagnostic testing.
Genetic screening means the routine screening of healthy people for genetic markers.

b. Can you give another example of genetic screening?

Learning aim: 1.1
Model answer
A correct example of genetic screening. For example: - *Screening for Down syndrome* - *Cholesterol measurements* - *Guthrie test*

2. a. Can you explain the founders in which life stages of people, genetic tests and screenings can be applied?

Learning aim: 1.2
Model answer
Preconception (not for screening)
Pre implantation (not for screening)
Prenatal
After birth/adults

b. What kinds of decisions can be associated with testing in various life stages (assuming that the decision to test already has been made) ?

Learning aim: 1.2
Model answer
Preconception (not for screening): the decision to get a baby or to have pre implantation genetic diagnostics.
Pre implantation (not for screening): whether or not someone decides to implant an embryo.
Prenatal: to continue the pregnancy or to adjust the environment to the unborn child with a disorder.
After birth/adults: whether to get children or not or how you live your life.

What is being tested

The founders have noticed that there are many different genetic markers that can be used in order to define the risk for, or the presences of, a genetic disorder. Earlier they thought that only one genetic marker was being used to define risks, namely changes in the DNA (genotype).

3. Can you name the other genetic markers that can be used to define risks?

Learning aim: 2.1
Model answer
RNA
Polypeptides/proteins
Metabolites
Phenotype
Family history

As previously mentioned, for more than 2500 disorders genetic tests (screenings) are available. You want to make the founders understand that the role of genes in these disorders may differ and that these differences are important for the nature of the test, the meaning of the test results, and a possible treatment.

4. Explain to the founders the meaning of the concepts monogenetic, polygenetic, and multifactorial. Appoint at least the following aspects: the meaning itself, the severity and prevalence of the disorders that belong to the group, and the meaning of the test results.

Learning aim: 2.2	
Model answer	
Below	
Disorder	**Answer**
Monogenetic	*The answer must show that the teachers understands that:* - *Meaning: disorder is caused by changes in one gene* - *Severity: most of the time severe* - *Prevalence: rare* - *Test results: very reliable*
Polygenetic	*The answer must show that the teachers understands that:* - *Meaning: the disorder is caused by the interaction of multiple genes (containing changes)* - *Severity: is variable, but less severe than monogenetic disorders* - *Prevalence: not common* - *Test results: Not very reliable*
Multifactorial	*The answer must show that the teachers understands that:* - *Meaning: the disorder is caused by interactions between (multiple) genes and non genetic factors* - *Severity: most disorders are not severe, but there severe disorders exist* - *Prevalence: very common* - *Test results: unreliable*

5. a. Based on your previous answer, can you argue why monogenetic disorders should be part of the companies offer?

Learning aim: 2.2
Model answer
The disorders are severe
The test results are reliable

b. Based on your previous answer, can you argue why multifactorial disorders should be part of the companies offer?

Learning aim: 2.2
Model answer
High prevalence
Can usually be influenced by lifestyle changes.

'Genetics for You' decided to focus on genetic health screenings for persons. Therefore they will not offer tests that provide information about ancestry and paternity.

6. Indicate how the company could argue that the results of offered genetic screenings may contribute in various ways to an (potential) improvement of the health of the tested person.

Learning aim: 2.3
Model answer
Provides risks and responses to risks
Prescription of the correct medication (pharmacokinetics)
Carrier ship/risk for relatives

The utility of a genetic test

The founders of 'Genetics for You' can not just decide to start offering genetic screenings. The screenings have to be useful. The utility is determined by the technical quality, the clinical relevance, and the validity.

7. Can you explain these quality aspects to the founders?

Learning aim: 3.1
Model answer
Technical quality is determined by the quality of the test process, the actions, and processing; the test results should be reproducable
The clinical relevance depends on the action options that are offered by the outcome of a test.
The validity of a test depends on whether a test measures what should be measured (sensitivity/false positives and specificity/false negatives).

The outcome of a genetic test

In response to the submitted DNA sample 'Genetics for You' calculates the risks for several disorders. However, the calculated risks of complex disorders are not definitive and may change in time (the first day you have an risk of 0,1 for developing a heart disorder and a month later this risk has been increased to 1,2). 'Genetics for You' heard that often customers do not understand this and therefore they want to place information about this phenomenon on their website.

