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The Status of the Concepts 'Hereditary Trait' and 'Phenotype' in Secondary School Textbooks

Research Setting:

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Abstract:

In this time of rapid genetic knowledge development, students' genetic understanding is still problematic in secondary education. The current emphasis in genetics education on Mendelian segregation and single-gene inheritance models lacks the richness of today's scientific understanding and may lead to a poor understanding of genetics and a deterministic view on genetics. Many factors in this problem have been identified, but it is likely that also student understanding of the genetic concepts *hereditary traits* and *phenotype* is problematic and much narrower than in biological science. Textbooks are an important factor contributing to students' understanding. In this study, the two major Dutch secondary school biology textbooks were analysed and use of the terms *hereditary traits* and *phenotype* was classified according to six categories in which the concepts were expected to differ from scientific use. It was found that only part of the aspects of these concepts as described in the categories were covered in the textbooks and that conceptual variation and inconsistent use of the terms may contribute to problematic genetic understanding. Most of the textbook examples of *hereditary traits* and *phenotype* were about simple genetically determined Mendelian monogenetic characteristics. The next generation therefore might be insufficiently prepared for future decisions on genetics related problems.

Contents

1. Introduction:

Citizens of the 21th century live in an era of rapid genetic development. Examples of relevant issues in the field of genetics are discussions about cloning, genetic testing, gene patents, assisted reproduction (SCST, 2000), jurisdiction in cases of genetically and environmentally disadvantaged suspects (Feresin, 2009), selection of embryos for male or female sex (Valkenberg, 2014), genetic modified food, testing for risk traits in sports, and headlines in newspapers about genes for traits such as alcoholism (Sadler, 2004). To cope with these issues effectively, citizens should have a basic understanding of genetics and an ability to make informed decisions in political or personal issues. A particular group of citizens who should have these skills are secondary school students, who are the next generation with the task to cope with genetic information and applications responsibly and effectively (Jennings, 2004).

An important prerequisite for such genetic literacy is correct understanding of genetic concepts. When people have a poor understanding of genetic terminology and concepts, this has dangers such as misunderstanding of information from health care providers (Lanie et al., 2004). Central and strongly interrelated concepts in genetics are *environment*, *genes*, *hereditary traits* and *phenotype*. From extensive studies on students' understanding of genetics it became clear that correct and functional understanding of genetics is difficult for students, which is at least partly due to an incomplete understanding of genetic concepts (Shaw, Van Horne, Zhang, & Boughman, 2008; Gericke et al., 2014; Knippels, Waarlo, & Boersma, 2005).

Many studies have been done specifically on scientific use and educational use and understanding of the concept *gene* and how the use and understanding of this concept can be improved in education (Portin, 1993; Boujemaa et al. 2010; Flodin, 2009). However, scientific use of the concepts *phenotype* and *hereditary traits* has been only minimally studied, for example by Nachtomy, Shavit, and Yakhini (2007) and by Mahner and Kary (1997). Nachtomy et al. concluded that gene expression profiles could also be regarded as phenotypes (see also paragraph 6.1). Mahner and Kary found that at the time of the study the current scientific state of the key concepts *genome*, *genotype* and *phenotype* was far from satisfactory. Very few studies about educational use and understanding of the concepts phenotype and hereditary trait are known, for example by Gericke and Hagberg (2010b). Gericke and Hagberg found that the concept *hereditary traits* is very complex and that it depends on which model organism is used and on which biological organizational level the trait is described. Further, they found that the terms *hereditary trait* and *phenotype* are ill defined in textbooks and that there is conceptual variation (diversity of meanings) in the use of the terms, though the textbooks do not take the resulting incompatibility of the terms explicitly or implicitly into account. These findings indicate that the use of the terms *phenotype* and *hereditary trait* can be problematic in secondary education. Research about genetic concepts in secondary education indicates that students' conceptions of hereditary

traits is limited to outer visible traits like eye colour, hair colour and skin texture (Wood-Robinson, Lewis, & Leach, 2000). Further findings about educational use of the terms are discussed in paragraph 3.5 of the theoretical basis.

According to concept learning theories, students have a prototype in their mind for each concept – an image that captures the essence of the concept (Woolfolk, Walkup & Hughes, 2008). As the students' prototype of *hereditary trait* seems to be limited to outer visible and static characteristics, students might be unable to use the concept for situations in which other traits than outer visible traits are treated. Further, as the flexibility of the use of a concept in new situations depends on a clear understanding of the concept (Perkins & Salomon, 1992; Van Oers, 1987; Vosniadou & Ioannides, 1998), poor definition of the terms *hereditary trait* and *phenotype* (as indicated by Gericke & Hagberg, 2010b) might contribute to this inability.

Correct educational translation of the scientific scholarly knowledge of the terms *phenotype* and *hereditary trait* to students is important for at least two further theoretical reasons and two practical reasons.

First of all, as Gifford (2000) indicates, *genes* and *traits* are interdependent concepts: 'A *conceptual issue about how to describe and classify traits can have relevance to, and even be a prerequisite for, the conceptual question of what counts as a gene* (…) *in various contexts, genes and traits codefine each other'*[1] (This will be further discussed in the theoretical basis). Thus, clarity about the concept of the gene enables clarity of the concept of traits, while at the other side, confusion about the one causes confusion about the other. Therefore, good understanding of the term *traits* is necessary for a good understanding of the term *gene* and other related genetic and biological concepts. Gould & Lewontin (1979) for example indicated in their paper about adaptationism that the question "What is a trait?" is a vital issue in understanding adaptation.

Secondly, meaningful learning (as opposed by *rote* learning) requires clearness of concepts (Novak & Canãs, 2008), thus meaningful learning about genetics requires clearness of the concepts *hereditary trait* and *phenotype*. Otherwise, as it is the case in rote learning, concepts are not related to other concepts and previous knowledge and thus create a lacuna in genetic literacy.

There can also be practical consequences of an incomplete or incorrect understanding of the concept of a *hereditary trait* or *phenotype*: Chamany, Allen, & Tanner (2008) for example showed that a weak experience of physicians to identify hereditary traits at both the molecular and organismal level led to a public conflation of the carrier status of sickle cell anaemia with the trait of having sickle cell anaemia proper in Columbia. The problems of secondary school students resulting from incomplete or incorrect understanding of the terms could include unnecessary fear for getting familial diseases.

[1] Gifford, *Gene concepts and genetic concepts*, p. 54 - 56

Finally, as textbooks are major sources of what is learnt about genetics in the classroom, it is important for textbook writers and curriculum developers to monitor whether recent scholarly knowledge about *hereditary traits* and the *phenotype* is sufficiently covered in educational textbooks.

In the process of translation of scientific scholarly knowledge to students, a number of actors can be distinguished: science, textbooks, teachers and students (Gericke & Hagberg, 2007). Since textbooks are major sources for knowledge learnt in the classroom (Gericke et al., 2014; Knippels, Waarlo, & Boersma, 2005), especially for biology teaching, these are the best candidates for studying how genetic terms are presented to the next generation citizens (Gericke, 2009).

2. Research aim

The aim of this research is to analyse the use of the terms *phenotype* and *hereditary traits* in secondary school biology textbooks in order to infer the risk of problematic understanding of the terms *phenotype* and *hereditary traits*.

