

# Research Paper

Which meaning do students, with knowledge of genetics on upper secondary school biology level, attribute to the concept ‘hereditary trait’?

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Date: January 2016

Intended journal: Journal of Biological Education



## Abstract

The field of genetics education is well researched as nowadays genetic literacy is found very important for the general public in order to be able to make well-informed decisions concerning genetics-related issues. Although several genetic concepts are thoroughly researched, few is published on the concept 'hereditary trait'. This research studies students' understanding of this concept. A questionnaire is held under 149 students with knowledge of genetics on upper secondary school biology level, in which students (1) gave a definition of the concept 'hereditary trait', (2) gave 5 examples of hereditary traits, (3) classified a list of traits as hereditary (yes/no/maybe) and (4) gave an explanation to their classification. It became clear that students have a limited view on the concept. Students mentioned for the most part only traditional examples of traits which are visible on the outside, independent from environmental factors, have a high variance within a population and do not change over time, such as: eye colour, skin colour and hair shape. They do recognize that hereditary traits are inherited or passed on from parents to their offspring and that they are coded in DNA/genes/chromosomes, but many students do not consider traits as hereditary which are also (partly) influenced by environmental factors, are not visible on the outside of an organism, with no variance within a population, are dynamical (change over time) and are on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers). Criteria to exclude these types of traits were mainly: if there is no variation within a population, if they are (partly) influenced by environmental factors or if it is caused by a mutation. Furthermore, a few misconceptions became clear. The results have implications for genetics education.

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## Introduction

Science is not only part of the school curriculum in order to prepare students for further careers in science, it is also found important that the general public has some familiarity with science. Therefore, in the late 1950's the term scientific literacy was introduced (DeBoer, 2000). Since the introduction of the term scientific literacy, many different meanings are given to this concept but there is still no consensus of its exact definition (Popli, 1999; Norris & Philips, 2003). It is probably best described as “what the public should know about science in order to live more effectively with respect to the natural world” (DeBoer, 2000). Considering that developments in science and technology can generate issues that affect society (i.e. socio-scientific issues), citizens need to be prepared to make well-informed decisions on these socio-scientific issues (Sadler, 2004). Hence, scientific literacy is found an important part of science education and much research has been done on how to effectively educate students to deal with such socio-scientific issues (Ratcliffe & Grace, 2003; Lewis & Leach, 2006).

The field of genetics in particular has evolved rapidly in the past decades, especially after the sequencing of the human genome. Research in human biology and medicine has been brought to a whole new level and numerous possibilities have been predicted and already used nowadays, such as: localized medicine, individualized preventive medicine, gene therapy and predictive genetic tests (Collins & McKusick, 2001; Subramanian *et al*, 2001; Williams, 2001). These emerging applications also bring along concerns and dilemma's (medical, ethical, social) creating the need for genetic literacy among the public. Citizens need to be prepared to make well-informed decisions concerning genetics (Jennings, 2004; Bowling *et al*, 2008; Shaw *et al*, 2008).

The establishment of genetic literacy in the general public starts with education. Therefore, genetics is an inherent part of the biology curriculum. The main interrelated concepts which are emphasised in secondary education are heredity, gene, environment, phenotype and hereditary traits (College voor examens 2013a, 2013b & 2014). As it is important that these concepts are fully understood in order to achieve genetic literacy, much research has already been done on students' understanding or conceptual status on these concepts (Wood-Robinson, 1994 & 1995; Venville & Treagust, 1998; Hickey & Kindfield, 1999; Lewis, Leach & Wood-Robinson, 2000a,b,c; Lewis & Wood-Robinson, 2000; Marbach-Ad & Stavy, 2000; Wood-Robinson *et al*, 2000; Marbach-Ad, 2001; Lewis & Kattmann, 2004; Duncan & Reiser, 2007; Tsui & Treagust, 2007 & 2010; Boujemaa *et al*, 2010) and in the general public (Lanie *et al*, 2004). These studies show that these concepts are often still not fully understood

and that misconceptions occur, resulting in implications for education. Several assessment tools are developed in order to measure the understanding of genetics, such as the Genetics Concept Assessment (Smith *et al.*, 2008) and the Genetics Literacy Assessment Instrument (Bowling *et al.*, 2008) in order to improve education as it can be used as a pre- and post-test.

Although the field of genetics education is thoroughly investigated, research mainly focussed on students' understanding of specific concepts underlying transcription and translation. Yet, no research has been done specifically on students' understanding of concepts that emphasize the effects of protein synthesis, such as: phenotype, genetic traits or hereditary traits. Research on students' understanding on the latter aspects is especially important because Duncan & Reiser (2007) already mentioned that focussing on the mechanisms of translation might lead students' astray from the general principle of the relation between genes and protein-function. Moreover, they showed that students do understand that proteins are important elements in the body (enzymes and building of muscle, nails and hair), yet lack to understand that proteins also have a role in genetic phenomena, such as: signalling, regulation, transport, etc. Therefore, it is important to investigate students' understanding of concepts concerning the effect of genes, i.e. genetic/hereditary traits or phenotype.

This study focusses on students' understanding of the concept 'hereditary trait'. The scientific meaning of the concept hereditary trait has changed over time and there is still no clear definition of this concept. As the field of genetics initially started by research on hereditary traits that showed visible differences on the outside of an organism, such as Mendel's study on peas, hereditary traits were first seen as simple, visible and monogenetic characteristics. It was only until the field of molecular biology emerged and more research could be done on the underlying mechanisms, that it became clear that the relation between genes and their environment is very complex resulting in more phenomena to be seen as hereditary traits. The aim of this study is to investigate to what extent these types of phenomena are recognized and seen as hereditary traits by secondary school students, who have been educated on this subject in upper secondary biology classes. This results in the following research question: *Which meaning do students, with knowledge of genetics on upper secondary school biology level, attribute to the concept 'hereditary trait'?* To answer this question, four sub questions are formulated:

1. *What definition give students to the concept 'hereditary trait'?*
2. *What kind of examples of hereditary traits do students come up with?*
3. *Which traits are classified by students as hereditary?*
4. *What type of arguments do students use to classify a trait as not hereditary?*

The results can inform a revision of the genetics curriculum in secondary education.

## Theoretical background

### A brief history of genetics

Since ancient times the observation that certain traits are passed on from parents to their offspring is used in order to improve plants and animal species through selective breeding. The modern field of genetics, which studies the underlying process of heredity, probably started in the mid-19<sup>th</sup> century. Charles Darwin (1859) was the first to describe extensively how there must be an underlying mechanism which results in the diversity within and among species. Thereafter, Gregor Mendel (1866) discovered with his research on peas that some characteristics were transferred from generation to generation, independently from each other and through a fairly simple mathematical way. Mendel is sometimes called the ‘Father of genetics’ as his model forms the basis to understand how characteristics are passed on through generations. Hence, the field of genetics started by studying hereditary traits, the description of hereditary traits was there long before the description of genes. As it only contained visible heredity, only hereditary traits were described in which there were clearly visible differences on the outside of organisms.

Improvements of microscopes and colouring methods made it possible to study the molecular basis of heredity. Already in the late 19<sup>th</sup> century it was found that genetic information is passed on by the nucleus of cells and this was followed by a lot of research on chromosomes, mitosis and meiosis. In the beginning of the 20<sup>th</sup> century it became clear that the ‘hereditary factors’ that Mendel described previously were linked to the chromosomes. However, it was still unknown from which chemical compounds hereditary traits consists of and how they were translated into phenotype. The discovery of the chemical structure of DNA by Watson and Crick in the mid-20<sup>th</sup> century was a big breakthrough in genetics as scientists were now able to perform research on identification, function and regulation of genes; the field of molecular biology arose. Shortly after, the ‘central dogma of molecular biology’ was formulated, when it was discovered that DNA consists of a code which is transcribed and translated to result in certain characteristics.

Since then, tremendous progress is made in this new field of molecular biology in the late 20<sup>th</sup> century. For example, recombinant DNA technologies made it possible to produce human-insulin by bacteria and tests for certain diseases such as sickle-cell disease and cystic fibrosis were developed. Moreover, the rapidly developing field of bioinformatics and sequence technologies made it possible to process a lot of data resulting in the finishing of the ‘Human Genome Project’ in 2000, even earlier than predicted. The complete human genome was sequenced and made publicly available online. This made it possible to also search for

genetic components of traits and disorders for which was no evidence of a relation at first. It became clear that the relation between genes and traits is very complex in most cases.

In the last decade it became more clear how internal and external factors influence the regulation of genes and therefore influence the phenotype. This influence on gene-expression is also passed on to new cells during growth and development and sometimes even to the next generation: the new field of epigenetics emerged.

The meaning of the concept ‘hereditary trait’ has shifted from a simple, visible and monogenetic characteristic to a very complex relation between genes, the environment and traits. As stated in the introduction, nowadays it is important for the general public to have some understanding of genetic concepts as the still rapidly developing field of genetics not only bring numerous possibilities, but also concerns. People should be able to make well-informed decisions on topics which concerns genetics. As genetic literacy among citizens starts with education, it is relevant to study genetics education as part of the biology curriculum in secondary education.

### Problems in genetics education

As it is clear how important it is for the general public to have some understanding of genetics and that genetic literacy among the public starts with education, much research has already been done on genetics education. However, it is still considered as one of the most challenging subjects in biology education. Knippels (2002) divided the main domain-specific difficulties into five categories:

- (1) *Domain specific vocabulary and terminology.*
- (2) *Mathematical content of genetic tasks.*
- (3) *Cytological processes of cell division, which mainly relates to chromosome structure and its processes.*
- (4) *Abstract nature due to the sequencing of the biology curriculum, which separate mainly meiosis and genetics.*
- (5) *The complex nature of genetics: a macro-micro-problem, how to relate concepts and processes from different systematic levels.*

Problems in these categories are not isolated but are all related to one another which contribute to difficulties in students’ understanding of genetics. It is clear that still a lot of misconceptions occur among students’ understanding of genetics (Cho, Kahle, Nordland, 1985; Browning & Lehman, 1988; Lawson & Thompson, 1988; Shaw, *et al*, 2008) which means that genetics education is not adequate.

Gericke *et al* (2007, 2009, 2010, 2014) extensively investigated science versus school-science. These studies revealed that a lot of incoherence occurs in school textbooks which may contribute to the lack of understanding of genetics in students. Moreover, the study of

Draanen (2015) also compared science to school science by investigating Dutch biology schoolbooks, specifically about the concept ‘phenotype’ and ‘hereditary traits’. He also showed incoherence in these textbooks. As most teachers use school textbooks as a guide to teach the different subjects and students use them to learn the different subjects, incoherence in school textbooks might be of great influence on students’ understanding.

Poor teaching of genetics causes poor understanding of genetics by students, i.e. poor scientific literacy. Dougherty (2009) states:

*‘Taken together, current teaching practices may be producing a public that is unprepared to participate effectively as medical consumers in a world where personalized medicine will rely increasingly on genetic testing, risk assessment, predispositions, and ranges of treatment options that include biological and behavioural components.’*

Therefore, it is important that more research is done on students’ understanding of genetics in order to improve genetics education.

### Potential differences between the scientific meaning of the concept ‘hereditary trait’ and students’ understanding of the concept

On the basis of scientific research, assumptions can be made on what points education and science are likely to differ from each other in the description of what is considered as a hereditary trait (Draanen, 2015). These following six categories were used to investigate students’ understanding of the concept ‘hereditary trait’.