8. How could 'Genetics for You' explain the changing risks?

Learning aim: 4.1
Model answer
New research may yield new information relating to the degree of involvement of different genes in developing a disorder.
During risk calculation, non genetic factors and family history are not included.
A lot of different alleles might be known for one disorder, which are not all examined in the test.

9. Suppose 'Genetics for You' has the resources (time, money, and material) to do whatever they want within their screenings, what additional questions should they ask (the consumer) to predict their risks even better?

Learning aim: 4.1
Model answer
They should take into account non genetic factors (lifestyle, present phenotype)
They should take into account family history

'Genetics for You' is aware of the fact that genetic screenings do not solely have positive effects, but also potential negative effects. They realize that it is important to inform consumers about this. They just do not have a good notion about the potential negative effects.

10. Can you explain 'Genetics for You' the negative effects that a genetic screening might have for consumers?

Learning aim: 4.2
Model answer
Ethical aspects, such as: what to do with the results
Legal aspects, such as: insurance
Social aspects, such as: consequences of knowledge for the persons and his relatives

Inheritance

Thanks to genetic screening consumers might find out that they have a high risk for a certain disorder. In many people this raises questions about the possibility of passing on the disorder to their children. Initially the chance of passing on the disorder will be determined by Mendelian inheritance. Unfortunately, for complex disorders the chances determined by Medelian inheritance often do not match the actual phenotype.

11. Can you explain why inheritance sometimes differs (seems to differ) of the normal Mendelian inheritance?

Learning aim: 5.1
Model answer
Incomplete penetrance: the genetic abnormality is present, but this does not always lead to the specific phenotype.
Pleiotropy: genes might be part of multiple molecular pathways
Interactions between genes
Interactions between genes and non genetic factors
High/low risk genes

Within genetics there is a distinction between the concepts congenital, genetic, and heritable. These concepts are used interchangeably what might cause confusion. 'Genetics for You' might be confronted with these concepts when describing disorders. To avoid confusion and to make sure that within the organization everybody is talking about the same thing, 'Genetics for You' wants to place a description on their website.

12. Can you explain why congenital, genetic, and heritable disorders are not interchangeable?

Learning aim: 5.2
Model answer
A congenital disorder does not have to be genetic and therefore not heritable (it is possible). The disorder could be caused by events during pregnancy, for example an illness of the mother.
Heritable means that the disorder can be inherited of one or both parents and can be transferred to potential progeny. A heritable disorder therefore is always genetic as well as congenital.
A genetic disorder could be heritable or congenital but this does not have to be true. It could be caused by de novo mutations and can occur in each life stage. In this case the disorder is genetic but not heritable or congenital (however, a de novo mutation that happens during pregnancy might cause a child with a congenital disorder).

Final

As a result of your help 'Genetics for You' has a much better picture of the genetic testing or screening practice and the difficulties that come along with it. However, it is possible that certain aspects have not been addressed because they did not think of it.

13. The area below can be used for further comments, hints, and so forth.

Learning aim: not applicable
Model answer
Not applicable

Results from teachers

The cases filled in by the biology teachers were used to provide an answer to the second sub question: *Which conceptions of the genome and genetic testing do high school biology teachers use to understand genetic testing?* The answer to this question is defined by the outcome of the case filled in by the teachers. This part will provide a summary of the difficulties that teachers encounter for each learning objective.

> *1.1 Indicate that genetic tests can be carried out in persons with a disease history or a disease running in the family (genetic test), as well as in healthy persons (genetic screening).*

The case shows that the difference of genetic testing and genetic screening is not clear to all teachers. Some have notions about the fact that genetic screening is a part of genetic testing and about half of the teachers mention that testing is more specific than screening. However, none of the teachers mentions that testing is carried out in persons with a disease history or a disease running in the family and that screening is carried out in healthy persons. Although the difference between testing and screening was not fully understood, almost all teachers were able to mention one or more examples of genetic screening.

> *1.2 Indicate that genetic tests can take place in various life stages (preconception, pre-implantation, prenatal, and after birth/adults) and that, in the case of complex disorders, the timing of the test might be critical for the meaning of an outcome.*

The life stages preconception, prenatal, and after birth/adults were known to most teachers as well as most decisions that associated with testing in the various life stages. However, pre implantation was not mentioned by any of the teachers.