In short, a *hereditary trait* is a physical or mental characteristic of an organism transmissible from parent to offspring (Kenneth, 1963). The terms 'character' or 'characteristic' are synonyms of the term 'trait' (King & Stansfield, 2002). In this paper, the term 'trait' will be used. The *phenotype* is originally the total of all hereditary traits of an organism, thus every organism has its unique phenotype (Johannsen, 1909). Though the terms *phenotype* and *hereditary trait* are often used interchangeable, both in scientific and educational settings (Shaw, Van Horne, Zhang, & Boughman, 2008), there is a small content difference between the words. Further, there are a number of genetic terms closely related with *hereditary trait* and *phenotype* that are relevant to discuss as well. Both issues will be discussed in the theoretical basis.

This study consists of two parts. In the first part, different definitions and attributes of *hereditary traits* or *phenotypes* as used in biological and medical research will be categorized and described. Secondly, the two major Dutch secondary school biology textbooks for upper secondary education will be analysed based on the developed categorization. Both textbook definitions of the terms hereditary trait and phenotype and textbook examples of hereditary traits will be compared with attributes and definitions used in actual biological research.

3. Theoretical basis

In this theoretical basis, the linguistic differences and relations between the concept 'phenotype' and the concept 'hereditary trait' are discussed. Secondly, the differences and relations between the concepts 'phenotype' and 'genotype' and between 'gene' and 'hereditary trait' are discussed. Third, educational definitions of the concepts 'phenotype' and 'hereditary traits' as found in scientific literature about education are listed and discussed. Finally, it is described which conceptual problems can theoretically arise in the educational use of the concepts 'phenotype' and 'hereditary trait'. This theory is used for the structure of the analysis of the textbooks.

3.1 Terms used in defining a concept

A number of linguistic terms are relevant to describe differences between the definitions and meaning of the concepts *phenotype* and *hereditary trait*. These terms, which will be discussed hereafter, are the word *attribute* and the word-pairs *hypernym/hyponym* and *holonym/meronym.*

a) Attribute

According to the mathematical lattice theory (Wille, 1982), a concept consists of a set of objects and a set of attributes. The objects all share certain attributes, but they also have attributes not unique for the concept. For example, all birds (objects) share the attributes feathers, toothless beaks, hollow bones, perching feet, wishbones, deep breastbones and stump-like tailbones (Padian, 1998), but not all birds have the attribute 'flight' or 'long beak'.

In this research on the concepts *hereditary trait* and *phenotype*, it is important to distinguish attributes shared by all hereditary traits and those used for only a specific subset of hereditary traits. Further, it is important to distinguish explicitly described attributes of hereditary traits and attributes implicit in examples of hereditary trait (Ray Bareiss, 2014).

b) Hypernym/hyponym

Some concepts are a hypernym[2], a word which encompasses the meaning of multiple mutually semantically related words. For example, the concept 'bird' is a hypernym encompassing the meaning of amongst others the concepts 'passerine' and 'eagle'. 'Passerine' and 'eagle' are encompassed by the hypernym 'bird' and are thus called hyponyms[3] (Fromkin, Rodman, & Hyams, 2007).

In this research, it is important to make a distinction between the concept 'hereditary trait' in general (thus as a hypernym) and specific hereditary traits like blue eyes. 'Blue eyes' is thus a hyponym of 'hereditary trait'.

[2] From Greek ὑπέρ, 'over' and ὄνομα, 'name'
[3] From Greek ὑπό, 'under' and ὄνομα, 'name'

A concept refers to objects, 'bird' for example refers to e.g. 'passerine' and 'eagle', thus to a collection of hyponyms. The whole collection of all birds, thus of all hyponyms of *bird*, is called the *extension* of the concept *bird*. Another word for the extension of a concept is *category* (Ray Bareiss, 2014), but because of practical reasons, in this paper the term *extension* will be used. Thus the extension of the concept *hereditary trait* is the whole collection of all hereditary traits.

In this research, it is attempted to map the extension of *hereditary trait* for its range today, thus beyond simple Mendelian traits.

c) Holonym/meronym

Another term to describe concepts is *holonym*[4], which is a word for the whole of parts. For example, the concept 'hand' is a holonym for 'finger'. 'Finger' is part of the hand and is thus called a meronym[5].

In this research, it is important to distinguish between *phenotype* as the whole of hereditary traits, thus *phenotype* is a holonym for *hereditary trait.* As described in 3.2, however, *phenotype* can also be used as a synonym to *hereditary trait.*

3.2 How are the concepts 'phenotype' and 'hereditary trait' related?

In the same sense as the concept *bird*, the concept *hereditary trait* consists of distinct hereditary traits (hyponyms) like 'blue eyes', 'height' and 'a heart' and of attributes like 'inheritable via DNA in germ cells' and 'outward visible'. All hereditary traits share the attributes 'inheritable via DNA in germ cells', but not all hereditary traits are outward visible. It should be noted here that the term *genetic traits* also occurs in genetic vocabulary to denote hereditary traits (for example Knippels, Waarlo, & Boersma, 2005), but as this term is not as common as term *hereditary trait*, this term will not be discussed in this paper.

The term *phenotype*[6] has originally been coined by Wilhelm Johannsen together with the word *genotype* in 1909 (Schulze & McMahon, 2005; Dunn, 1965). Johannsen states that the phenotype of an individual is the sum of all his hereditary traits and that also the environment plays a role in the establishment of the phenotype of an organism; he defines a phenotype of an individual as '*the sum total of all of this expressed characters*. *The single organism, the individual plant, an animal, a man, "What he is and what he does," [wie er geht und steht] has its phenotype, i.e. he appears as a totality of characters which are determined by interplay between "inherited predispositions [Anlage] and elements of the environment*"'[7].

[4] From Greek ὅλον, 'whole', and ὄνομα, 'name'
[5] From Greek μέρος, 'part' and ὄνομα, 'name'
[6] From Greek φαίν-ομαι, 'to appear' (Johannsen, 1909) or φαίνω (Johannsen, 1926)
[7] Dunn, *A Short History of Genetics,* p. 92

Because the concept *phenotype* was originally used to describe the total of an organism's hereditary traits, *phenotype* is a holonym of all hereditary traits. However, in the course of time, scientists started to use the term *phenotype* also for specific characteristics either of a class of organisms or of individual organism instead of only for the whole of an organism's characteristics. Formulations like 'collagen phenotype' (Benya & Shaffer, 1982), or of a 'cardyomyocyte phenotype' (Bird et al., 2003; Toma, Pittenger, Cahill, Byrne, & Kessler, 2002) are abound. Even 'the expression level of a gene' is called an 'inherited phenotype' (Cheung & Spielman, 2002). Thus, the concept *phenotype* has become a synonym of *hereditary trait* in addition to its use as a holonym. It is well possible that if students have problems with the terms *phenotype* and *hereditary trait*, these are partly due to the described changed conceptual relations.

3.3 How are the concepts 'phenotype' and 'genotype' related?

Originally, the concept *phenotype* paralleled that of the genetic complement *genotype* (Gifford, 2000; Mahner & Kary, 1997). Johannsen originally used the term *genotype* for the whole of genes and defined it as the constitution of the two gametes that unite to produce the individual (Dunn, 1965). The phenotype was defined as the result of the interplay between the genotype and environmental factors, thus as the sum of all traits of an organism. However, according to Mahner and Kary (1997), scientists later started to use the term *genotype* for specific allele pairs either of classes of organisms and of individual organisms. This shift has been confirmed by philosopher Richard Lewontin (2011): *'In practice, genotypic and phenotypic descriptions are not total but partial, restricted to some subset of the characteristics of the organism that is regarded as relevant for a particular explanatory or experimental purpose'*[8].