#### 1. Clearly visible traits versus not-visible traits

Secondary school students seem to restrict themselves to outer visible or physical traits (Thomas, 2000; Wood-Robinson, Lewis, & Leach, 2000; Venville & Treagust, 1998) while in science, also internal traits and traits invisible for the naked eye (Bearden & Freimer, 2006) are mentioned, for example the production of insulin and serotonin (Wojczynski & Tiwari, 2008), the colour of the peritoneum of lizards (Porter, 1967) and chemical signals of flowers (Van Doorn, 1997).

#### 2. Traits on organism level versus traits on lower organisation levels

Scientists describe hereditary traits on different organizational levels: on molecular level (proteins, hormones, enzymes) and on cellular-, tissue-, organ- and organism level (Roseman et al., 2006; Dupré, 2008; Wojczynski & Tiwari, 2008; Duncan et al., 2009). Sociobiologists add psychological and behavioural levels to these (Machalek and Martin, 2010; Goldberg, 1993; Strickberger in Churchill, 1974). Cystic fibrosis, for example, has features on tissue-,

organ- and organism level (Dupré, 2008; Duncan, Rogat, & Yarden, 2009; Duncan & Reiser, 2007). Secondary school students, in contrast, do not seem to realize that traits do have features on other organizational levels than the organismal level (Duncan & Reiser, 2007).

3. Traits which are independent from environmental factors versus traits which are heavily influenced by the environment

The use of the terms ‘hereditary traits’ and ‘phenotype’ also differs for phenomena heavily influenced by environmental influences. Jiménez-Aleixandre (2014) mentions chance, gene regulation, cell environment, body environment (e.g., hormones) and external environment (e.g., diet, health care) as examples of such environmental factors jointly responsible for phenotypes, while Dupré (2012) adds care, training, knowledge to this list of environmental factors. Beside genetic and environmental influences, epigenetic factors influence hereditary traits (Gericke, Hagberg, dos Santos, Joaquim & El-Hani, 2014; Meijer, Bomfim, El-Hani, 2013). Students consistently distinguish physical traits, which they regard as hereditary, and character traits like temperament, which they attribute to environmental factors (Thomas, 2000). Further, it is well possible that they do not see the role of epigenetics in the development of traits because little information about epigenetics is available to them (Billingsley & Carlson, 2010). However, twin studies in science show that an important part of the variation in traits like intelligence and extraversion is explained by genetic variation and they can therefore be regarded as hereditary traits (Plomin, Owen, & McGuffin, 1994). Also sportsmanship and musicality are hereditary traits which are established by an interaction between genes and environmental factors. Even apparently obvious genetically determined traits like body height and facial appearance are partly determined by environmental factors (Thomas, 2000).

4. Traits with a high variance within a population versus traits with no variance within a population

Secondary school students are likely to regard traits which are universal in a population and which do not have variants or different alleles not as a hereditary trait. Examples of such traits are the manufacture of haemoglobin and the development of nerves, tissue and hormones (Bodmer, 1997). Though much research focuses on traits which vary within a population, for example on diseases, Mendelian traits or crop size (Dupré, 2012), there is also research to traits which do not vary within a population, like the embryonic development of the chicken (Hamburger & Hamilton, 1951). Mendelian traits constitute an important part of the genetics curriculum (Venville & Treagust, 1998; Shaw, Van Horne, Zhang, & Boughman,

2008; Gericke *et al.*, 2014) which could cause underexposure to non-variable (non-Mendelian) traits as examples of hereditary traits.

#### 5. Static traits versus dynamical traits (which change over time)

A hereditary trait can change during the lifetime of an organism. Examples of this are the hereditary traits lactose intolerance which can disappear on higher age or diabetes type II which appears on later age, but also hair colour and eye colour; these can turn lighter or darker during the lifetime of an individual (Christensen, 2000; Imesch, Wallow, & Albert, 1997). Some hereditary traits change overnight while others remain constant for a longer time (Lewis & John in Lenartowicz, 1975). However, it is well possible that students do not always recognize traits which can change during the lifetime of an individual (dynamic traits) as a hereditary trait. They are more likely to think of outer traits which they possess themselves as examples of hereditary traits. Further, as traits make up one's phenotype, it is well possible that students do not regard the phenotype as changing, though the phenotype in fact changes from day to day (Sinott in Lenartowicz, 1975).

#### 6. Traits on organism level versus traits on lower organisational levels which influence the chance of or sensitivity to certain traits or diseases (biomarkers)

Another aspect in which the content of the concept 'hereditary traits' or 'phenotype' can differ between science and education is the whether the chance of getting a disease or other trait is regarded as hereditary trait or not. A mutated gene often increases this chance, but it depends on many factors whether the phenotype occurs indeed. In other words, a problem can be present on only one, non-outer organizational level, which makes it possible to measure whether problems can arise on other organizational levels. The chance of getting a disease is therefore sometimes measured by looking at increased or decreased levels of biomarkers like iron levels in the blood. A biomarker which is known to be at least partly hereditary is called an endophenotype (Wojczynski & Tiwari, 2008). Gottesman & Gould (2003) state that an endophenotype can be neurophysiological, biochemical, endocrinological, neuroanatomical or cognitive. According to Nachtomy (2007), even the mRNA level of the BRCA-1 gene involved in breast cancer can be regarded as a phenotype. As students often receive limited examples of hereditary traits and as it is likely that they do not learn about biomarkers, it is well possible that they do not regard traits for which susceptibility can be measured with endophenotypes or biological markers as hereditary traits.

## Students' understanding of genetic concepts

Much research has already been done on students' conceptual status or understanding of several genetic concepts, however not specifically on the concept 'hereditary trait'. Nevertheless, the same methodology could be used in order to assess students' understanding of this specific concept. Therefore, an overview on literature on students' understanding or conceptual status on genetics/genomics/heredity/genes and the methods that were used is given.

Some studies examined the change in students' conceptions after a genetics course (Venville & Treagust, 1998; Duncan & Reiser, 2007) or certain activities such as the use of GenScope™/BioLogica™ (Hickey & Kindfield, 1999; Tsui & Treagust, 2003, 2007 and 2010). A part of the data on students' understanding of several genetic concepts were gathered from the worksheets and online tools which were used during the learning activities. Though, to gain more insight in students' reasoning also open-ended questionnaires and interviews were used.

Other research did not involve learning activities, but assessed students' understanding of genetic concepts near the end of their compulsory science education, (Lewis, Leach & Wood-Robinson, 2000a,b,c; Lewis & Wood-Robinson, 2000; Marbach-Ad & Stavy, 2000 and Lewis & Kattmann, 2004), as will be done in this research. Here also written questions, including open-ended and fixed questions, interviews and discussions were used.

Moreover, in order to test students' understanding of genetics, several assessment tools were made, such as the Genetics Concept Assessment (GCA) (Smith, Wood & Knight, 2008) and the Genetics Literacy Assessment Instrument (GLAI) (Bowling *et al*, 2008). These are validated questionnaires using questions with fixed answers.

Furthermore, students' conceptions about the concept of gene were investigated in University students (Boujemaa *et al*, 2010) making use of a questionnaire consisting open-ended questions. And the public understanding of basic genetic concepts was investigated (Lanie *et al*, 2004), which used in-depth telephone interviews.

As most research on students' understanding of genetic concepts made use of questionnaires including open-ended and fixed questions and several in-depth interviews, the same approach is used in this study.

## Methodology

This study investigates which meaning students, who have been educated on genetics in upper secondary biology classes, attribute to the concept ‘hereditary trait’. To investigate students’ understanding quantitatively most researchers made use of written questionnaires with fixed questions (see Theoretical Background, ‘Students’ understanding of genetic concepts’). However, in order to get more insight in students’ reasoning also open-ended questions or the possibility to explain were added and interviews and discussions were held. Moreover, in most cases the questions included a certain example or introduction about the specific concept upfront the question to measure understanding rather than replication of the concepts.

Hence, in this study there is also made use of written questionnaires, including open-ended questions and fixed questions with the option to explain. Unfortunately there was not enough time to also include several in-depth interviews, although they would give more insight in students’ reasoning.

### Research instrument - Questionnaire

At first, students were asked open-ended questions about the concept hereditary traits: “What is a ‘hereditary trait’?” and “Give five examples of hereditary traits”. These involve no examples or explanations and therefore test which examples come first to mind when considering the concept ‘hereditary trait’. This part is done before the questions with examples and explanations of traits because they may influence the students’ answers on these first questions.

The second part of the questionnaire consisted of multiple questions in which students were asked to classify examples of traits, illustrating the six different categories of hereditary traits, as hereditary or not, with an explanation of their categorization. The general format is as follows:

1. Explanation of the trait...

Would you regard this as a hereditary trait?

Yes

No

Not sure

Because.....

.....

As stated in the theoretical background, six categories of hereditary traits can be distinguished on which students might have a limited view. For each category multiple

examples are used. Preferably, three at both ends of the category, so six questions per category in total, in which ‘a’ is the one end of the category from which is expected students to be able to classify it correctly as a hereditary trait and ‘b’ is the other end of the category where students might not be able to classify it correctly as a hereditary trait. The ‘a-categories’ often include the same examples as these are the more traditional ones, which are compared to the new meanings. In the questionnaire itself, these questions are mixed up so students themselves cannot determine whether examples belong together. The following overview of the categories and examples is also given in appendix III to make it easier to look up while reading this paper.

- C1 Clearly visible traits (a) versus not-visible traits (b)
- a.
    1. The colour of the human iris.
    2. Natural hair shapes, such as strait or curly.
    3. Having a light or dark skin.
  - b.
    1. Haemophilia, a disease in which certain clotting factors are missing in the blood, leading to continuous bleeding after a vessels is damaged.
    2. Gilbert’s syndrome, an enzymatic disorder leading to high levels of bilirubin in blood.
    3. PKU, Phenylketonuria, a disease in which phenylketon builds up in the body due to a defect in the hepatic enzyme phenylalanine hydroxylase (PAH).
- C2 Traits on organism level (a) versus traits on lower organisational levels (b)
- a.
    1. Cystic fibrosis (CF), also known as mucoviscidosis, a disease in which patients have difficulty breathing and are more at risk for lung infections.
    2. Colour blindness, the inability to distinguish colours.
    3. Having a light or dark skin.
  - b.
    1. Abnormal mucus production in lung cells of patients with Cystic fibrosis (CF).
    2. The absence of a pigment type in the retina leading to colour blindness.
    3. High production of pigment in skin cells in people from African origin.
- C3 Traits which are independent from environmental factors (a) versus traits which are heavily influenced by the environment (b)
- a.
    1. The ability to fold your tongue.
    2. The colour of the human iris.
    3. Natural hair colour.
  - b.
    1. Having a musical talent.
    2. Being good at sports.
    3. Having a high IQ score.
- C4 Traits with a high variance within a population (a) versus traits with no variance within a population (b)
- a.
    1. Having a light or dark skin.
    2. The colour of the human iris.
    3. Natural hair shapes, such as strait or curly.
  - b.
    1. The possession of limbs.
    2. The ability to metabolize glucose.
    3. The ability of the skin to repair itself.
- C5 Static traits (a) versus dynamical traits (which change over time) (b)
- a.
    1. The colour of the human iris.
    2. Having ‘clubbed thumbs’, the tips of the thumbs being short and round with a short and wide nailbed.
    3. Having a tip-tilted nose.
  - b.
    1. Having lactose intolerance at a young age, which results in bowel problems when dairy-products are ingested. However, at when they grow older this often disappears.
    2. Older people having larger ears as they keep on growing during life-time.
    3. Sexual characteristics which develop during puberty.