> *2.1 Indicate that genetic tests include all tests that can indicate genetic characteristics, thus the analysis of DNA, RNA, polypeptides, metabolites, phenotype, and family history.*

The answers show that most teachers associate genetic testing or screening with investigations of the DNA. Only two out of nine teachers could give other examples than markers based on DNA. Besides, some teachers talked about non genetic factors and lifestyle of relatives as predictive markers, which is not correct. The lack of knowledge in this learning objective was also demonstrated by the fact that only one teacher indicated to manage it; four teachers believed to partly manage and four believed not to manage it.

> *2.2 Indicate that disorders can be monogenic, polygenic or multifactorial and name the implications that this might have for a genetic test and a potential treatment for the disorder.*

Almost all teachers are able to correctly define the concepts monogenic, polygenic, and multifactorial. However, the implications of these concepts for a disease are not clear. Most teachers believe that monogenic disorders are easier to treat than polygenic or multifactorial disorders. Besides, some teachers seem to overvalue the use of treatments, mainly gene therapy. Finally, the answers seem to indicate that most teachers do not realize

that the prevalence of multifactorial (and polygenic) disorders is much higher than for monogenic disorders.

2.3 Indicate that a genetic screening (depending on the type of screening) in addition to the risks for certain disorders can inform persons about carrier status (and thus risks for newborns), the risk for relatives, and (positive or negative) reactions on medication.

Eight out of nine teachers indicate that they manage this learning objective. However, it is interesting to see that almost none of them mentioned the benefits of genetic screening for adjusting the right medication or the risks for family. Teachers do mention the risk for disorders and carrier status of the tested person.

3.1 Indicate that the usefulness of a test is determined by the technical quality, the clinical relevance, and the validity (sensitivity/false positive and specificity/false negative).

Only three teachers believe that they do manage the learning objective. This is reflected by the answers to the associated question. Least problems arise when defining the technical quality and the clinical relevance. Only four out of five teachers were able to give a (almost complete) description of validity.

4.1 Indicate that the outcome of a genetic test for complex disorders will never be definitive because: new research might reveal new insights about risk increasing or decreasing alleles; not all alleles are taken into consideration during the test (they may vary in different ethnical groups or might be protected by patents); non genetic factors and family history might influence the outcome.

Teachers are aware of the fact that risks might be altered by non-genetic factors or family history. However, most teachers do not mention new researches (seven out of nine) as well as that they do not mention that not all alleles are taken into consideration during the test.

4.2 Indicate that the outcome of a genetic test has positive effects as early identification as well as potential negative effects as false concerns or problems for access to certain professions or insurances and that this might raise ethical, legal, and social questions.

Seven out of nine teachers believe they manage this learning objective. However, the provided answers indicate that they are especially focused on potential psychological consequences of genetic testing and do not think of the consequences of ELSA when asked to name negative effects.

5.1 Indicate how Mendelian heredity works and explain that heredity may differ caused by the complexity of interactions of the genome (incomplete penetrance, pleiotropy, influence of high/low risk genes, and interactions between genes and between genes and non genetic factors).

Seven out of nine teachers indicate that they are partially aware of the aspects of this learning objective. Only two teachers indicated which part they did not manage, namely the concepts between brackets. This might be the same for the other teachers. The answers to

the question indicate that teachers are most conscious off the interactions between non-genetic factors and genes that make the heredity of multifactorial diseases complex; other aspects (incomplete penetrance, pleiotropy, high/low risk genes) are rarely mentioned.

5.2 Name the differences between congenital, heritable, and genetic, give accompanying examples and indicate the implications for inheritance.

Almost all teachers can provide a description of the individual concepts (congenital, heritable, and genetic). However not all of them clarified why these concepts cannot be used interchangeably. Especially the difference between congenital and genetic and/or heritable has not been described precisely. Together with three teachers that indicate only to partially manage the learning objective, this might indicate that this aspect needs some additional clarification.