In 1920, professor of botany Hans Winkler introduced the term *genome* to denote the totality of genes (Lenartowicz, 1975). This can be seen as a result of the use of *genotype* for specific allele pairs: a new term was needed to describe the whole of allele pairs. Even the Human *Genome* Project reflects such a use of the term *genome*. As equivalent of the term *genome,* the term *phenome* was coined, which is defined as '*physical totality of all traits of an organism or of one of its subsystems'* (Mahner & Kary, 1997).

If we compare the use of the term *genotype* with the use of the term *phenotype,* we can basically identify a shift from the use of the terms *genotype* respectively *phenotype* first from the whole of genes respectively traits, and secondly replacement of the terms *genotype* and *phenotype* as descriptors of wholes by the terms *genome* and *phenome.* It is interesting to note that indeed a field of phenomics has arisen as an equivalent of the field of genomics (Houle, Govindaraju, & Omholt, 2010). Further, we can conclude that there is much *conceptual variation,* which is *'the range of different historical/scientific meanings that*

[8] http://plato.stanford.edu/archives/sum2011/entries/genotype-phenotype/

a concept might have'[9] of the terms *genotype* and *phenotype.* This complicates the task of selecting definitions of the terms for educational use and it is well possible that students' problems with the terms *phenotype* and *hereditary trait* as far as present, are therefore partly due to the described conceptual variation.

3.4 How are the concepts 'hereditary trait' and 'gene' related?

As 'hereditary trait' and 'gene' are the meronyms of the respectively 'phenotype' and 'genotype' original concepts, they exhibit the same relation as the phenotype to the genotype: hereditary traits are established through the interaction of genes and the environment. In the early 20th century, in the context of breeding, genes were regarded as the causes of phenotypic differences, like smooth versus wrinkled pea texture. However, though single genes can indeed make the *difference* between certain traits, they cannot be held responsible for the establishment of the individual trait. In fact, the relation between individual traits and genes is many-to-many: one gene can influence many traits (called pleiotropy) and one trait is often coded for by many genes (Dupré, 2012). Still, the formulation *'gene for'* suggesting a one-to-one relation between a gene and a trait lingers in science and it the media. For example, Thomas (2000) describes that media reports announced the findings of genes for speech, baldness, dyslexia, asthma, faithfulness, better motherhood, athletic prowess, snoring and even suicide. In fact, the term 'gene' in these contexts is confused with for example a quantitative trait locus (QTL), which is in fact a stretch of DNA linked to or containing genes underlying a trait (Crusio, 2002). Another problem of the formulation 'gene for' is the fact that many scientists describe the mutation or absent gene causing a disease as 'the gene for' the disease, while in fact, the cause of an abnormality is a mutation or multiple mutations in the allele and not the allele itself (Dupré, 2012). Further, if an abnormal trait is known to be correlated with a certain allele, like 'the allele for diabetes', the normally functioning allele is sometimes erroneously regarded as 'the allele for normal blood sugar regulation' while in fact, a number of alleles are involved in healthy blood sugar regulation. This can be illustrated with the fact that there are many possible mutations that can cause blood coagulation problems, but that the absence of such a mutation in an allele does not mean that the allele itself is the only allele for blood coagulation. This can be also be explained with the analogy of a Jenga tower game. In this game, players have to build a tower from wooden bricks and subsequently players have to remove bricks without letting the tower fall. The first one who removes a brick with the result of the fall of the tower has lost. Though one brick may be responsible for the fall of the tower because the brick is removed, this does not mean that that single brick is the only constituent of the tower: all bricks are involved! The same is true for abnormalities: normally functioning alleles should not be held responsible for the normal or healthy equivalent of an abnormal trait (Dupré, 2012).

[9] Gericke, *Science and School Science*, p. 17

3.5 Definitions, hyponyms and attributes of 'phenotype' and 'hereditary traits' in education

A small literature study has been performed for this research, indicating that students in previous decades used the definitions *hereditary trait* for only a few specific types of hereditary traits. Pupils' definitions of phenotypic traits were often about physical traits, for example *'what people look like'*[10], eye colour, hair colour, skin texture (Wood-Robinson, Lewis, & Leach, 2000) or *'what it looks on the outside'* (Venville & Treagust, 1998). However, some studies report that students also mentioned psychological traits like intelligence (Knippels, Waarlo, & Boersma, 2005). However, Lewis & Wood-Robinson (2000) found in a research with 14-16 year olds that only about 8 percent of the respondents thought of mental, emotional, or behavioural traits while the majority referred to physical traits when thinking of characteristics determined by the DNA. Thus, it is likely that psychological traits are not prevalent among students' conceptions of hereditary traits. According to Knippels et al., some students also regarded height as hereditary trait. Though height is in fact a trait that can change during development, it is likely that the students meant 'adult height'. After all, little attention is paid to continuous variation in genetics education (Jiménez-Aleixandre, 2014). Lewis and Kattmann (2004) suggest that students regard hereditary traits as heirlooms, which are transferred like goods from generation to generation.

According to Gericke & Hagberg (2010b), secondary school textbooks mainly focus on one-to-one relation between genes and functions or traits, while this relation is, as discussed before, in fact many-to-many. It is likely that one-to-one relations mainly occur in examples of Mendelian traits because these are traits of which the differences can indeed be caused by (mutations in) single genes (Dupré, 2012).

Gericke & Hagberg (2010b) further reported about definitions of *trait* in educational textbooks. They found the following definitions (literature sources already present in Gericke & Hagberg, 2010b):

1. Particular characteristic or feature of an organism (Evans et al., 2005a, p. 481)
2. One genetically determined characteristic of an organism (Leonard and Penick, 2003, p. 562)

These are very vague definitions which are not likely to add anything to the knowledge of learners about the concept *hereditary trait* except maybe reinforcing the association of traits with genetic determination. It can thus be expected that students' definitions of phenotypic traits are mainly discrete, physical, genetically determined, outer visible traits. This can be confirmed with the conclusion of Dougherty (2009) that mostly rare, single-gene (Mendelian) traits are taught in secondary education*, 'leaving students with the mistaken assumption that this fully describes inheritance'*[11] which results in the fact that '*many*

[10] Wood-Robinson, *Young People's Understanding,* p. 31.
[11] Dougherty, 2009, p. 10

students view all phenotypes through the lens of Mendelian inheritance'[12]. More about Mendelian traits and their present state in science can be found in paragraph 6.3.

Only very little data are available of what students regard as *phenotype*. Gericke & Hagberg (2010b) found that the relationship between the *phenotype* concept and the *trait* concept is generally avoided in educational textbooks. However, they found the following definitions of *phenotype* (literature sources already present in Gericke & Hagberg, 2010b):

1. A certain set of traits (Henriksson, 2007a, p. 42)
2. A specific set of traits. (Karlsson et al. 2007, p. 172)
3. The observable form of a characteristic or trait (Evans et al. 2005a, p. 313)

Also these definitions are very vague; only the last definition suggests that observability is an important attribute of the concept *phenotype*.

3.6 How is a concept learned in education?

According to learning psychologist Joyce, four components are crucial in learning new concepts: examples (or: *exemplars*) and non-examples, relevant and irrelevant attributes, the name of the concept and a definition (in: Woolfolk, Walkup & Hughes, 2008). A non-example is for instance: a bat can fly but is a mammal and thus not a bird, therefore it is a non-example of a bird. Both adults and children can, with sufficient domain knowledge, generalize a concept (that is, formulate the attributes essential to the concept) on the basis of one example of the concept. They can thus infer a hypernym from the attributes of a hyponym. When they are encountered with new examples of the concept, their generalizations of the concept are determined predominantly by their original generalization and only partly by the attributes of the new examples. When more examples are provided, the attributes of the new examples play a bigger role in determining the attributes essential of the concept (Carmichael & Hayes, 2001).