4. Huntington's disease, a neurodegenerative disorder which affects muscle coordination and mental decline and behavioural symptoms, physical symptoms usually begin at the age of 30-40.
- C6 Traits on organismal level (a) versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers) (b)
- a.
    1. Having a light or dark skin.
    2. The colour of the human iris.
    3. Natural hair shapes, such as straight or curly.
  - b.
    1. Having a deviant haemoglobin type caused by a Sickle cell allele without having disease symptoms.
    2. Having a higher risk on breast cancer caused by a mutation in the BRCA-1 gene.
    3. Having a lower level of a certain type of neurotransmitter associated with a higher chance on aggressive behaviour.

To avoid that students recognize that all these traits are for some part hereditary, also a few non-hereditary traits are included.

1. Down-syndrome, a genetic disorder caused by a third copy of the chromosome 21.
2. Getting skin cancer after too much exposure to UV-light.
3. Having a congenital mental disability due to oxygen deficiency during birth.
4. Being born with 'spina bifida', a developmental disorder in which the embryonic neural tube isn't completely closed.
5. Being born with the foetal alcohol syndrome.

At the end of the questionnaire students are again asked the question "What is a 'hereditary trait'?" to see if their definition has changed after they saw the other examples.

It is made clear to the participants not to adjust their previous answers. The full questionnaire (in Dutch) can be found in 'Appendix I'.

### Pilot questionnaire

Before sending out the final questionnaires to all the participants, it was first tested on a few students. Students were observed while answering the questions and afterwards an interview was held. Part B of the questionnaire in particular, which contains questions with examples of traits to which students need to answer if they would classify it as a hereditary trait (yes/no/maybe) and explain their answer, might need adjusting. If lack of concentration is observed in students after a certain amount of questions, the questionnaire is too long and should be shortened. Also, when students take a lot of time answering a certain question or have a question about a certain question it might be that the explanation of the example needs to be adjusted or is too difficult and should be removed completely from the questionnaire.

Afterwards an interview was held with the students about the length of the questionnaire and the difficulty of the examples of traits. Students were then able to explain what exactly they did not understand about the example in order to adjust it when necessary.

The results of the pilot and the adjustments to the questionnaire can be found in the result section of this research proposal.

## Participants

The questionnaires were held in biology classes of upper secondary education in the fifth year of the pre-university tract (in the Netherlands, 5VWO). These students have had extensive education on the topic of genetics, only to be repeated once again just upon their final exams in the next (last) year. For practical reasons there is not chosen to use 6VWO students although they would be even more representative. However, it is expected that also VWO5 students should know most of the curriculum. If misconceptions or lack of understanding occurs in these students they will probably also occur within other students of secondary education of lower levels.

In total 149 students participated from five different teachers on four different schools, this is shown in table 1.

*Table 1: Participating schools, teachers and students*

School nr.	Name	City	Teacher	Number of students
1	Amersfoortse Berg	Amersfoort	A	26 (7 male, 19 female)
	Amersfoortse Berg	Amersfoort	B	25 (18 male, 7 female)
2	Minkema College	Woerden	C	22 (9 male, 13 female)
3	Roland Holst College	Hilversum	D	57 (22 male, 35 female)
4	De Werkplaats	Bilthoven	E	19 (6 male, 13 female)

The participating schools are located in four different cities in the Netherlands. Most schools probably contain mostly students with high educated parents which makes the results less generalizable.

In total, five different teachers were involved in this study. As it is assumable that teachers differ in their teaching and thereby make use of different examples when teaching genetics, there is controlled for the five different teachers. All teachers have at least one year teaching experience in upper secondary education.

## Data analysis

### Definitions of the concept 'hereditary trait'

In part A and C of the questionnaire students were asked to give a definition of the concept 'hereditary trait'. In order to be able to analyse the results quantitatively, the given definitions were analysed to contain the following characteristics or concepts:

- a) Heredity / passing on / etc.
- b) DNA / genes
- c) From birth / fertilisation
- d) Independent from environmental factors

Students were asked to give a definition upfront and after the questionnaire, both answers will be compared to see if the questionnaire itself influenced their understanding.

### Examples of hereditary traits

In part A of the questionnaire students were asked to give five examples of hereditary traits. All the given traits were analysed and categorised in:

Traditional traits (natural eye/hair colour, shape of the nose, body length, etc).

Medical conditions (e.g. blood type, certain diseases, colour blindness, etc).

Behavioural characteristics (e.g. IQ, character, musicality, etc).

### Classification of hereditary traits

Part B of the questionnaire consisted of 33 questions with explanations of different traits to which students had to answer if they would regard it as a hereditary trait (yes/no/maybe) and give an explanation to their answer. In order to be able to analyse the explanations quantitatively, the explanations were coded to contain one or more characteristics or concepts following a coding scheme (see Appendix II).

### Reliability

To ensure the reliability of the analysis 10% of the questionnaires (fifteen), chosen randomly, were analysed by another assessor and compared to the analysis of the researcher.

## Results

### Pilot questionnaire

Before sending out the questionnaires to all the participants, it was first tested on four students, two boys (18 and 16 years old) and two girls (both 16 years old). These students were observed while answering the questions to see if there would be a lack of concentration after a certain amount of questions and to give the opportunity to ask questions if an example of a trait was not clear. Afterwards an interview was held with the students about the length of the questionnaire and the difficulty of the examples of traits.

### Length of the questionnaire

All of the students finished within 25-30 minutes. The students found it pretty long, but not too long to answer the questions with full concentration. Therefore, the questionnaire wasn't shortened afterwards.

### Difficulty of examples

There were two examples that needed more explanation in order to make it better understandable for the students. One student was not sure about 'bilirubin' and therefore the explanation "a product of breaking down haemoglobin" was added. And it became clear that the explanation that Huntington's disease is a 'neurodegenerative' disorder is distracting as two students thought that 'generative' has something to do with heredity. Therefore the explanation was changed in 'the breakdown of neurons in certain parts of the brain'.

The adjusted version of the full questionnaire can be found in 'Appendix I' (in Dutch).

### Definitions of the concept 'hereditary trait'

In the beginning (part A) and at the end (part C) of the questionnaire students were asked to give a definition of the concept 'hereditary trait'. In order to be able to analyse the results quantitatively, the given definitions were analysed to contain the following characteristics or concepts:

- A) Heredity / passing on / etc.
- B) DNA / genes / chromosomes
- C) From birth / fertilisation
- D) Independent from environmental factors

The results of the definition of the concept 'hereditary trait' by students in the beginning of the questionnaire (Part A) are shown in table 2 and 3. Table 1 shows the total number of students who have mentioned characteristics in the above mentioned categories. Most of the students mentioned that hereditary traits are traits that are inherited or passed on from parents

to their offspring (92%) and/or mentioned that it is determined by DNA/genes/chromosomes (69%). A few (5%) also mentioned that it is present from birth. And 3% of the students also mentioned that it is not influenced by environmental factors.

Table 2: Results definition 'hereditary trait' in Part A

Category	Percentage of students
A) Hereditary, passed on, etc.	92% (n=137)
B) DNA/genes/chromosomes	69% (n=103)
C) From birth / fertilisation	5% (n=8)
D) Independent from environmental factors	3% (n=5)

Most students mentioned several characteristics from different categories. Table 3 therefore specifically shows the number of students that mentioned certain characteristics from one or more categories combined, including an example. It shows that most of the students mentioned that it is inherited or passed on from parents to their offspring and mentioned that it is determined by DNA/genes/chromosomes (category A and B combined). And 39 students only mentioned that it is inherited or passed on from parents to their offspring. Only 14 students answered another combination.

Table 3: Number of students

Category	Number of students	Example
A + B	91	"A trait that is passed on from parents to their offspring due to transmission of certain genes"
A	39	"A trait which you inherit from your parents"
B	7	"A trait which is caused by genes."
A + C	5	"A trait which you inherit from your parents and is present from birth."
A + D	2	"A trait that is passed on from parents to their offspring. Environmental factors didn't have any influence."
B + C	2	"A physical trait that is determined by genes and present from birth."
B + D	1	"A trait which is determined by your DNA without being influenced by environmental factors or learned behaviour."
A + B + D	1	"A trait which is in your genes. You get it from genes from your parents. It is not influenced by environmental factors."
C + D	1	"A trait that is congenital and you can't change anything about."

Interestingly, 7 of the students which mentioned characteristics from category A and B, also added that hereditary traits "are located on the X and Y chromosomes".

Students were asked to give a definition upfront (Part A) and after the questionnaire (Part C) and both answers were compared to see if the questionnaire itself influenced their meaning. Table 4 shows the amount of students that added certain characteristics to their definition at the end of the questionnaire. The characteristics have been divided in the previously mentioned categories.

Table 4: Results definition 'hereditary trait' in Part C

Added characteristics	Percentage of students
None	82% (n=122)
B) DNA/genes/chromosomes	9% (n=13)
D) Independent from environmental factors	5% (n=7)
A) Hereditary, passed on, etc.	2% (n=3)
C) From birth / fertilisation	1% (n=2)
A + D	1% (n=1)

Table 4 shows that most of the students (82%) did not change their minds about the definition of the concept 'hereditary trait' after finishing the questionnaire. Only a few, 26 in total, added certain characteristics. Most of the students (11 students) that changed their definition first only mentioned that a hereditary trait is inherited or passed on from parents to their offspring (category A) and afterwards added that it is determined by DNA/genes/chromosomes (category B).

To ensure the reliability of this analysis, 15 questionnaires (10%) were also analysed by another assessor. The analysis of the definition of the concept 'hereditary trait' corresponded for 97%.

### Examples of hereditary traits

In part A, in the beginning of the questionnaire, students were also asked to give five examples of hereditary traits. Table 5 shows that most of the students mentioned traditional traits which are on organism level, clearly visible on the outside, not influenced by environmental factors, static over a lifetime and with a high variance in a population, such as: natural eye colour, hair colour, shape nose, length body, etc. (83%). These traits correspond with the 'a' end of the categories described in the theoretical background, from which was expected that students would regard them as hereditary traits. However, from the third given example also other types of traits were mentioned such as: medical conditions (e.g. certain diseases, blood type, colour-blindness, immune system, allergies, weight, predisposition to get overweight, hormonal regulation blood type and certain diseases, 11%) and behavioural characteristics (e.g. IQ, intelligence, character, talent for sports, music, art, etc. 5%).

Table 5: Results of examples of hereditary traits given by students

Examples	Example number:					Total
	1	2	3	4	5	
Traditional traits	142	138	129	103	104	83% (n=616)
Medical conditions	5	9	16	27	23	11% (n=80)
Behavioural characteristics	1	2	4	14	14	5% (n=35)
None, unclear	1			5	8	2% (n=14)

Fifteen questionnaires (10%) were also analysed by another assessor to ensure reliability of the analysis. The analysis of the examples of hereditary traits corresponded for 100%.

## Classification of hereditary traits

Part B of the questionnaire consisted of 33 questions with explanations of different traits to which students had to answer if they would regard it as a hereditary trait (yes/no/maybe) and give an explanation to their answer. A quantitative analysis is made of students' classification. Moreover, the explanations of the students were coded to contain one or more characteristics or concepts following a coding scheme in order to give insight in students' argumentation. Students that classified traits as hereditary for the most part gave arguments such as: children resemble their parents, the information for the trait is coded in DNA/genes or that they learned about it in school. Therefore, only the argumentations when answered 'no' or 'maybe' are described in this results section. Except for the control traits, traits which are not hereditary, for which it is more interesting to know why students do classify these traits as hereditary.