Conclusion and discussion

Which content knowledge related to the genome and genetic testing should be added or adjusted in (post) teacher training to create a better understanding of genetic testing in context? To answer this question the required content knowledge has been obtained through interviews with experts in the field of genetic testing and is converted into ten learning objectives (table 1). This provided an answer to the first sub question: Which conceptions of the genome and genetic testing are required to understand current and future practices in genetic testing? The learning objectives consist of concepts as well as relations between concepts that citizens, and therefore in this research biology teachers, should posses for understanding genetic testing. A part of the required content knowledge that is found by this research corresponds to the findings of Van der Zande et al. (2010), for example concepts as high/low risk genes, multifactorial disorders, and polygenic disorders. These concepts are not yet a part of the biology curriculum, but might be incorporated in the future. The novelty in the results of this research compared with the research of Van der Zande et al. (2010) is that this research described the knowledge as learning aims instead of a list of concepts. This makes it easier to apply the knowledge in education. Interesting is another resemblance between the two researches. Both seem to add a great value to the characteristics of genetic testing. The experts interviewed for this research especially assigned a major role to complexity, in particular to the complexity of the genome. Although only four experts were part of the research, three of four interviewees emphasized the importance of a basic understanding of the complexity of the genome for a correct understanding of genetic testing. Although perhaps not immediately apparent, the characteristics can be found in virtually all the learning objectives, stressing their importance. However, the both researches also show differences, for example Van der Zande et al. (2010) stated that the required content knowledge should include single nucleotide polymorphism, risk assessment, and sequencing. This distinction in research outcome might be due to the difference in focus of the interviews. Where the research of Van der Zande et al. (2010) was aimed at (the context of) genetic testing, this research was aimed at knowledge of the genome in relation to genetic testing. Besides, in the research of Van der Zande et al. (2010) clients and medical ethicist were also involved in determining the

required content knowledge. In the end, it can be concluded that the combination of this research together with the research of Van der Zande et al. (2010) provide a good overview of the required content knowledge that can be used for future changes in the biology curriculum as well as for the development of (post) teacher training strategies.

The determined required content knowledge was used to create a case which, when filled in by biology teachers, provided insights in their current knowledge. This provided an answer to the second sub question: Which conceptions of the genome and genetic testing do high school biology teachers use to understand genetic testing? Before proceeding to the content knowledge of biology teachers, it is important to keep in mind that this research only did an initial assessment of teachers' content knowledge on genetic testing. However, this initial assessment already provides some insight into the current content knowledge and potential deficiencies of teachers. The answers of teachers to the case presented an overview of the current knowledge of teachers. This identified that almost all teachers had at least some knowledge about each learning objective; none of the subjects was completely new to them. Based on a comparison of the teacher answers compared with the model answers defined by the learning objectives the potential difficulties could be identified. The deficiencies identified by this comparison are summarized in table 2. This list is a first assessment and will probably be adjusted based on future research. Teachers often do not mention concepts, as for the various genetic markers, or give wrong explanations, as for the concept validity. Furthermore, there are a few learning objectives for which teachers indicate that they believe to manage them, while the answers to the case questions associated with the specific learning objective indicate that the teachers did not or partly manage the learning objective. An example of this is that eight out of nine teachers indicate that they know all consequences of genetic testing, but almost none actually mention the consequences of genetic testing for relatives and medication. The same applies to knowledge of ELSA, in the case most teachers mention solely psychological consequences of genetic screening, while seven out of nine indicate to manage the learning objective. Unfortunately this method cannot reveal the real reason for these differences. Two possible conclusions might be drawn from this observation. First it could be that teachers indeed do manage the learning objectives, but that they are unconscious of this knowledge at the moment they have to apply it in context. When in the end they are confronted with the knowledge they realize that they do know the aspects mentioned by the learning objective. Second, it could be that

Table 2 Possible knowledge deficiencies of biology teachers identified by this initial assessment.

- **Genetic testing versus genetic screening**
- **Pre implantation**
- **Genetic markers (except for DNA)**
- **Implications (severity, prevalence, and treatment) of a disease that is monogenic, polygenic or multifactorial**
- **Genetic screening informs about reactions to medication and risks for relatives**
- **Validity**
- **Uncertainty caused by new research and because not all alleles are taken into consideration**
- **ELSA**
- **Complexity of the interactions of the genome (except for interactions between genes and between genes and non genetic factors)**
- **Congenital versus genetic/heritable**

teachers emphasize some concepts above others and that these undervalued concepts should receive more attention, this could be an indication for the need of restructuring of knowledge concerning genetic testing. More research is needed to identify the deficiencies of biology teachers more precisely. In order to do this in more detail it should be considered to do interviews instead of a survey or to use interviews as an additional method. Furthermore, it could be wise to make a comparison between teachers whose knowledge will be tested in context and teachers whose knowledge will be tested without a context.