According to concept learning theories, students form, as soon as they learn a concept, a prototype in their mind for each concept – an image that captures the essence of the concept (Woolfolk et al., 2008). This prototype is developed when new examples of the concept are provided to the learner. For young children, the prototype of the concept 'bird' might be, for example, a robin. It is important to note that the robin prototype lacks many attributes of birds like hawks, ducks and ostriches.

For learning the concept *hereditary trait,* this would mean that students with enough knowledge of genetics can formulate the attributes essential to the concept on the basis of examples of hereditary traits provided to them. However, as already described in the

[12] Dougherty, 2009, p. 7

introduction, students seem to hold traits like blue eyes as prototype of concept *hereditary trait,* which suggests that many attributes and hyponyms of the concept *hereditary trait* as it is used in science, are not known or not clear to them.

Scientific examples of hereditary traits reveal that there are many other attributes relevant to the concept hereditary trait in general or to specific groups of hereditary traits, thus to the complete extension of *hereditary trait* or to part of the extension of this concept. Further, many specific hereditary traits relevant in science do not have (all) the attributes of the students' blue eyes prototype, for example, many traits relevant in science are not outer visible. Students might not realize that the attributes of their prototype of *hereditary trait* are valid only for a limited set of *hereditary trait* hyponyms.

In analogy to the robin prototype example, which lacks attributes like the sharp claws of hawks, the flightlessness of ostriches and the swimming ability of ducks, students thus might miss important attributes of specific groups of hereditary traits. In addition, they might lack the insight that the attributes of traits like blue eyes, like their outer visibility, do not apply to all hereditary traits. To obtain a more complete understanding of the concept *hereditary trait,* students would have to be provided multiple examples of hereditary traits having attributes not present or not clear in the blue eyes prototype.

Woolfolk et al. (2008) suggest that when complicated concepts are taught (or when the concepts are taught to younger or less knowledgeable people), more examples should be provided than for simpler concepts. Further, non-examples should be provided to draw the boundaries of the concept and its extension. For example, discussing why a bat is not a bird aids pupils in defining the boundaries of the concept 'bird'.

For learning the concept *phenotype,* the case is complicated. For the original use of the term as a holonym, students would just have to have a good understanding of the concept *hereditary trait,* as hereditary traits are the parts of someone's phenotype. However, as the term *phenotype* is now also used as synonym of *hereditary trait*, students would have to develop the concept *phenotype* in the same way as the concept *hereditary trait.* This would cause much confusion because students would then have to judge every time they recognize the term *phenotype* whether it is used as holonym of *hereditary trait* or as synonym. Thus, part of the textbook analysis will be how they treat this conceptual variation of the term *phenotype.*

As it is thus clear that examples, non-examples as well as attributes and definitions are important in learning concepts, the textbooks in this study have been analyzed for examples and non-examples, relevant and irrelevant attributes and definitions of *phenotype* and *hereditary trait*.

4. *Research Question*

In order to find out whether the use and definition of the terms hereditary trait *and* phenotype *in secondary school textbooks enables a flexible use of these terms in new situations, textbook content and scientific knowledge concerning these terms should be compared. The research question therefore is:*

To what extent do biology textbooks cover the current scientific definitions, attributes and hyponyms of the concepts 'hereditary trait' and 'phenotype'?

This research question is divided in four sub-questions:

a) What attributes and hyponyms of the concepts *hereditary trait* and *phenotype* can be distinguished in actual science?

b) What definitions of the concepts *hereditary trait* and *phenotype* can be distinguished in actual science and what attributes or hyponyms do they reveal?

c) Which of these attributes and hyponyms are represented in secondary education biology textbooks and to what extent?

d) What definitions are used in secondary education biology textbooks for *hereditary trait* and *phenotype?*

5. *Material and Methods*

In this section, it is described what material and methods were used to answer each of the sub-questions of the research question.

a) What attributes and hyponyms of the concepts *hereditary trait* and *phenotype* can be distinguished in actual science?

To inventorise the relevant attributes and hyponyms of the concepts *hereditary trait* and *phenotype*, four genetics dictionaries from different centuries have been studied as well as a number of scientific and philosophical books and articles that are specific about the concepts *hereditary trait* and *phenotype* or about evolutionary or genetic concepts in general. The books which were used are 'Conceptual Issues in Evolutionary Biology' (Sober, 2006), 'The Book of Man: The Human Genome Project and the Quest to Discover Our Genetic Heritage' (Bodmer, 1997) and 'Processes of Life' (Dupré, 2012). The articles which were used are by Lenartowicz (1975) about the *genotype/phenotype* distinction, Mahner & Kary (1997) about *phenotype* and *genotype* definitions, Churchill (1974) about the *genotype* concept, Violle (2007) about the *trait* concept for plants, Nachtomy, Shavit, & Yakhini about gene expression and the concept of the *phenotype* and Houle, Govindaraju, & Omholt (2010) about *phenomics.*

Attributes and hyponyms as explicitly formulated in texts or as implicit in examples of *hereditary traits* or *phenotypes* are listed in 6.1. It is the aim of this research to find potential problematic attributes and hyponyms in educational texts, therefore attributes and hyponyms used in science (as found in the analysed books and articles) different from or supplementary to the educational attributes and hyponyms (as described in paragraph 3.5 of the theoretical basis) were categorized in six categories. These categories are listed in 6.2.

b) What definitions of the concepts *hereditary trait* and *phenotype* can be distinguished in actual science and what attributes or hyponyms do they reveal?

To obtain a broad overview of definitions of *hereditary trait* and *phenotype*, the same books and articles were studied as for a). It was checked whether the categories as described in 6.2 also occurred in the definitions. The found definitions were listed in a table and can be found in paragraph 6.3a and 6.3b. In the right column, the corresponding categories of attributes or hyponyms are indicated with a 'C' followed by the number of the category. The books and articles have also been searched for non-examples of hereditary traits, which result can be found in paragraph 6.3c.

c) Which of these attributes and hyponyms are represented in secondary education biology textbooks and to what extent?

The categories of attributes as described in a) were used as a framework for the educational textbook analysis.

The newest version of the textbooks of two Dutch biology methods have been studied which are available as traditional books (*Nectar* and *Biologie voor Jou*). *Biologie voor Jou* is used on 60% of Dutch secondary schools while *Nectar* is used on 30%[13]. The textbooks of both havo and vwo level (which are the highest levels of secondary education) have been used. Only books from the upper secondary level of havo and vwo have been analysed as the concept of genetics is introduced and further developed in this stage.

The *Biologie voor Jou* upper level series consists of four havo books and six vwo books (two books per grade) while *Nectar* consists of two havo books and three vwo books. The vwo6 book of *Nectar* is still in press, thus an available draft version was used for the 6vwo book. The two vwo6 books of *BvJ* are not already available and therefore have not been analysed.