### Category 1 - Clearly visible traits versus not-visible traits

Table 6 shows the results of students' classification of traits in category 1 'Clearly visible traits versus not-visible traits'. The greater part of the students regarded the clearly visible traits (C1a: natural hair shape, eye colour and skin colour) as a hereditary traits. Out of the 26 students that gave an explanation when answered 'no' or 'maybe', 14 mentioned in their argumentation that it could (also) be influenced by environmental factors.

The not-visible traits (C1b: Hemophilia, PKU and Gilbert's disease) were also for the most part regarded as hereditary traits, however, there were a lot of students that answered 'no' or 'maybe' compared to the visible traits. The students that did not classify these traits as hereditary, for the most part mentioned in their explanation that it is caused by a mutation (14 out of 66) or just that it is not coded in DNA/genes (12 out of 66). Interestingly, a lot of students appear to think that a mutation only takes place later on in life, only affect body cells and are therefore not hereditary. This finding is more elaborated in the description of category 6, where even more students gave such explanation. Out of the 128 students that gave an explanation when answered 'maybe', 80 students did not know the disease.

Table 6: Students' classification of traits in category 1 – Clearly visible traits (C1a) versus not-visible traits (C1b)

Category	Trait	Yes	No	Maybe	Blanc
C1a	<b>2. Hair shape</b>	135	1	13	0
	<b>3. Eye colour</b>	143	2	4	0
	<b>4. Skin colour</b>	142	1	6	0
C1b	<b>11. Haemophilia</b>	82	23	44	0
	<b>12. PKU</b>	52	44	53	0
	<b>13. Gilbert's</b>	71	22	55	1

The differences between the answers to C1a and C1b are significant with  $P < 0,001$ .

## Category 2 - Traits on organism level versus traits on lower organisational levels

Students' classification of traits in category 2 'Traits on organism level versus traits on lower organisational levels' are shown in table 7. Three traits: natural skin colour, colour blindness and Cystic Fibrosis were explained on organism level (C2a) and on lower organisational levels (C2b).

- a.
  1. Cystic fibrosis (CF), also known as mucoviscidosis, a disease in which patients have difficulty breathing and are more at risk for lung infections.
  2. Colour blindness, the inability to distinguish colours.
  3. Having a light or dark skin.
- b.
  1. Abnormal mucus production in lung cells of patients with Cystic fibrosis (CF).
  2. The absence of a pigment type in the retina leading to colour blindness.
  3. High production of pigment in skin cells in people from African origin.

Most of the students classified the traits when described on organism level (C2a) as hereditary. The students that did not regard these traits as hereditary, for the most part mentioned in their explanation that it could (also) be influenced by environmental factors (7 out of 41) or that it is caused by a mutation (6 out of 41). Out of the 61 students that answered 'maybe' and gave an explanation, for the most part (18 students) did not know enough about the trait.

Approximately the same results were seen when the traits were described on lower organisational levels (C2b). Note that the traits described on organism level were placed in the beginning of the list of traits and the traits described on lower organisational levels at the end of the list to avoid recognition. However, still a lot of students mentioned in their explanation something like "same as the previous question about [...]". Most of the students that did not classify these traits as hereditary mentioned that is influenced by environmental factors (6 out of 33) or that it is caused by a mutation (5 out of 33). Out of the 40 students that answered 'maybe' and gave an explanation, 22 students did not know enough about the trait, especially with Cystic Fibrosis and colour blindness.

Table 7: Students' classification of traits in category 2 – Traits on organism level (C2a) versus traits on lower organisational levels (C2b)

Category	Trait	Yes	No	Maybe	Blanc
C2a	<b>4. Skin colour</b>	142	1	6	0
	<b>8. Colour blindness</b>	107	17	25	0
	<b>9. CF</b>	83	29	37	0
C2b	<b>31. CF</b>	85	31	32	1
	<b>32. Colour blindness</b>	119	15	14	1
	<b>33. Skin Colour</b>	134	10	4	1

The differences between the answers to C2a and C2b are not significant with  $P > 0,5$ .

**Category 3 - Traits which are independent from environmental factors  
versus traits which are heavily influenced by the environment**

The results of students classification of traits in category 3 ‘Traits which are independent from environmental factors versus traits which are heavily influenced by the environment’ are shown in table 8. Most students regarded the traits which are independent from environmental factors (C3a: Natural hair colour, eye colour and the ability to fold your tongue) as hereditary. However, especially the ability to fold your tongue was also often regarded as not hereditary or maybe. In their argumentation, most of the students mentioned that it is (also) influenced by environmental factors (55 out of 72), as “the tongue is a muscle and can be trained”.

The greater part of the students also regarded the traits which are heavily influenced by environmental factors as hereditary. However, a lot of students also answered ‘no’ or ‘maybe’. Most of these students mentioned in their argumentation that it can (also) be influenced by environmental factors (146 out of 191).

*Table 8: Students’ classification of traits in category 3 – Traits which are independent from environmental factors (C3a) versus traits which are heavily influenced by the environment (C3b)*

Category	Trait	Yes	No	Maybe	Blanc
C3a	<b>1. Hair colour</b>	149	0	0	0
	<b>3. Eye colour</b>	143	2	4	0
	<b>5. Fold tongue</b>	71	48	30	0
C3b	<b>21. Musical talent</b>	70	45	34	0
	<b>22. Sports talent</b>	69	37	43	0
	<b>23. IQ</b>	79	32	37	1

The differences between the answers to C3a and C3b are significant with  $P < 0,001$ .

**Category 4 - Traits which a high variance in a population versus traits  
with no variance within a population**

Table 9 shows the results of students’ classification of traits in category 4 ‘Traits which a high variance in a population versus traits with no variance within a population’. The traits with a high variance within the population (C4a) were the same as the clearly visible traits in category 1, most of the students regarded these traits as hereditary. The ones that did not, for the most part mentioned in their argumentation that it could (also) be influenced by environmental factors (14 out of 26).

The traits with no variance within a population (C4b: the ability to metabolize glucose, the ability to repair the skin and the possession of limbs) were not classified as hereditary by most of the students. For the most part, these students mentioned in their argumentation that

it is not hereditary because everyone can or has it (115 out of 161) and the same argumentation was found in the ones that answered ‘maybe’ (23 out of 51).

Table 9: Students’ classification of traits in category 4 – Traits with a high variance in a population (C4a) versus traits with no variance within a population (C4b)

Category	Trait	Yes	No	Maybe	Blanc
C4a	<b>2. Hair shape</b>	135	1	13	0
	<b>3. Eye colour</b>	143	2	4	0
	<b>4. Skin colour</b>	142	1	6	0
C4b	<b>15. Glucose metabolism</b>	65	67	17	0
	<b>16. Skin repair</b>	61	65	23	0
	<b>17. Limbs</b>	50	82	17	0

The differences between the answers to C4a and C4b are significant with  $P < 0,001$ .

### Category 5 - Static traits versus dynamical traits (which change over time)

Students’ classification of traits in category 5 ‘Static traits versus dynamical traits (which change over time)’ is shown in table 10. Most of the students regarded the static traits (C5a: eye colour, clubbed thumbs and having a tip-tilted nose) as hereditary. The ones that did not, especially at the clubbed thumbs, mentioned in their argumentation that they did not know enough about the trait (18 out of 52).

Three of the four dynamical traits (C5b: sexual characteristics, big ears of elderly and Huntington’s disease) were regarded as hereditary by most of the students. However, lactose intolerance was classified as not hereditary by most of the students. The students that did not regard these traits as hereditary for the most part mentioned in their explanation that it is the same in everyone (46 out of 142), especially with sexual characteristics and big ears of elderly, or that it changes over time (39 out of 142). Students that answered ‘maybe’, mostly mentioned in their explanation that they did not know enough about the trait (34 out of 83).

Table 10: Students’ classification of traits in category 5 – Static traits (C5a) versus dynamical traits (which change over time) (C5b)

Category	Trait	Yes	No	Maybe	Blanc
C5a	<b>3. Eye colour</b>	143	2	4	0
	<b>6. Clubbed thumbs</b>	109	6	34	0
	<b>7. Tip-tilted nose</b>	141	3	5	0
C5b	<b>18. Sexual characteristics</b>	78	50	21	0
	<b>20. Big ears elderly</b>	68	62	18	1
	<b>28. Lactose intolerance</b>	45	72	31	1
	<b>30. Huntington’s</b>	97	23	28	1

The differences between the answers to C5a and C5b are significant with  $P < 0,001$ .

### Category 6 - Traits on organism level versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker)

The results of students' classification of traits in category 6 'Traits on organism level versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker)' are shown in table 11. The traits on organism level (C6a) were the same as the clearly visible traits in category 1 and the traits with a high variance within the population in category 4. Most of the students regarded these traits as hereditary. The ones that did not, for the most part mentioned in their argumentation that it could (also) be influenced by environmental factors (14 out of 26).

The traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker) (C6b: having a higher risk on breast cancer caused by a mutation in the BRCA-1 gene, having a deviant haemoglobin type caused by a Sickle cell allele without having disease symptoms and having a lower level of a certain type of neurotransmitter associated with a higher chance on aggressive behaviour) were regarded as hereditary by most of the students. The students that did not regard these traits as hereditary, for the most part gave an explanation, such as "it is caused by a mutation" (28 out of 48). As mentioned earlier, these students seem to believe that a mutation is never hereditary. Some students even elaborated "a mutation is not hereditary", "a mutation is not inherited but only takes place in one's body" or "this mutation is not visible in the whole body". The ones that answered 'maybe' to these traits did not know enough about the trait (33 out of 60). Moreover, the ones that answered maybe to the trait about breast cancer mentioned in their explanation that they were not sure when the mutation took place and if it is hereditary (10 out of 18).

Table 11: Students' classification of traits in category 6 – Traits on organism level (C6a) versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker) (C6b)

Category	Trait	Yes	No	Maybe	Blanc
C6a	<b>2. Hair shape</b>	135	1	13	0
	<b>3. Eye colour</b>	143	2	4	0
	<b>4. Skin colour</b>	142	1	6	0
C6b	<b>24. Risk Breast cancer</b>	96	33	20	0
	<b>26. Hemoglobine</b>	126	10	12	1
	<b>27. Risk aggression</b>	79	31	38	1

The differences between the answers to C6a and C6b are significant with  $P < 0,001$ .

### Control traits – Not hereditary

Table 12 shows the results of students' classification of the five control traits (Down Syndrome, having a congenital mental disability due to oxygen deficiency during birth, being born with 'spina bifida', a developmental disorder in which the embryonic neural tube isn't completely closed, getting skin cancer after too much exposure to UV-light and being born with the fetal Alcohol syndrome). These traits were classified as not hereditary by most of the

students. However, a lot of students regarded Down Syndrome and Fetal Alcohol Syndrome as hereditary.

Out of the 27 students that classified Down Syndrome as hereditary and gave an explanation, 16 students mentioned that it is caused by a fault in the DNA/genes/chromosomes and therefore hereditary. The ones that answered ‘maybe’, for the most part mentioned that they did not know if it was passed on from parents to their offspring because parents without Down Syndrome can have children with Down Syndrome (8 out of 16).

Most of the students that regarded being born with Foetal Alcohol Syndrome, mentioned in their argumentation that it is present from birth (5 out of 16). Almost all of the students that answered ‘maybe’ and gave an explanation did not know what Foetal Alcohol Syndrome implies (25 out of 26).