Many researchers have indicated that education should follow a more context-based approach (Bennet et al., 2006; Sadler, 2009). Besides the changes this will induce in the curriculum, it will also demand a change in the knowledge of teachers. Therefore the assessment of teachers' content knowledge should be adapted to this development. This study demonstrated that the strategy as described by the flowchart in figure 2 is effective in translating context based expert knowledge into context based knowledge assessment, Instead of testing the content knowledge of teachers by letting them explain concepts, this research tried to assess the knowledge in a context-based manner via a case. Based on the answers provided by the teachers it could be concluded that most of the questions were well understood and that the case is a good representation of the learning objectives. Although the answers of the teachers provided a good first impression, some drawbacks were also identified. The answers provided by teachers are mostly short and unclear and, as described before, some thoughts of teachers about managing the learning objectives did not correspond with the provided answers. This could be an indication of the fact that the questions do not stimulate teachers to fill in everything they know. On the other had, as described above, it could provide insight in the fact that teachers emphasise some concepts above others; a situation that not always will be desirable. The strategy used in this research is not bound to this topic. The strategy can easily be adjusted to other (school) topics for assessing teachers' content knowledge in context, which is desirable because the concept-context approach is not tied to biology education. Based on this research new (post) teacher training activities might be developed containing the concepts and relations between concepts as well ass the characteristics and ELSA of genetic testing. Resulting activities (for example the case presented in this research) will help teachers to obtain the knowledge they need and to adjust this knowledge in context in order to teach about all aspects of genetic testing and feel more comfortable teaching it. In the end it will all be in the interest of students, because genetic testing might become a significant issue in their future and they need to be prepared to live in a genomic world.

References

Baarda, D. B., De Goede, M. P. M., & Teunissen, J. (2005). *Basisboek kwalitatief onderzoek*. Groningen, Wolters-Noordhoff.

Ball, D. L., Thames, M. H., & Phelps, G. (2008). Content knowledge for teaching: What makes it special? *Journal of Teacher Education, 59* (5), 398-407.

Balmain, A., Gray, J., & Ponder, B. A. (2003). The genetics and genomics of cancer. *Nature Genetics, 33*, 238–244.

Bennett, J., Lubben, F., & Hogarth, S. (2007). Bringing science to life: A synthesis of the research evidence on the effects of STS and context based approaches to science teaching. *Science Education*, *91*, 347–370.

Boerwinkel, D. J., Verhoeff, R., & Waarlo, A. J. (2008*).* A framework for rethinking science curricula in the genomics-era. In D.J. Boerwinkel and A.J. Waarlo (eds), *Rethinking science curricula in the genomics era*, 13-24. Available from www.society-genomics.nl

Borry, P., Howard, H.C., Sénécal, K., Avard, D. (2010). Health-related direct-to-consumer genetic testing: a review of companies' policies with regard to genetic testing in minors. *Familial cancer, 9*, 51-59.

Bowling, B. V., Acra, E. E., Wang, L., Myers, M. F., Dean, G. E., Markle, G. C., Moskalik, C. L., Huether, C. A. (2008). Development and evaluation of a genetics literacy assessment instrument for undergraduates. *Genetics, 178*, 15-22.

Chi, M.T.H., Slotta, J.D., & de Leeuw, N. (1994). From things to processes: A theory of conceptual change for learning science concepts. *Learning and Instruction, 4*, 27-43.

Corbin, J., & Strauss, A. (1990) *Basics of Qualitative Research: Grounded Theory Procedures and Techniques*, Newbury Park, CA: Sage.

Darling-Hammond, L. & Hammerness, K. (2002). Toward a pedagogy of cases in teacher education. *Teaching Education, 13*(2), 125-135.

Dougherty, M. J. (2009). Closing the gap: Inverting the genetics curriculum to ensure an informed public. *American Journal of Human Genetics, 85*(10), 6-12.

GeneTests database. (2011). Retrieved April 10, 2012, retrieved from http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/labdirgrowth.shtml

Gericke, N.M. & Hagberg, M. (2007) Definition of historical models of gene function and their relation to students' understanding of genetics. *Science & Education, 16,* 849–881.

Hashweh, M.Z. (1987). Effects of subject matter knowledge in the teaching of biology and physics. *Teaching & teacher education, 3*(2), 109-120.

Henneman, L., Timmermans, D.R.M., Van der Wal, G. (2005). Public experiences, knowledge and expectations about medical genetics and the use of genetic information. *Community genetics, 7*, 33-43.