The textbooks were analysed by inventorizing all hereditary traits or phenotypes (in the synonym version of this concept, see paragraph 3.2). A table has been constructed in which every found trait that in one way or another is linked to a genetic basis was listed. The criterion for whether a trait x is linked to a genetic basis was the use of formulations such as 'gene for *x*', '*x* lies in the genes', '*x* is genetically determined', 'allele for *x*', '*x* is hereditary', '*x* is inborn'. In addition, traits listed in twin research tables can be interpreted as hereditary and are thus included. Traits which were described as being probably hereditary, epigenetic

[13] Source: anonymous

or non-hereditary (thus not with certainty hereditary) were listed and analysed as described under d) of this paragraph. Also traits or phenotypes in captions of images, questions and additional or extra (non-compulsory) chapters were included. If a trait appeared twice in one subparagraph, the trait was listed only once. However, if a trait appeared more often in a paragraph (but not in one subparagraph), the trait was listed for occurrence in each subparagraph. In the case that the being hereditary or not of traits discussed in questions was unclear, the answers book (only available for *BvJ*) was consulted. For traits in questions in *Nectar* books, the questions were answered with an educated guess.

The inventorized hereditary traits were scored for whether they were called *phenotype* or not. Further, they were categorized in 'outer traits', 'diseases/anomalies', 'internal regulation', 'structural elements', 'psychological/behavioural characteristics' or 'other traits'. This distinction is important to find out whether textbook examples of traits are indeed restricted to outer traits. The categorization is a modification of the categories used by Chantal de Ruijter (2009). Finally, for every trait it was checked which attributes as described in the theoretical framework applied to it.

An interrater reliability analysis using the Kappa statistic was performed to determine consistency between the main rater and the second rater in categorizing the traits (Landis & Koch, 1977).

These results can be found in paragraph 7.2.

d) What definitions are used in secondary education biology textbooks for *hereditary trait* and *phenotype?*

To inventorise the definitions used in the textbooks, the books were all precisely read and found definitions were listed. Further, non-examples of hereditary traits or phenotypes (in the synonym version of this concept, see paragraph 3.2) as found in c) were analysed for their information about what is a *hereditary trait* or *phenotype* indeed. In addition, traits marked as probably hereditary were analysed for their information about the definitions of *hereditary trait* or *phenotype.* Finally, information in the books explicitly about attributes and hyponyms of *hereditary trait* and *phenotype* was inventoried. These findings can be found in paragraphs 7.1 and 7.2.

6. Results - I

In this section, it is first described what attributes and hyponyms of 'hereditary trait' and 'phenotype' – as far as these are not present in the educational prototype of simple monogenetic Mendelian traits – were found in scientific books, papers and dictionaries.

After that, the differences between the found educational and scientific definitions are integrated in a category framework and further described.

Finally, it is described what definitions were found in these studied scientific books, papers and dictionaries and whether they reveal the same attributes and hyponyms as found in the first section of this chapter.

6.1 Hyponyms or attributes (implicit in examples or explicit) in scientific texts

In this section, attributes and hyponyms of *hereditary trait* explicitly mentioned or found in particular examples of hereditary traits or phenotypes (used in the synonym version) are discussed. Corresponding attribute categories they provide information about, are indicated in bold after each example.

Nachtomy, Shavit, & Yakhini (2007): In their paper about gene expression and the concept of the phenotype, philosopher Nachtomy and his colleagues argue that the phenotype encompasses phenomena at different organizational levels. Thus cancer, for example, applies at cellular level, tissue level or organ level. The attribute that traits have entities on different organizational levels will be further discussed in **C2.** Further, the writers state that also invisible traits can be regarded as phenotypic traits. According to them, even mRNA profiles can be regarded as such. They quote Darvasi who writes: '*Height and weight are typical examples, but it is perfectly reasonable to consider gene expression levels as a quantitative trait, too.'* The notion that also invisible traits and mRNA expression patterns (both hyponyms) can be hereditary traits will be further discussed in respectively **C1** and **C6**.

Dupré (2012): In his book about the philosophy of biology, philosopher John Dupré argues that the assumption that traits are determined by genes is problematic. He shows that there are many environmental conditions that influence traits; someone's length of six feet can be arrived at by different combinations of genes interacting with environmental influences. The attribute of *hereditary trait* that also environmental factors play a role in the establishment of traits is discussed in **C3.**

Bodmer (1997): In his book about the Human Genome Project, Bodmer describes that genes have been found which determine, though in a complex way, intelligence, personality and the manufacture of haemoglobin, insulin and human growth hormone. The notion that the manufacture of haemoglobin is a hereditary trait indicates that traits that do not have differences within a population are also hereditary traits. This hyponym of *hereditary trait* is

also discussed by Dupré (2012) who states that in the strict Mendelian sense, there are no genes for universal traits. This hyponym will be further discussed in **C4**.

Sober (2006): In their paper about evolutionary psychology, philosophers Tooby & Cosmides (2000) provide a list of attributes of adaptations. As adaptations are defined as 'phenotypic properties' that were present and relevant during the species' evolution, this list is as relevant for traits as for adaptations. They write that it is worth noting that traits ('phenotypic properties') can develop at specific times during the life cycle, for example teeth or breasts. This indicates that traits can change, which attribute is discussed in **C5**.

Houle, Govindaraju, & Omholt (2010): In their paper about the emerging field of phenomics, Houle and his colleagues argue that a biomarker is an endogenous phenotype that can be measured and used as an indicator of other biological processes, predominantly of diseases. This indicates that also endogenous entities functioning as predictors can be counted as hereditary traits: this hyponym will be further discussed in **C6**.

6.2 Categories of limited or incorrect educational use of the concept 'hereditary trait'

In this paragraph, the attributes of hereditary traits or phenotypes presumably less common in secondary education, as found in scientific definitions and examples of hereditary traits and phenotypes, are described in six categories. These categories were used for the educational textbook analysis, of which the results can be found in chapter 7. The bold part of the title of each category describes the attributes as found in scientific texts but presumably less present in educational textbooks. The left, non-bold part of the title of each category describes the narrow opposite version of these attributes as commonly found in education.

In the remainder of this paper, references to the scientific part of a category will be like *'the scientific side of category* x*'* while references to the hypothesized narrow educational part as described in paragraph 3.5 will be like *'the educational side of category x'*. These categories C1 to C6 have been used as a framework for comparison of scientific and educational textbook use of the terms hereditary traits and phenotype.

C1

Outer visible traits vs. also **non-outer visible traits**

Secondary school students seem to restrict themselves to outer visible and 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).

C2

Traits on organism level vs. **traits on multiple organizational levels**

Secondary school students do not seem to realize that traits do have features on other organizational levels than the organismal level (Duncan & Reiser, 2007) and biological textbooks make little notion of biological organizational levels (Knippels, 2002; Gericke 2010b). Scientists, on the contrary, 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, Rogat, & Yarden, 2009). Sociobiologists add psychological and behavioural levels to these (Machalek and Martin, 2010; Goldberg, 1993; Strickberger in Churchill, 1974). The disease cystic fibrosis, for example, has features on tissue-, organ- and organism level (Dupré, 2008; Duncan, Rogat, & Yarden, 2009; Duncan & Reiser, 2007).

C3

Environment-independent traits vs. **partially environment-dependent traits**

Students consistently distinguish physical traits, which they regard as hereditary, and character traits like temperament, which they attribute to environmental factors (Thomas, 2000). Further, many students doubt that genes play any role in personality, addiction and cardiovascular efficiency. They ascribe these traits to environmental factors like peer groups, will power and exercise (Dougherty, 2009). 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). In addition, sportsmanship and musicality are hereditary traits that are established by interplay 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). 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.[14]

C4

Traits with variance within a population vs. **traits without variance within a population**

[14] In a study by Knippels (2005, p. 111) it was found that students of the control group of a small study who only had a basic genetic knowledge also realised that traits are determined by the environment as well. However, as it could be that they regard environmental factors as only important for non-Mendelian traits, it is still possible that most students do not consciously know that environmental factors play a role in every trait.