Table 12: Students’ classification of control traits - not hereditary

Category	Trait	Yes	No	Maybe	Blanc
Control	<b>10. Down Syndrome</b>	40	92	17	0
	<b>14. Oxygen deficit</b>	5	142	2	0
	<b>19. Spinal bifida</b>	18	111	19	1
	<b>25. Skin cancer</b>	7	134	7	1
	<b>29. Fetal alcohol syndrome</b>	40	80	28	1

The differences between the answers to the control traits are significant with  $P < 0,001$ .

Fifteen questionnaires (10%) were also analysed by another assessor to ensure reliability of the analysis. The analysis of classification of hereditary traits (yes/no/maybe) corresponded for 99%.

### Students’ argumentation

The percentage of students that gave a certain type of argumentation when answered ‘no’ or ‘maybe’ to the traits in category C1b, C2b, C3b, C4b, C5b and C6b are shown in table 13 and 14 respectively. The argumentation when answered ‘yes’ or ‘maybe’ to the control traits are shown in Table 15.

Table 13 shows the type of arguments when answered ‘no’ per category. The following four type of argumentations were predominantly given by the students.

#### *No variance within a population*

For example: “Everyone can do it”. This type of argument was especially given in category 4 (Traits with a high variance in a population versus traits with no variance within a population) and 5 (Static traits versus dynamical traits (which change over time))

*(Partly) influenced by environmental factors*

For example: “This could also be trained”. This type of argument was especially given in category 2 (Traits on organism level versus traits on lower organisational levels) and 3 (Traits which are independent from environmental factors versus traits which are heavily influenced by the environment)

#### *Caused by a mutation*

For example: “This is caused by a mutation”. This argumentation was especially given in category 1 (Clearly visible traits versus not-visible traits) and 6 (Traits on organism level versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker)).

#### *Not consistent over time*

For example: “It changes during life”. This type of argument was especially given in category 5 (Static traits versus dynamical traits (which change over time)).

Table 13: Type of arguments given by students when answered ‘no’ per category

	<b>C1b</b>	<b>C2b</b>	<b>C3b</b>	<b>C4b</b>	<b>C5b</b>	<b>C6b</b>
	% No	% No	% No	% No	% No	% No
	20% (n=89)	13% (n=56)	26% (n=114)	48% (n=214)	46% (n=207)	17% (n=74)
<b>Type of argument</b>						
No variance within a population	2%	0%	1%	71%	32%	2%
(Partly) influenced by environmental factors	14%	18%	70%	1%	4%	8%
Caused by a mutation	27%	15%	0%	1%	2%	52%
Not consistent over time	3%	3%	0%	1%	27%	2%
Not coded in DNA / genes / chromosomes	18%	0%	10%	4%	4%	8%
No resemblance parents and offspring	12%	12%	6%	4%	3%	6%
Knowledge	11%	3%	1%	1%	1%	0%
Personal experience	0%	0%	1%	0%	1%	0%
Not present from birth	2%	0%	0%	0%	1%	0%

The percentage of students given certain type of arguments when answered ‘maybe’ to category C1b, C2b, C3b, C4b, C5b and C6b are shown in table 14. The following four argumentations were mostly given by students:

#### *Knowledge*

For example: “I don’t know this disease”. This argumentation was especially given in category 1 (Clearly visible traits versus not-visible traits), 2 (Traits on organism level versus traits on lower organisational levels), 5 (Static traits versus dynamical traits (which change over time)) and 6 (Traits on organism level versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker)). These, however, are not really argumentations for classifying a certain trait as hereditary, they simply do not know enough about the trait.

*(Partly) influenced by environmental factors*

For example: “This is for some part inheritable, but could also be trained”. This type of argument was especially given in category 1 (Clearly visible traits versus not-visible traits), category 2 (Traits on organism level versus traits on lower organisational levels), category 3 (Traits which are independent from environmental factors versus traits which are heavily influenced by the environment) and 5 (Static traits versus dynamical traits (which change over time)).

*No variance within a population*

For example: “Because everyone has it”. This type of argument was especially given in category 4 (Traits with a high variance in a population versus traits with no variance within a population).

*Caused by a mutation*

For example: “I don’t know how the mutation started”. This type of argument was especially given in category 6 (Traits on organism level versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarker)).

Table 14: Type of arguments given by students when answered 'maybe' per category

	C1b	C2b	C3b	C4b	C5b	C6b
	% Maybe	% Maybe	% Maybe	% Maybe	% Maybe	% Maybe
	34% (N=152)	11% (N=50)	26% (N=114)	13% (N=57)	22% (N=98)	16% (N=70)
<b>Type of argument</b>						
Knowledge	63%	55%	7%	12%	43%	55%
(Partly) influenced by environmental factors	14%	21%	82%	8%	18%	7%
No variance within a population	0%	3%	1%	45%	12%	0%
Caused by a mutation	5%	5%	0%	0%	0%	18%
Coded in DNA / genes / chromosomes	13%	3%	0%	12%	2%	10%
Resemblance parents and offspring	12%	3%	8%	4%	2%	7%
Present from birth	5%	0%	0%	4%	0%	0%
Not consistent over time	1%	0%	0%	0%	7%	0%
Personal experience	0%	0%	0%	0%	4%	0%

Table 15 shows the percentage of students given a certain type of argument when answered ‘yes’ or ‘maybe’ to the control traits. Students that classified these traits as hereditary, for the most part (38%) mentioned in their explanation that it is coded in DNA/genes/chromosomes. This could indicate a misconception, namely that students seem to confuse ‘hereditary’ to ‘genetic’, as not every genetic influence is also hereditary. The students that answered ‘maybe’, for the most part (55%) did not know enough about the trait.

Table 15: Type of arguments given by students when answered 'yes' or 'maybe' to the control traits

	<b>Controls</b>	
	<b>% Yes</b>	<b>% Maybe</b>
<b>Type of argument</b>	15% (N=110)	23% (N=73)
Coded in DNA / genes / chromosomes	38%	2%
Knowledge	7%	55%
Resemblance parents and offspring	23%	16%
Present from birth	25%	0%
Caused by a mutation	7%	9%
(Partly) influenced by environmental factors	4%	8%
Personal experience	3%	0%
Variance within a population	0%	0%
Consistent over time	0%	0%

Fifteen questionnaires (10%) were also analysed by another assessor to ensure reliability of the analysis. The coding of argumentations (following a coding scheme; see Appendix II) corresponded for 93%.

## Conclusion

To answer the main research question ‘Which meaning do students, with knowledge of genetics on upper secondary school biology level, attribute to the concept ‘hereditary trait’?’ four sub questions need to be answered.

### What definition give students to the concept ‘hereditary trait’?

In the beginning and at the end of the questionnaire, students were asked to give a definition of the concept hereditary trait, most students answered that they are traits that are inherited or passed on by parents to their offspring (92%) and/or that it is coded in DNA/genes/chromosomes (69%).

### What kind of examples of hereditary traits do students come up with?

When asked to give five examples of hereditary traits in 83% of the cases the students mentioned traditional examples of traits which are visible on the outside, independent from environmental factors, have a high variance within a population and do not change over time, such as: eye colour, skin colour and hair shape. From the third given example also other types of traits were mentioned such as: medical conditions (e.g. blood type and certain diseases; 11%) and behavioural characteristics (e.g. IQ, character, musicality; 5%).

### Which traits are classified by students as hereditary?

Part B of the questionnaire consisted of 33 questions with explanations of different traits to which students had to answer if they would regard it as a hereditary trait (yes/no/maybe) and give an explanation to their answer. As stated in the theoretical background, six categories of hereditary traits can be distinguished on which students might have a limited view:

- C1 Clearly visible traits (a) versus not-visible traits (b)
- C2 Traits on organism level (a) versus traits on lower organisational levels (b)
- C3 Traits which are independent from environmental factors (a) versus traits which are heavily influenced by the environment (b)
- C4 Traits with a high variance within a population (a) versus traits with no variance within a population (b)
- C5 Static traits (a) versus dynamical traits (which change over time) (b)
- C6 Traits on organismal level (a) versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers) (b)

For each category multiple examples were used at both ends of the category, in which ‘a’ is the one end of the category from which is expected students to be able to classify it correctly as a hereditary trait and ‘b’ is the other end of the category where students might not be able to classify it correctly as a hereditary trait.

The greater part of the students classified all the traits in category 1, 3, 5 and 6 as hereditary, both at the 'a' and 'b' end of the categories. However, to the traits at the 'b' end of the category also a lot of students answered 'no' or 'maybe'. This difference was significant with  $P < 0,001$ . In category 4 even a bigger difference was seen as most of the students did not classify the traits at the 'b' end of the category (the ability to metabolize glucose metabolism, the ability of the skin to repair itself and the possession of limbs) as hereditary. This finding corresponds with the research of Duijts (2016) which makes clear that all the scientists state that hereditary traits or phenotype must concern traits for which there is variance within a population in order to be able to perform research on it. The difference in classification of traits in category 2 was not significant with  $P > 0,5$ . In this case three traits: natural skin colour, colour blindness and Cystic Fibrosis were explained on organism level (C2a) and on lower organisational levels (C2b). The traits described on organism level were placed in the beginning of the list of traits and the traits described on lower organisational levels at the end of the list to avoid recognition. However, still a lot of students mentioned in their explanation something like "same as the previous question about [...]". This means that students recognize that traits on lower organisational levels are also hereditary.

To avoid that students recognize that all these traits are for some part hereditary, also a few non-hereditary traits were included. These traits were classified as not hereditary by most of the students. However, a lot of students regarded Down Syndrome and Foetal Alcohol Syndrome as hereditary.

### What type of arguments do students use to classify a trait as not hereditary?

Students were also asked to add an explanation to their answer (yes/no/maybe) to all these traits to give more insight in students argumentation. These explanations when answered 'no' or 'maybe' to the traits in category C1b, C2b, C3b, C4b, C5b and C6b were analysed to contain certain characteristics following a coding scheme (see Appendix II). The type of arguments given by students differed per category. There were three arguments which were predominantly present in students explanation to classify a trait as not hereditary or maybe.

#### *No variance within a population*

This argument was especially given in category C4b (traits with no variance within a population; 71%) and C5b (dynamical traits (which change over time); 32%) when answered 'no' and C4b when answered 'maybe' (traits with no variance within a population; 45%).

#### *(Partly) influenced by environmental factors*

This argument was especially given in category C2b (traits on lower organisational levels; 18%) and C3b (traits which are heavily influenced by the environment; 70%) when answered

‘no’ and category C3b (traits which are heavily influenced by the environment; 79%) and C4b (traits with no variance within a population; 23%) when answered ‘maybe’.

*Caused by a mutation*

This argument was especially given in category C1b (not-visible traits; 27%) and category C1b (not-visible traits; 52%) when answered ‘no’ and in C6b (traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers); 18%) when answered ‘maybe’.

**Which meaning do students, with knowledge of genetics on upper secondary school biology level, attribute to the concept ‘hereditary trait’?**

This study indicates that students have a limited view on the concept ‘hereditary trait’. When asked to give a definition of the concept, most of the students only mention that these traits that are inherited or passed on by parents to their offspring (92%) and/or that it is coded in DNA/genes/chromosomes (69%). However, scientist state that they must be (for some part) also influenced by environmental factors, otherwise this term (or the term phenotype) is not used (Duijts, 2016). This limited view is also seen in the argumentation of students when they did not classify a trait as hereditary or answered ‘maybe’. A lot of students then mentioned in their argumentation something like “because it can be trained” or “because it can also be influenced by other factors”. Moreover, when asked to give five examples of hereditary traits mostly traditional examples of traits (which are visible on the outside, independent from environmental factors, have a high variance within a population and do not change over time, such as: eye colour, skin colour and hair shape) were mentioned (83%).