Kim, S., Phillips, W., Pinsky, L., Brock, D., Phillips, K., & Keary, J. (2006). A conceptual framework for developing teaching cases: A review and synthesis of the literature across disciplines. *Medical Education, 40*(9), 867–876.

Kolstø, S. D. (2001). Scientific literacy for citizenship: Tools for dealing with the science dimension of controversial socioscientific issues. *Science Education, 85*(3), 291–310.

McInnerney, J. D. (2002). Education in a genomic world. *Journal of Medicine and Philosophy, 27*(3), 369–390.

Molster, C., Charles,T., Samanek, H. & O'Leary, P. (2009). Australian Study on Public Knowledge of Human Genetics and Health. *Public Health Genomics, 12,* 84–91.

Portin, P. (2009). The elusive concept of the gene. *Hereditas 146,* 112–117.

Posner, G., Strike, K, Hewson, P., & Gertzog, W. (1982). Accommodation of a scientific conception: Toward a theory of conceptual change. *Science Education 66*(2), 211–227.

Sadler, T. D. (2009). Situated learning in science education: Socio-scientific issues as contexts for practice. *Studies in Science Education, 45*(1), 1–42.

Shulman, L. S. (1986). Those who understand: Knowledge growth in teaching. *Educational Researcher, 15*(2), 4–14.
Shulman, L. S. (1987). Knowledge and teaching: Foundations of the new reform. *Harvard Educational Review, 57*(1), 1–22.
Shulman, L. (1992). Toward a pedagogy of cases. In: Shulman, J. ed. *Case Method in Teacher Education* (pp. 1-30). New York: Teachers College Press.
Van der Zande, P., Waarlo, A.J., Brekelmans, M., Akkerman, S.F., & Vermunt, J. D. (2010). A Knowledge Base for Teaching Biology Situated in the Context of Genetic Testing. *International Journal of Science Education*, 1–31.

Appendices

Labelling system

Topic	Sub topic	label
1. Who qualifies for a genetic test	1.1 Distinction between tests	1.1.1. Genetic testing (diagnostic)
		1.1.2. Genetic screening
	1.2 Different life stages	1.2.1. Pre-implantation
		1.2.2. Prenatal
		1.2.3. Preconception
		1.2.4. After birth
2. What is being tested	2.1. Look at genetic markers	2.1.1. Genotype (DNA, RNA, gene, chromosomes)
		2.1.2. Phenotype (appaerence)
		2.1.3. Metabolites (polypeptides, enzyms etc.)
	2.2 Disorder background	2.2.1. Monogenetic
		2.2.2. Polygenetic
		2.2.3. Multifactorial
	2.3. Provides information about	2.3.1. Carrier status
		2.3.2. Disease risks
		2.3.3. Pharmacogenetics
		2.3.4. Ancestry
	2.4. Genetic markers	2.4.1. DNA
		2.4.2. RNA
		2.4.3. Polypeptides
		2.4.4. Metabolites
		2.4.5. Phenotype
3. The utility of a genetic test	3.1. Technical quality	3.1.1. Technical quality
	3.2. Klinical relevance	3.2.1. Treatable/treatments possible
		3.2.2. Prevention
	3.3. Validity	3.3.1. Sensitivity/False positives
		3.3.2. Specificity/False negatives
	3.4. Improvements	3.4.1. Higher heredity
		3.4.2. Less complex
		3.4.3. Higher prevalence
4. The outcome of a genetic test	4.1. Not definitive	4.1.1. New research
		4.1.2. Not all allele variants are taken into considerations
		4.1.3. Differences between ethnic groups
		4.1.4. Non genetic factors
		4.1.5. Family history
	4.2. Consequences	4.2.1. Ethical
		4.2.2. Legal
		4.2.3. Social
	4.3 Interpreting risks	4.3.1. Interpreting risks
5. Inheritance	5.1. Mendelian inheritance	5.1.1. Mendelian inheritance
	5.2. Differ from Mendelian inheritance	5.2.1. Penetrance
		5.2.2. Pleiotropy
		5.2.3. Interactions between genes
		5.2.4. Interactions between genes and non genetic factors
		5.2.5. High/low risk genes
	5.3. Confusing concepts	5.3.1. Genetic
		5.3.2. Congenital (de novo)
		5.3.3. Heritable