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. According to Dougherty (2009), '*students' world of genetics is one of either/or traits, rather than quantitative characters (…) discrete traits commonly discussed in genetics courses.*[15]

C5

Traits which remain the same vs. **traits which change**

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). Even the blood type of organisms can change (Dean, 2005). 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 (continuous traits) as a hereditary trait. They are more likely to think of discrete traits (Jiménez-Aleixandre, 2014). 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 (Sinnott in Lenartowicz, 1975). In this category, two kinds of changing traits could be distinguished: discrete traits which appear (generally without disappearing) at a certain moment or during a specific timespan in an organism's development (like teeth, secondary sex characteristics and the development of lactose intolerance) and continuous traits which are abstractions of traits and which can change during development (like hair and eye colour).

This category is thus not about traits which are supposed to be changed during evolution (like single cycle blood circulation to double cycle blood circulation).

C6

Traits on organism level vs. **traits on lower organizational levels which influence the chance of or susceptibility to getting certain traits or diseases**

[15] Dougherty, 2009, p. 7

Another aspect in which the content of the concept 'hereditary traits' or 'phenotype' can differ between science and education is the whether measurable internal biological components which are not outer visible and which indicate the chance of getting a disease or other trait is regarded as hereditary trait or not. The chance of getting a disease is sometimes measured by looking at increased or decreased levels of components like prostate-specific antigen (PSA) levels in the blood. Component levels which are known to be hereditary are called endophenotypes, while the term 'biological marker' is used for any (thus also visible and non-hereditary) measurable indicator of a disease (Wojczynski & Tiwari, 2008; Lenzenweger, 2013). Gottesman & Gould (2003) state that an endophenotype can be neurophysiological, biochemical, endocrinological, neuroanatomical or cognitive. According to Nachtomy (2007), even the mRNA expression profile of the BRCA-1 gene involved in breast cancer (Miki et al., 1994) 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 or endophenotypes, it is well possible that they do not regard endophenotypes or biological markers which are not DNA themselves, as hereditary traits.

It is important to note that sometimes the term intermediate phenotype is used for the endophenotype (Winawer, 2006); however, as this term is also used for phenotypes resulting from incomplete dominance alleles (Lenzenweger, 2013), in this paper the term endophenotype will be used.

6.3 Definitions of phenotype and hereditary traits in scientific texts

Since no current general accepted definitions of the terms hereditary trait and phenotype could be found in literature, a collection of definitions – ranging from historical to modern – of the terms have been derived from scientific literature and dictionaries. These findings are presented and discussed below and found trait attributes and hyponyms implicit in the definitions are marked with the corresponding category number of the framework in paragraph 6.2.

***a) Phenotype* definitions**

In literature, a number of different definitions of *phenotype* were found. Most of them reflect either the holonym version of the term phenotype or the synonym version of the term. Some of them mention both uses. The term phenotype is thus used for the whole of traits or for specific traits; however sometimes also to characterize an individual. For example, a *Caenorhabditis elegans* worm displaying a helically twisted body roller mutation can be called a roller worm (Cox, Laufer, Kusch, & Edgar, 1975). This use can be found in the definition by Sinnott (in: Lenartowicz, 1975). Further, the term can be used for a class of organisms that share the same phenotype, the offspring of two crossed identical hybrids AaBb x AaBb for example: while nine genotypes can be distinguished in the offspring, only four phenotypes can be distinguished. This use can be found in the definitions by Knight (1948) and Kenneth (1963).

Mahner and Kary (1997) summarize the common usages of the term phenotype:

1) *the set of trait types of an organism or of one of its subsystems*
2) *an organism characterized by a certain (usually partial) phenotype*
3) *a class of organisms sharing the same (usually partial) phenotype*

In table 1, definitions of *phenotype* as found in the dictionaries, books and articles that are mentioned in the Material and Methods section are displayed. As related versions of the term *phenotype*, like *phenotypic, phene* and *phenome*, also provide information about the meaning of *phenotype*, these terms are displayed as well. Attributes of traits as revealed in the phenotype definitions are made bold and in the right column, a 'C' followed by a number corresponding with a certain category as described in paragraph 6.2.

Table 1. Phenotype definitions and definitions of related terms.

	Concept	*Category*
	Phenotype	
Knight (1948) p. 114	*A group of individuals of similar appearance but not necessarily of similar genetic constitution.*	
Kenneth (1963) p. 436	*The characters of an organism due to the response of genotypic characters to the **environment**; a group of individuals exhibiting the same phenotypic characters.*	***C3***
Churchill (1974) p. 6	*From: Strickberger (1968): 'The phenotype refers to all the manifold biological appearances, including **chemical, structural and behavioral attributes**, that we can observe about an organism but excludes its genetic constitution. The genotype defines only the particular genetic material that an organism inherits from its parents. Therefore, although the phenotype **changes with time** as the appearance of the organism changes, the genotype remains relatively constant except for the rare genetic changes known as mutations.'*	*C1* *C5*
Lenartowicz (1975) p. 30	*From: Sinnott et al. (1958): 'The sum total of all characteristics, such as color, form, size, behavior, chemical, composition and structure, **both external and internal**, gross and microscopic. (...) The phenotype of an individual **changes with time**, as illustrated, for example, by a series of photographs of a person taken at different ages from infancy to senility. But we also know that the more subtle physiological changes constantly occur in an individual, so that the phenotype is never exactly the same from one moment to the next. (...) We recognize persons or individuals of any species of animal, or plant, by their phenotypes.'*	*C1* ***C5***
MacLean (1987) p. 301	*Expressed characteristics of an organism, resulting from the activity of the genotype modified by the **influences of the environment**. Although originally the term was taken to mean the observable character (i.e., shape, form, colour, behaviour) it is now used in a more extensive sense to include both **microscopic character and molecular character**. Thus the primary aspect of the phenotype is the selection of **proteins synthesized** at the*	***C3*** ***C1*** ***C6***

	*instigation of the genome, and the **grosser aspects of the phenotype result from the structural or enzymic properties of these proteins**.*	***C2***
Mahner & Kary (1997) p. 55 - 56	*From: Suzuki et al. (1989): '(i) The form taken by some character or group of characters in a specific individual (ii) outward manifestation of a specific genotype.'* *From: Rieger et al. (1991): 'The observable properties (structural and functional) of an organism'* *From: Hartl et al. (1987): 'The observable properties of an organism'* *From: Futuyama (1986): 'The manifestation (...) of the interaction of this [genetic, DvD] information with the physical and chemical factors – the environment in the broadest sense – that enable the blueprint to be realized.'* *From: Dobzhansky et al. (1977): 'Its [an organism's, DvD] appearance – its morphology, **physiology**, and ways of life - what we can observe.'* *From: Lewontin (1992): 'a **consequence** of the development of that individual from a zygote through a historical sequence. That sequence is a result (...) of the previous state of the organism, of its genotype, and of the environment in which it is developing.'* *From: Herskowitz (1977): 'Collection of traits possessed by a cell or organism that results from the interaction of the genotype and the **environment**.'*	***C1*** ***C5*** ***C3***
King & Stansfield (2002) p. 295	*The observable properties of an organism produced by the genotype in conjunction with **the environment**. In a more restricted sense, phenotype is used for the effect a gene produces, in comparison with its mutant alleles, on the morphology of the organism in which it resides. Some genes control the behaviour of the organism, which in turn generates an artefact outside the body. R. Dawkins used the term extended phenotype to refer to the production of such an artefact (spider webs, bird nests, and beaver dams are examples).*	***C3***
	Phenotypic	
Knight (1948) p. 114	*Appertaining to the physical make-up of an organism or group of organisms as distinct from their genetic make-up. The phenotypic effect of any particular gene on an organism is its outward, measurable, quantitative or qualitative effect on that organism.*	
Kenneth (1963)	*Pert.* Phenotype, *appl.* Characters arising from reaction to ***environmental stimulus***.	***C3***
	Phene	
MacLean (1987) p. 301	*Phenotypic character (see PHENOTYPE) controlled by genes. Note however, that in fact all 'genetically determined' characteristics are more accurately understood as resulting from an interaction of gene expression and **environmental influence**.*	***C3***
King & Stansfield (2002) p. 295	*A phenotypic character controlled by genes.*	
	Phenome	
Mahner & Kary (1997) p. 56	*From: Lewontin (1992):'The actual physical manifestation of the organism, including its morphology, **physiology** and behaviour.'*	***C1***