The results of students’ classification of hereditary traits indicate that students do regard the traits on lower organisational levels (C2b) as hereditary. The not visible traits (C1b), traits which are heavily influenced by environmental factors (C3b), dynamical traits (which change over time) (C5b) and traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers) (C6b) were classified as not hereditary or ‘maybe’ by a lot of students. Especially, the traits with no variance within a population (C4b) were regarded by most students as not hereditary, this is consistent with the view of scientists (Duijts, 2016).

To regard a trait as not hereditary or maybe, most of the students mentioned in their argumentation that there is no variance within a population, that it is (partly) influenced by environmental factors or that it is caused by a mutation.

## Discussion

In this section the generalizability and limitations of this study are discussed. And implications for further research and practice are given.

### Generalizability

The questionnaires were held among a heterogeneous group of 149 students (62 male, 87 female) from five different teachers from four different schools in four different cities in the Netherlands. The five teachers were involved in this study had at least one year teaching experience in upper secondary education. It is assumable that teachers differ in their teaching and thereby make use of different examples when teaching genetics. However, no big differences were seen between the students of the different teachers. Moreover, the sample population is big enough in order to be able to generalize the results for the population at large. Nonetheless, there are some limitations to the generalizability concerning the sample group. First, the students were chosen from biology classes of upper secondary education in the fifth year of the pre-university tract (in the Netherlands, 5VWO). These students have had extensive education on the topic of genetics, only to be repeated once again just upon their final exams in the next (last) year. If misconceptions or lack of understanding occurs in these students they will probably also occur within other students of secondary education of lower levels. However, it has already been a year since these students have been educated on this topic and therefore a lot of conceptual knowledge is not clear anymore. Second, most of the schools participating in this study probably contain mostly students with high educated parents which makes the results less generalizable. Therefore, it would be interesting to perform this research on other schools, which contain more students with less higher educated parents, and to use VWO6 students after they repeated this topic in biology class to be more representative. Moreover, it would be interesting to perform this research on students that do not follow biology classes to see if biology education makes any difference.

### Limitations and implications for future research

There were also several limitations to the questionnaire itself. It became clear that a lot of students did not know enough about certain traits given in part B of the questionnaire to which students had to answer if they would regard it as hereditary (yes/no/maybe). Around 50% of the students that answered 'maybe' to traits in category 1, 2, 5, 6 and the control traits mentioned in their argumentation that they did not know enough about the trait. Therefore, this is not regarded as an argument for classifying a trait as hereditary or not. In future research it might be better to use other examples which are clear to the students to be able to

get more insight in their classification of traits. Moreover, there were a lot of students that did not give an explanation to their answers, especially approaching the end of the questionnaire. Therefore, it might be better to shorten the questionnaire in order to maintain a good concentration and motivation to explain their answers. Also, it would be helpful when teachers could walk around and check if students are seriously answering the questionnaire.

Most of the results were significant and had a high inter-rater reliability and are therefore discussed further on under ‘implications’. However, the difference in students classification of traits in category 2 ‘Traits on organism level (C2a) versus traits on lower organisational levels (C2b)’ were not significant with  $P > 0,5$ . In this category three traits: natural skin colour, colour blindness and Cystic Fibrosis were explained on organism level (C2a) and on lower organisational levels (C2b).

- a.
  1. Cystic fibrosis (CF), also known as mucoviscidosis, a disease in which patients have difficulty breathing and are more at risk for lung infections.
  2. Colour blindness, the inability to distinguish colours.
  3. Having a light or dark skin.
- b.
  1. Abnormal mucus production in lung cells of patients with Cystic fibrosis (CF).
  2. The absence of a pigment type in the retina leading to colour blindness.
  3. High production of pigment in skin cells in people from African origin.

About the same results were found for the three traits, either described on organism level or lower organisational levels (see table 7). Note that the traits described on organism level were placed in the beginning of the list of traits and the traits described on lower organisational levels at the end of the list to avoid recognition. However, still a lot of students mentioned in their explanation something like “same as the previous question about [...]”. This means, however, that students do recognize the traits on lower organisational levels as hereditary.

Moreover, research of Duijts (2016) indicates that scientists are not even clear about the concept ‘hereditary trait’. Some researchers seem to find that the term is confusing and therefore better not to be used because it indicates that traits can be grouped as hereditary or not hereditary while in fact every trait is for some part hereditary. A few researchers even find that the concept hereditary trait equals genotype. Most researchers, therefore, prefer to use the term ‘phenotype’ when research is done about traits with a certain genetic compound. However, there is no real consensus about the meaning of that concept as well, for example: if it must be visible on the outside of an organism or also if it is present on lower organisational levels. In future research it might be interesting to find out which meaning students contribute to the concept ‘phenotype’. The same questionnaire could be used, or adjusted using the previously described limitations and implications for future research.

### Implications for genetics education

As this research is for a big part generalizable and most of the results were significant, several implications for genetics education can be given. The results indicate a limited view of the concept ‘hereditary trait’ which needs to be addressed in genetics education. At first, students fail to recognize that hereditary traits are always (for some part) influenced by environmental factors. A lot of students even appear to think that if a trait can be influenced by environmental factors, such as: training, it is not hereditary. Teachers in genetics education must emphasise the fact that every hereditary trait is for some part influenced by environmental factors. Especially, as traits which are important for scientific literacy are for a great part influenced by environmental factors. Therefore, it might be more important to teach students more about these traits than about the colour of the human iris, natural hair and skin colour, etc.

Secondly, when asked to give five examples of hereditary traits mostly traditional examples of traits (which are visible on the outside, independent from environmental factors, have a high variance within a population and do not change over time, such as: eye colour, skin colour and hair shape) were mentioned (83%). This was already expected as these traits are often used in biology textbooks used in upper secondary education. However, Draanen (2016) performed research on often used textbooks in upper secondary biology education to find out how many examples of traits on the ‘b’ end of the six previously described categories are given. These results are shown in figure 1 and show that also a lot of not visible traits (C1b) are given by textbooks.

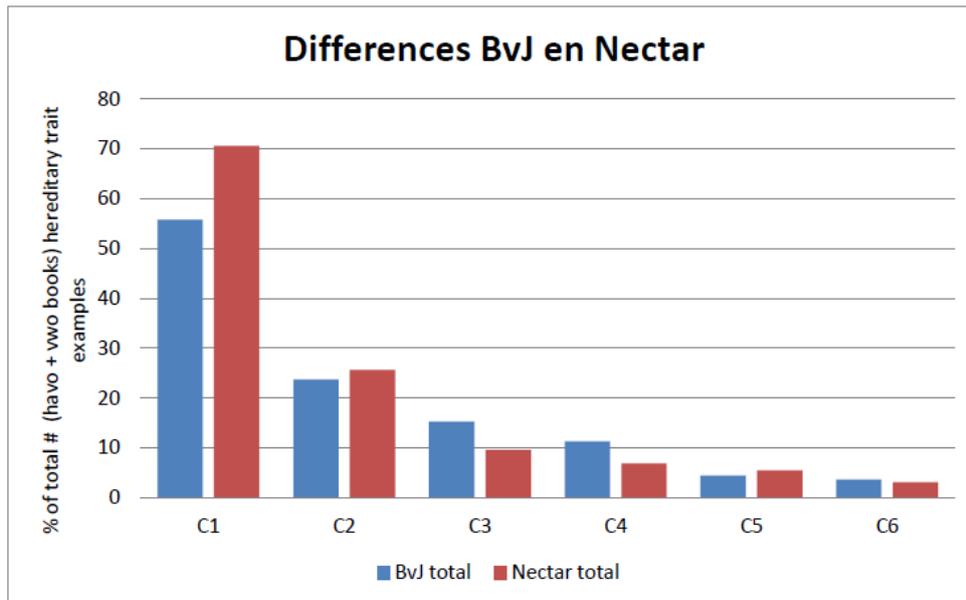


Figure 1: Hereditary traits mentioned in two commonly used textbooks in upper secondary biology education (Draanen, 2016)

Figure 1 makes clear that not many examples of traits which are heavily influenced by environmental factors (C3b), have no variance within a population (C4b), are dynamical (which change over time) (C5b) and on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers) (C6b). This finding could explain that a lot of students did not regard traits in these categories as hereditary. It might be helpful to adjust schoolbooks to contain more of these examples as schoolbooks are mostly used as guidelines for teachers. Especially, as most of the students regarded the traits with no variation in a population (C4b) as not hereditary, there seem to be a misconception that DNA only contains genes for variable traits. However, most of the coding DNA influences traits that everyone has.

Thirdly, a few misconceptions became clear among the students which need to be addressed by teachers in order to establish better understanding of genetics. When asked to give a definition of the concept 'hereditary trait' 7 students (from four different teachers) who mentioned in their definition that hereditary traits are traits that are inherited or passed on by parents to their offspring and that it is coded in DNA/genes/chromosomes especially mentioned that they "are located on the X and Y chromosomes". Of course, hereditary traits are not limited to the sex chromosomes, but can occur on all the other chromosomes. Maybe the focus on Mendelian inheritance, practicing the crossing-schemes of gender-related traits might lead students astray from this bigger picture. Moreover, it was interesting to find that a lot of students that did not classify a certain trait as hereditary supported their answer with the argument "because it is caused by a mutation" (category 1: 27%, category 6: 52%). A lot of students appear to think that a mutation always takes place later on life, only affect body cells

and are therefore not hereditary. Some students even elaborated “a mutation is not hereditary”, “a mutation is not inherited but only takes place in one’s body” or “this mutation is not visible in the whole body”. Maybe the focus on the environmental factors that could cause mutations, such as: radiation, smoking, etc. might form a limited view on how mutations can occur. More emphasis must be given to the heredity of mutations by biology teachers. Furthermore, students seem to confuse ‘hereditary’ to ‘genetic’, as 38% of the students mentioned in their explanation to regard a control trait as hereditary that they do because it is coded in DNA/genes/chromosomes.

## References

- Bearden, C. E., & Freimer, N. B. (2006). Endophenotypes for psychiatric disorders: ready for primetime?. *Trends in Genetics*, 22(6), 306-313.
- Billingsley, J., & Carlson, K. A. (2010). Epigenetic Effects of Diet on Fruit Fly Lifespan: An Investigation to Teach Epigenetics to Biology Students. *The American Biology Teacher*, 72(4), 231-234.
- Bodmer, W. F. (1997). *The book of man: the Human Genome Project and the quest to discover our genetic heritage*. Oxford University Press.
- Boujemaa, A., Pierre, C., Sabah, S., Salaheddine, K., Jamal, C., & Abdellatif, C. (2010). University Students' Conceptions about the Concept of Gene: Interest of Historical Approach. *Online Submission*, 7(2), 9-15.
- 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(1), 15-22.
- Browning, M. E., & Lehman, J. D. (1988). Identification of student misconceptions in genetics problem solving via computer program. *Journal of Research in Science Teaching*.
- Cho, H. H., Kahle, J. B., & Nordland, F. H. (1985). An investigation of high school biology textbooks as sources of misconceptions and difficulties in genetics and some suggestions for teaching genetics. *Science Education*, 69(5), 707-719.
- Christensen, A.J. (2000). Patient-by-treatment context interaction in chronic disease: A conceptual framework for the study of patient adherence. *Psychosomatic Medicine*, 62, 435-443
- Churchill, F. B. (1974). William Johannsen and the genotype concept. *Journal of the History of Biology*, 7(1), 5-30.
- College voor examens (2013a). Syllabus centraal examen 2015 Biologie VMBO. Utrecht.
- College voor examens (2013b). Syllabus centraal examen 2015 Biologie VWO. Utrecht.
- College voor examens (2014). Syllabus centraal examen 2015 Biologie HAVO. Utrecht.
- Collins, F. S., & McKusick, V. A. (2001). Implications of the Human Genome Project for medical science. *Journal of the American Medical Association*, 285(5), 540-544.
- Darwin, C. (1859). *On the origin of species*. London: Murray, 434.