b) Hereditary trait definitions

In table 2, definitions of *hereditary trait* as found in the dictionaries, books and articles that are mentioned in the Material and Methods section are displayed. As most sources defined *hereditary* and *trait* separately, definitions of these terms are displayed separately in the table. As related versions of the term *hereditary trait*, like *character, heredity* and *hereditary disease* also provide information about the meaning of *hereditary trait*, these terms are displayed as well. Attributes of traits as revealed in the *hereditary trait* definitions are made bold and in the right column, a 'C' followed by a number, which is specific for each type of attribute. In the text following the table, the meaning 'hereditary' and a number of different types of hereditary traits (continuous, discrete, inborn, congenital and innate traits) as come across during the literature analysis are discussed.

Table 2. Hereditary trait definitions and related terms definitions

	Concept	*Category*
	Character/Characteristic	
Knight (1948) p. 24	*The phenotypic result of the interaction of a gene or group of genes and the **environment**.*	***C3***
King & Stansfield (2002) p. 61	*Any detectable phenotypic property of an organism; synonymous with phenotype, trait.*	
	Hereditary	
Knight (1948) p. 67	*(i) In the clinical sense: transmitted with unbroken continuity from generation to generation. (ii) In the genetic sense: controlled by a genetic mechanism which is capable of being transmitted from generation to generation although the outward signs of the presence of this mechanism may only be apparent under specific conditions.*	
Kenneth (1963) p. 242	*Transmissible from parent to offspring, as characteristics, physical or mental.*	
	Heredity	

Knight (1948) p. 68	*The transmission or parental qualities, expressed or latent, to the progeny.*	
Kenneth (1963) p. 242	*The organic relation between successive generations; germinal constitution.*	
MacLean (1987) p. 185	*Phenomenon whereby characteristic traits are transmitted from one generation to another. It results from the transmission of genes from parents to offspring. Patterns of inheritance within a group of progeny result from the assortment of such genes in the GAMETES and ZYGOTES.*	
King & Stansfield (2002) p. 61	*A familial phenomenon wherein biological traits appear to be transmitted from one generation to another. The science of genetics has shown that heredity results from the transmission from parents to offspring. The genes interact with one another and with their environment to produce distinctive characteristics or phenotypes. Offspring therefore tend to resemble their parents or other close relatives rather than unrelated individuals who do not share as many of the same kinds of genes.*	
	Hereditary disease	
MacLean (1987) p. 185	*Pathological condition resulting from the expression of a gene or genes, usually a mutant allele of a normal gene, or the lack of a gene or genes.*	
King & Stansfield (2002) p. 179	*A pathological condition caused by a mutant gene.*	
	Trait	
MacLean (1987) p. 391	*Particular phenotypic property of an individual, especially if expressed only to a slight degree. Thus individuals heterozygous for the sickle cell haemoglobin allele are said to have SICKLE CELL TRAIT.*	
Violle et al. (2007) p. 884	*Any morphological, **physiological** or phenological feature measurable at the individual level, **from the cell to the whole-organism level**, without reference to the environment or any other level of organization.*	***C1*** ***C2***

Jablonka & Lamb (2005) identified three dimensions of heredity other than only via DNA: epigenetic, behavioural and symbolic. According to them, *'heredity is through genes and other transmissible biochemical and behavioural entities'*[16]. Recent research showed that also epigenetic configurations and methylations of DNA which do not change the primary structure of DNA itself but still account for part of individual's traits, can be passed on to next generations. Because of these mechanisms even environmentally acquired characteristics like fear can be transmitted to offspring. Thus, in this sense, acquired characteristics *can* be hereditary. A young mammal, for example, can develop a preference for a specific type of food after it receives information about it through the milk. An example of behavioural inheritance is that a bird can learn its young to use a specific nesting site.

[16] Jablonka, Evolution in Four Dimensions, p. 356

Further, wealth and education can be transmitted from human parents to their children (Dupré, 2012). The discussion about the existence of extra-genetic inheritance is also relevant for evolutionary theory, which probably should include it in its fundamental concepts (Laland et al., 2014). However, as this paper is about genetic concepts, only genetic inheritance will be discussed.

Third, it should be noted here that hereditary traits can be continuous or discrete. According to Wojczynski & Tiwari (2008), discrete traits are traits that are either present or absent, for example hypertension and arthritis. Continuous traits (also called quantitative traits) are traits that have a range of possible values, for example arm length and weight (Dougherty, 2009).

Another important notion is that traits can be congenital, inborn or innate. The word *congenital* is used in medicine and law, especially for defects (Xie, 2002; Fernald, 1986). Congenital traits are present at birth, but not necessarily hereditary (Knight, 1948; Henderson, 1963; King & Stansfield, 2002). Errors occurring during foetal development, which are not inherited via DNA from the parents, like abnormal limbs due to drug use by the mother, are non-hereditary congenital traits. The term 'inborn' is mostly used for genetically determined biochemical disorders already present at birth that have pathological consequences (King & Stansfield, 2002). Thus inborn errors are in general hereditary. Finally, innate is a synonym for inherited (Knight, 1948; Henderson 1963), thus an innate trait is generally hereditary.

c) Non-examples of hereditary traits

To specify the boundaries of this concept, it is useful to identify what traits are regarded as *not* hereditary. Generally, hereditary traits are opposed to *acquired* traits (Lenartowicz, 1975). These are traits which are induced by the environment upon an organism (such as a lengthened neck due to wearing brass rings by a Kayan tribe woman) during its life and which are not hereditary because they do not change DNA in the germ line. In fact, in this case of *acquired* characteristics, two criteria separate hereditary from non-hereditary traits: a trait is only hereditary when (i) DNA is involved and (ii) it can be passed on through the germ line. Considering the first criterion, this implies that hereditary traits are always genetic traits, which are traits at least partly determined by DNA, but genetic traits are not necessarily hereditary. This is because some genetic traits inhibit the organism bearing the trait from reproducing. The second criterion reveals that a mutation in a somatic cell causing a genetic disorder is not hereditary, while a mutation in a germ cell causing a disorder is hereditary indeed.

7. Results – II

In the first two paragraphs of this section, the definitions, explicitly mentioned attributes and non-examples of the terms hereditary trait *and* phenotype *as found in the Biologie voor Jou and Nectar are presented and discussed. In paragraph 7.3, the examples the terms* hereditary trait *and* phenotype *as found in the Biologie voor Jou and Nectar are presented and it is discussed how many of these examples exhibit scientific use of the terms as described in the C1 to C6 categories.*

In *Biologie voor Jou*, in total (havo plus vwo books) 542 examples of traits were identified, in some cases explicit non-hereditary (*n* = 13), possibly hereditary (*n* = 25) or epigenetic (*n* = 7) but in the major part of the cases (*n* = 497) explicit or implicit hereditary. Most examples and definitions of hereditary traits and phenotypes were found in the havo4a chapter 'Heredity' and in the vwo 4b chapter 'Genetics'.