- DeBoer, G. E. (2000). Scientific Literacy: Another Look at Its Historical and Contemporary Meanings and Its Relationship to Science Education Reform. *Journal of Research in Science Teaching*, 37(6), 582-601.
- Dougherty, M. J. (2009). Closing the gap: inverting the genetics curriculum to ensure an informed public. *The American Journal of Human Genetics*, 85(1), 6-12.
- Draanen, D. (2015). What is a phenotype? In press. Utrecht University.
- Duijts, B. (2016). Definities en betekenissen van de begrippen erfelijke eigenschap en fenotype in verschillende wetenschapsdomeinen binnen de Life Sciences. In press. Utrecht University.
- Duncan, R. G., & Reiser, B. J. (2007). Reasoning across ontologically distinct levels: Students' understandings of molecular genetics. *Journal of Research in Science Teaching*, 44(7), 938-959.
- Duncan, R. G., Rogat, A. D., & Yarden, A. (2009). A learning progression for deepening students' understandings of modern genetics across the 5th-10th grades. *Journal of Research in Science Teaching*, 46(6), 655-674.
- Dupré, J. (2008). *The constituents of life*. Uitgeverij Van Gorcum.
- Dupré, J. (2012). *Processes of life: essays in the philosophy of biology*. Oxford University Press.
- Gericke, N. (2009). Science versus School-science: Multiple models in genetics-The depiction of gene function in upper secondary textbooks and its influence on students' understanding.
- Gericke, N. M., & Hagberg, M. (2007). Definition of historical models of gene function and their relation to students' understanding of genetics. *Science & Education*, 16(7-8), 849-881.
- Gericke, N. M., & Hagberg, M. (2010). Conceptual incoherence as a result of the use of multiple historical models in school textbooks. *Research in science Education*, 40(4), 605-623.
- Gericke, N. M., Hagberg, M., dos Santos, V. C., Joaquim, L. M., & El-Hani, C. N. (2014). Conceptual Variation or Incoherence? Textbook Discourse on Genes in Six Countries. *Science & Education*, 23, 381 - 416.
- Goldberg, L. R. (1993). The structure of phenotypic personality traits. *American psychologist*, 48(1), 26.

- Gottesman, I. I., & Gould, T. D. (2003). The endophenotype concept in psychiatry: etymology and strategic intentions. *American Journal of Psychiatry*, 160(4), 636-645.
- Hamburger, V., & Hamilton, H. L. (1951). A series of normal stages in the development of the chick embryo. *Journal of morphology*, 88(1), 49-92.
- Hickey, D. T., & Kindfield, A. C. H. (1999). Assessment-oriented scaffolding of student and teacher performance in a technology-supported genetics environment. Paper presented at the annual meeting of the American Educational Research Association, Montreal, Canada.
- Imesch, P. D., Wallow, I. H., & Albert, D. M. (1997). The color of the human eye: a review of morphologic correlates and of some conditions that affect iridial pigmentation. *Survey of ophthalmology*, 41, S117-S123.
- Jennings, B. (2004). Genetic literacy and citizenship: possibilities for deliberative democratic policymaking in science and medicine. *The Good Society*, 13(1), 38-44.
- Jiménez-Aleixandre, M. P. (2014). Determinism and Underdetermination in Genetics: Implications for Students' Engagement in Argumentation and Epistemic Practices. *Science & Education*, 23(2), 465-484.
- Knippels, M. C. P. J. (2002). Coping with the abstract and complex nature of genetics in biology education: The yo-yo learning and teaching strategy. Proefschrift Universiteit Utrecht.
- Lanie, A. D., Jayaratne, T. E., Sheldon, J. P., Kardia, S. L., Anderson, E. S., Feldbaum, M., & Petty, E. M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of genetic counselling*, 13(4), 305-320.
- Lawson, A. E., & Thompson, L. D. (1988). Formal reasoning ability and misconceptions concerning genetics and natural selection. *Journal of Research in Science Teaching*, 25(9), 733-746.
- Lenartowicz, P. (1975). Phenotype-genotype dichotomy: an essay in theoretical biology.
- Lewis, J., & Leach, J. (2006). Discussion of Socio-scientific Issues: The role of science knowledge. *International Journal of Science Education*, 28(11), 1267-1287.
- Lewis, J., Leach, J., & Wood-Robinson, C. (2000a). All in the genes? – young people's understanding of the nature of genes. *Journal of Biological Education*, 34(2), 74–79.

- Lewis, J., Leach, J., & Wood-Robinson, C. (2000b). What's in a cell? - young people's understanding of the genetic relationship between cells, within an individual. *Journal of Biological Education*, 34(3), 129-132.
- Lewis, J., Leach, J., & Wood-Robinson, C. (2000c). Chromosomes: the missing link — young people's understanding of mitosis, meiosis, and fertilisation. *Journal of Biological Education*, 34(4), 189-199.
- Lewis, J., & Kattmann, U. (2004). Traits, genes, particles and information: re-visiting students' understandings of genetics. *International Journal of Science Education*, 26(2), 195-206.
- Lewis, J., & Wood-Robinson, C. (2000). Genes, chromosomes, cell division and inheritance - do students see any relationship?. *International Journal of Science Education*, 22(2), 177-195.
- Machalek, R., & Martin, M. W. (2010). Evolution, Biology, and Society A Conversation for the 21st-Century Sociology Classroom. *Teaching Sociology*, 38(1), 35-45.
- Madden, D. (2005). Time for a genetic switch? *Journal of Biological Education*, 39(3), 100-101.
- Marbach-Ad, G. (2001). Attempting to break the code in student comprehension of genetic concepts. *Journal of Biological Education*, 35(4), 183-189.
- Marbach-Ad, G., & Stavy, R. (2000). Students' cellular and molecular explanations of genetic phenomena. *Journal of Biological Education*, 34(4), 200-205.
- Mendel, G. (1866). Versuche über Pflanzenhybriden. *Verhandlungen des naturforschenden Vereines in Brunn*, 4(3), 44.
- Meyer, L. M. N., Bomfim, G. C., & El-Hani, C. N. (2013). How to understand the gene in the twenty-first century?. *Science & Education*, 22(2), 345-374.
- Nachtomy, O., Shavit, A., & Yakhini, Z. (2007). Gene expression and the concept of the phenotype. *Studies in History and Philosophy of Science Part C: Studies in History and Philosophy of Biological and Biomedical Sciences*, 38(1), 238-254.
- Norris, S. P., & Philips, L. M. (2003). How literacy in its fundamental sense is central to scientific literacy. *Science Education*, 87(2), 224-240.
- Plomin, R., Owen, M. J., & McGuffin, P. (1994). The genetic basis of complex human behaviours. *Science*, 264(5166), 1733-1739.

- Popli, R. (1999). Scientific literacy for all citizens: different concepts and contents. *Public Understanding of Science*, 8, 123–137.
- Porter, W. P. (1967). Solar radiation through the living body walls of vertebrates with emphasis on desert reptiles. *Ecological Monographs*, 274-296.
- Ratcliffe, M., & Grace, M. (2003). *Science education for citizenship: teaching socio-scientific issues*. McGraw-Hill International.
- Roseman, J. E., Caldwell, A., Gogos, A., & Kurth, L. (2006, April). Mapping a coherent learning progression for the molecular basis of heredity. In *National Association for Research in Science Teaching Annual Meeting*.
- Sadler, T. D. (2004). Moral and Ethical Dimensions of Socioscientific Decision-Making as Integral Components of Scientific Literacy. *Science Educator*, 13(1), 39-48.
- Shaw, K. R. M., Van Horne, K., Zhang, H., & Boughman, J. (2008). Essay contest reveals misconceptions of high school students in genetics content. *Genetics*, 178(3), 1157-1168.
- Smith, M. K., Wood, W. B., & Knight, J. K. (2008). The Genetics Concept Assessment: A New Concept Inventory for Gauging Student Understanding of Genetics. *CBE – Life Sciences Education*, 7, 422-430.
- Strickberger, M. W. (1968). *Genetics*. Macmillan. New York.
- Subramanian, G., Adams, M. D., Venter, J. C., & Broder, S. (2001). Implications of the human genome for understanding human biology and medicine. *Journal of the American Medical Association*, 286(18), 2296-2307.
- Thomas, J. (2000). Learning about genes and evolution through formal and informal education. *Studies in Science Education*, 35(1), 59-92.
- Tsui, C. Y., & Treagust, D. F. (2003). Genetics reasoning with multiple external representations. *Research in Science Education*, 33(1), 111-135.
- Tsui, C. Y., & Treagust, D. F. (2007). Understanding Genetics: Analysis of Secondary Students' Conceptual Status. *Journal of Research in Science Teaching*, 44(2), 205–235.
- Tsui, C. Y., & Treagust, D. (2010). Evaluating Secondary Students' Scientific Reasoning in Genetics Using a Two-Tier Diagnostic Instrument. *International Journal of Science Education*, 32(8), 1073-1098.

- Van Doorn, W. G. (1997). Effects of pollination on floral attraction and longevity. *Journal of Experimental Botany*, 48(9), 1615-1622.
- Venville, G.J., & Treagust, D. F. (1998). Exploring conceptual change in genetics using a multidimensional interpretive framework. *Journal of Research in Science Teaching*, 35, 1031–1055.
- Wojczynski, M. K., & Tiwari, H. K. (2008). Definition of phenotype. *Advances in genetics*, 60, 75-105.
- Williams, M. S. (2001). Genetics and managed care: Policy statement of the American College of Medical Genetics. *Genetics in Medicine*, 3(6), 430-435.
- Wood-Robinson, C. (1994). Young people's ideas about inheritance and evolution. *Studies in Science Education*, 24, 29-47.
- Wood-Robinson, C. (1995). Children's biological ideas: knowledge about ecology, inheritance and evolution. *Learning science in the schools: Research informing practice*, 111-131.
- Wood-Robinson, C., Lewis, J., & Leach, J. (2000). Young people's understanding of the nature of genetic information in the cells of an organism. *Journal of Biological Education*, 35(1), 29-36.

## Appendix I – Questionnaire (Dutch)

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# WAT IS EEN ERFELIJKE EIGENSCHAP?

---

Beste Leerling,

Je doet mee aan een onderzoek naar leerlingdenkbeelden over het begrip ‘erfelijke eigenschap’. Dit onderzoek bestaat uit drie onderdelen welke je van voor naar achter dient te doorlopen.

In onderdeel A wordt je gevraagd om een definitie te geven van het begrip ‘erfelijke eigenschap’ en vijf voorbeelden hiervan te noemen. Vervolgens volgt in onderdeel B een lijst met 33 menselijke eigenschappen waarop je bij elke eigenschap dient aan te geven of je wel of niet denkt dat dit een erfelijke eigenschap is of dat je hier over twijfelt. Het is belangrijk dat je naast dit antwoord ook je uitleg hierbij geeft! Aan het einde, in onderdeel C, wordt je nogmaals gevraagd om een definitie te geven van het begrip ‘erfelijke eigenschap’ omdat dit mogelijk veranderd is nadat je onderdeel B hebt gemaakt.

Het is belangrijk om van voor naar achter te werken en je eerste ingeving op te schrijven. Ga dus niet terug naar een eerdere vraag om je antwoord te wijzigen nadat je idee hierover is veranderd.. Je wordt er niet op beoordeeld, de gegevens worden anoniem verwerkt.

Naam leerling: .....

Naam school: .....

Naam docent: .....

Klas: .....

Leeftijd: .....

Geslacht: jongen / meisje



## Onderdeel A

*Dit onderdeel is bedoeld om vast te stellen wat je verstaat onder het begrip ‘erfelijke eigenschap’ voorafgaand aan het onderzoek. Schrijf het antwoord meteen op. Aan het einde van de vragenlijst krijg je deze vraag nog eens.*

Wat is een ‘erfelijke eigenschap’?

.....

.....

.....

.....

.....

Geef vijf voorbeelden van erfelijke eigenschappen:

1. ....
2. ....
3. ....
4. ....
5. ....

Einde onderdeel A

## Onderdeel B

Hier volgt een lijst met 33 menselijke eigenschappen waarop je bij elke eigenschap dient aan te geven of je wel of niet denkt dat dit een erfelijke eigenschap is of dat je hier over twijfelt. Het is belangrijk dat je naast dit antwoord ook je uitleg hierbij geeft! Zeker als je 'Nee' of 'Ik twijfel' hebt ingevuld. Daarnaast is het belangrijk dat je de lijst van voor naar achteren doorloopt, ga niet terug naar een eerdere eigenschap om je antwoord te wijzigen als je idee hierover is veranderd.

1. *Natuurlijke haarkleur.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat

.....

.....

2. *Natuurlijke haarvorm, zoals krullend of steil.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat

.....

.....

3. *De kleur van de iris.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat

.....

.....

4. *Een lichte of donkere huidskleur.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat

.....

.....

5. *Het kunnen maken van een gootje met je tong.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

6. *Het hebben van hamerduimen, waarbij het topje van de duimen kort en rond is en gepaard gaat met een korte en brede nagel.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

7. *Het hebben van een wipneus.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

8. *Kleurenblindheid, het onvermogen om kleuren te onderscheiden.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

9. *Cystische Fibrose (CF), beter bekend als 'taaislijmziekte', een ziekte waarbij patiënten moeite hebben met ademen en een grote kans hebben op longontstekingen.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

10. *Syndroom van Down, een aangeboren afwijking die gepaard gaat met een verstandelijke beperking, veroorzaakt door een derde kopie van chromosoom 21.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

11. *Hemofilie, een ziekte waarbij bepaalde stollingsfactoren ontbreken in het bloed waardoor het bloed niet stolt wanneer een bloedvat beschadigd raakt.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

12. *Phenylketonurie (PKU), een ziekte waarbij phenylketon zich ophoopt in het lichaam door een defect leverenzym 'phenylalanine hydroxylase' (PAH).*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

13. *Syndroom van Gilbert, een stofwisselingsziekte wat hoge concentraties van bilirubine, een afbraakproduct van hemoglobine, in het bloed veroorzaakt.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

14. *Een aangeboren hersenafwijking door zuurstoftekort bij de geboorte.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

15. *Het vermogen om glucose te kunnen verbranden.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

16. *Het zelf herstellend vermogen van de huid.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

17. *Het bezit van ledematen.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

18. *Secundaire geslachtskenmerken, welke zich pas tijdens de puberteit ontwikkelen.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

19. *Geboren worden met een 'open ruggetje' (spina bifida), een ontwikkelingsstoornis waarbij de neurale buis die tijdens de embryonale ontwikkeling ontstaat niet goed sluit, ook wel het 'neuralebuisdefect' genoemd.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

20. *Grote oren van ouderen, aangezien deze blijven groeien gedurende een mensenleven.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

21. *Een muzikaal talent hebben.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

22. *Goed zijn in sport.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

23. *Een hoog intelligentie quotiënt (IQ) hebben.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

24. *Een hoger risico op borstkanker door een mutatie in het BRCA-1 gen.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

25. *Het krijgen van huidkanker door te veel blootgesteld te zijn aan UV-licht.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

26. *Het hebben van een afwijkend hemoglobine type door het bezit van het sikkelcel allel, zonder het hebben van symptomen van de ziekte.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

27. *Een lagere hoeveelheid van een bepaalde neurotransmitter, geassocieerd met een grotere kans op agressief gedrag.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

28. *Lactose intolerantie op een jonge leeftijd, wat darmproblemen veroorzaakt wanneer melkproducten worden ingenomen, maar wat op latere leeftijd vaak verdwijnt.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

29. *Geboren worden met het 'foetale alcohol syndroom'.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

30. *Ziekte van Huntington, een ziekte gekenmerkt door de afbraak van zenuwcellen in bepaalde delen van de hersenen wat de motoriek, het verstand en gedrag beïnvloed. De symptomen begin vaak rond het 30-40<sup>e</sup> levensjaar.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

31. *Afwijkende slijmproductie in de longcellen van patiënten met Cystische Fibrose (CF).*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

32. *De afwezigheid van een bepaald pigment type in het netvlies van mensen die lijden aan kleurenblindheid.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

33. *Hoge productie van pigment in de huidcellen van mensen van Afrikaanse afkomst.*

Denk je dat dit een erfelijke eigenschap is?

- Ja
- Nee
- Ik twijfel

Omdat .....

.....

## Onderdeel C

Wat is een 'erfelijke eigenschap'?

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Einde onderdeel C

Ontzettend bedankt voor je deelname! 😊

## Appendix II – Coding scheme (Dutch)

Code	Wanneer	Voorbeeld bij 'Ja'	Voorbeeld bij 'Nee'	Voorbeeld bij 'Twijfel'
oud	Vergelijking tussen ouders en kinderen	Wordt overgedragen. Kind vaak zelfde als ouders. Fam. Overgedragen/erfelijk.	Geen verband tussen ouder en kind. Is niet erfelijk. Wordt niet overgedragen.	Niet zeker of het erfelijk is / overgedragen wordt. Niet zeker of ouders met [...] ook kinderen met [...] krijgen.
geb	Vanaf de geboorte aanwezig	Is vanaf de geboorte aanwezig.	Is niet vanaf de geboorte aanwezig.	Niet zeker of het vanaf de geboorte aanwezig is.
dna	Concept: DNA/gen/chromosoom/allel	Ligt vast (in het DNA/gen/chromosoom/allel/erfelijk materiaal) Genetisch bepaald.	Ligt niet vast in het DNA/gen/chromosoom/allel/erfelijk materiaal. Heeft niets met genen te maken.	Niet zeker of het vast ligt DNA/gen/chromosoom/allel/erfelijk materiaal.
mut	Concept: mutatie	Wordt veroorzaakt door een mutatie/fout	Wordt veroorzaakt door een mutatie.	Niet zeker wanneer mutatie is ontstaan. Niet zeker of mutatie alleen in lichaamscellen of ook in geslachtscellen voorkomt.
omg	Omgevingsfactoren	Wordt niet beïnvloed door omgevingsfactoren.	Wordt beïnvloed door omgevingsfactoren. Kan je trainen.	Niet zeker of het ook door omgevingsfactoren wordt beïnvloed. Niet zeker of je het ook kunt aanleren/trainen.
ken	Kennis	Heb ik gelezen/gehoord/etc	Heb ik gelezen/gehoord/etc.	Omdat ik het niet weet/ken. Geen idee.
con	Constant	Eigenschap veranderd niet.	Dit veranderd bv. met leeftijd.	Omdat dit later weer verdwijnt.
var	Variatie	Zijn verschillen tussen mensen	Iedereen kan/heeft het.	Omdat iedereen dit kan/heeft.
pers	Persoonlijke ervaring	Omdat dit in mijn familie voorkomt.	Omdat ik iemand ken die dat heeft.	Mijn moeder heeft het en ik niet.
deels	Deels wel maar...	Deels erfelijk, maar wordt ook beïnvloed door omgevingsfactoren.	Deels erfelijk, maar wordt ook beïnvloed door omgevingsfactoren.	Deels erfelijk, maar wordt ook beïnvloed door omgevingsfactoren.
vaag	Te vage uitleg / onduidelijk			
overig	Geen van bovenstaande			

## Appendix III – Overview categories and examples used in Part B

- C1 Clearly visible traits (a) versus not-visible traits (b)**
- a.
    2. Natural hair shapes, such as straight or curly.
    3. The colour of the human iris.
    4. Having a light or dark skin.
  - b.
    11. Haemophilia, a disease in which certain clotting factors are missing in the blood, leading to continuous bleeding after a vessel is damaged.
    12. PKU, Phenylketonuria, a disease in which phenylketon builds up in the body due to a defect in the hepatic enzyme phenylalanine hydroxylase (PAH).
    13. Gilbert's syndrome, an enzymatic disorder leading to high levels of bilirubin in blood.
- C2 Traits on organism level (a) versus traits on lower organisational levels (b)**
- a.
    4. Having a light or dark skin.
    8. Colour blindness, the inability to distinguish colours.
    9. Cystic fibrosis (CF), also known as mucoviscidosis, a disease in which patients have difficulty breathing and are more at risk for lung infections.
  - b.
    31. Abnormal mucus production in lung cells of patients with Cystic fibrosis (CF).
    32. The absence of a pigment type in the retina leading to colour blindness.
    33. High production of pigment in skin cells in people from African origin.
- C3 Traits which are independent from environmental factors (a) versus traits which are heavily influenced by the environment (b)**
- a.
    1. Natural hair colour.
    3. The colour of the human iris.
    5. The ability to fold your tongue.
  - b.
    21. Having a musical talent.
    22. Being good at sports.
    23. Having a high IQ score.
- C4 Traits with a high variance within a population (a) versus traits with no variance within a population (b)**
- a.
    2. Natural hair shapes, such as straight or curly.
    3. The colour of the human iris.
    4. Having a light or dark skin.
  - b.
    15. The ability to metabolize glucose.
    16. The ability of the skin to repair itself.
    17. The possession of limbs.
- C5 Static traits (a) versus dynamical traits (which change over time) (b)**
- a.
    3. The colour of the human iris.
    6. Having 'clubbed thumbs', the tips of the thumbs being short and round with a short and wide nailbed.
    7. Having a tip-tilted nose.
  - b.
    18. Sexual characteristics which develop during puberty.
    20. Older people having larger ears as they keep on growing during life-time.
    28. Having lactose intolerance at a young age, which results in bowel problems when dairy-products are ingested. However, at when they grow older this often disappears.
    30. Huntington's disease, a neurodegenerative disorder which affects muscle coordination and mental decline and behavioural symptoms, physical symptoms usually begin at the age of 30-40.
- C6 Traits on organism level (a) versus traits on lower organisational levels which influence the chance or sensitivity to a certain trait or disease (biomarkers) (b)**
- a.
    2. Natural hair shapes, such as straight or curly.
    3. The colour of the human iris.
    4. Having a light or dark skin.
  - b.
    24. Having a higher risk on breast cancer caused by a mutation in the BRCA-1 gene.
    26. Having a deviant haemoglobin type caused by a Sickle cell allele without having disease symptoms.
    27. Having a lower level of a certain type of neurotransmitter associated with a higher chance on aggressive behaviour.