Nectar contained 301 instances of traits, of which 6 non-hereditary and 3 possibly hereditary. The other 292 instances were about traits hereditary indeed. In some cases, examples of traits were found in textbook questions[17], but as no answers book was available for Nectar, these questions have been answered with an educated guess and the answers were used for this results section. Most examples and definitions of hereditary traits and phenotypes were found in the 'Heredity' chapter in the havo5 and vwo4 chapter 'Heredity'.

In total, the number of hereditary traits found in the textbooks is *n* = 789.

The first paragraph of this section is about definitions of the concepts *phenotype* and *hereditary trait* and closely related concepts in the textbooks. As non-examples define the boundaries of concepts (Woolfolk, Walkup & Hughes, 2008), the found examples of non-hereditary and possibly hereditary traits are also discussed in this section. In addition, epigenetic effects or traits that are no real phenotypic traits are discussed in the first paragraph.

The second paragraph deals with attributes of hereditary traits as formulated in the textbooks. Attributes implicit in hyponyms and hyponyms of *hereditary trait* themselves, as found in the textbooks, are presented using the six-category framework as described in paragraph 6.2.

[17] For example Nectar havo 5, p. 10: *'Explain whether the following characteristics are hereditary and/or congenital: birth weight, sucking reflex, blood type, gender'*

7.1 BvJ and Nectar definitions of hereditary trait, phenotype and strongly related concepts.

In this section, the definitions of the terms 'phenotype' as found in the Biologie voor Jou and Nectar books is listed in a table. Further, the relations to the terms 'genotype' and 'hereditary trait' are discussed. Secondly, definitions of 'hereditary trait' are discussed and the relation between traits and genes as described in the textbooks is reported about.

a) Phenotype definitions and relations to 'hereditary trait'

In table 2, the phenotype and intermediate phenotype definitions as found in the havo and vwo books of Nectar and BvJ are listed. In short, Nectar describes the phenotype as the sum of all internal and external traits while BvJ describes it as the sum of all outer characteristics of an individual (or his/her appearance), though it is not made clear whether these characteristics are necessarily hereditary or not.

Table 2. Phenotype definitions and definitions of related terms.

	Nectar	*BvJ*
	Phenotype	
Havo	*The result of the interaction of hereditary factors and the environment is the phenotype.*	*All outer observable characteristics of an individual belong to the phenotype of an individual.*
Vwo	*All your traits – the outer and the traits which have a role in the functioning of your body – together form your phenotype.* *[In a caption]: all visible and measurable characteristics of a human constitute his phenotype.*	*By the phenotype of an individual we mean the observable characteristics of an individual.*
	Intermediate phenotype	
Havo	*A phenotype which is in between the two (respectively recessive and dominant, DvD) homozygous phenotypes.*	*Both alleles for flower colour come to expression in the phenotype to some extent. Such a phenotype we call intermediary.* *[Answers book]: a phenotype in which two unequal genes both come to expression.*
Vwo	*The cholesterol level of heterozygous (for the LDLR-gene which codes for receptors, DvD) people is, for an equal diet, between that of people with a normal number of receptors and people without receptors. They have an intermediate phenotype.*	*'Both alleles for the flower color come to expression to some extent in the phenotype. Such a phenotype we call intermediate. The phenotype is a mix (in Dutch: 'mengvorm') of both alleles (of a heterozygote organism, DvD).*

However, BvJ and Nectar also use the term phenotype for individual traits, for example '*the phenotype blood type A'* and (BvJ) or 'peas *with a yellow phenotype*' and '*the phenotype freckles*' (Nectar). BvJ even equals hereditary traits and phenotype in the sentence *'when two parents with equal phenotype have offspring with a deviating phenotype, both parents are heterozygous for this trait.'* Thus though phenotype and hereditary trait are defined respectively as holonym and meronym at the beginning of the 'Heredity' chapters, they are often used as synonyms later in the books without ample or no clarification of this conceptual variation. The only information provided by BvJ is that *'the concepts 'genotype' and 'phenotype' can have to do with all traits of an individual. In the classical study of inheritance you use them for the traits of which you study the inheritance*' (havo) and *'the concepts 'genotype' and 'phenotype' can relate to all traits of an individual. Often, these concepts are used for only one or two traits of which the inheritance is studied'* (vwo). This information is likely to be insufficient to make clear that *phenotype* can also be used as a synonym to *hereditary trait*. In the Nectar book, no information about the conceptual variation is provided at all.

In one Nectar instance *'Their trait, their phenotype is intermediate'* it is not clear whether *phenotype* is used as a holonym or as a synonym *to hereditary trait.*

It can be concluded that while the Nectar definition is in accordance with scientific definitions, especially with the definition by Sinnott et al. (1958), the BvJ definition about outer characteristics does not match scientific definitions.

b) *Relation between genotype and phenotype*

In both Nectar and BvJ, the genotype-phenotype relation is explicitly described.

BvJ states that 'a great part' of the phenotype is determined by the genotype, but that the phenotype is also determined by environmental factors like light, air, moisture, temperature, diet, education, diseases and injuries. Further, it is stated that, for many traits, the genotype determines the extreme limits while the environment determines how closely these limits are approached. Twin studies reveal the share of environmental factors contributing to the trait compared with the share of the genotype to the phenotype. *Genotype*, however, is also subject to conceptual variation: while it is first defined as the 'total package of genes of an organism', it is later also used for specific alleles or allele combinations. Thus, as it is the case for *phenotype, genotype* is defined as a holonym but later also used as synonym to *gene* or *allele*.

Later in the BvJ books, *gene expression* is connected with the phenotype in the formulation 'expressed in the phenotype'. This formulation is very problematic as, due to the definition of *phenotype* as sum of outer visible traits, it is not clear whether this means that gene is transcribed and translated to a protein or that the gene product is visible in the appearance of the organism. A good example of this is the sentence in the chapter about evolution: *'a mutated allele which is dominant is directly expressed in the phenotype'*. Another example is

the formulation *'some alleles are not expressed properly in the phenotype'* which is meant to explain incomplete dominance, however, this formulation can be understood either as a fault in gene expression or as a fault on a certain organizational level resulting in the fact that the alleles do not have a proper outer effect. Thus, the choice of defining phenotype as the collection of outer visible traits is thus very problematic for correct understanding of gene expression, codominance and evolution.

Nectar states that the phenotype is the result of the interplay between the genotype (genes in the DNA) and the environment. For each trait, the share of environmental factors contributing to the trait compared with the share of the genotype is different. Examples of environmental factors are training, parents, teachers, friends and work environment. In the same way as BvJ, the term 'genotype' is used for both the totality of genes or alleles and for individual genes or alleles: 'genotype' is contrasted with the environment as determining factors for traits, but there are also phrases like 'the genotype Aa'. It is worth noting that the definition of *genome* is different between the havo and vwo version: while the havo version reads that the genome is the whole of all genes, the vwo version states that the genome is the genotype (genes on the DNA) plus *the rest of the DNA* and mitochondrial DNA. Thus also non-coding DNA is included in the genome definition.

In both books, Nectar makes clear that recessive alleles of heterozygous individuals are not visible in their phenotype, but that co-dominant alleles of heterozygous individuals are *'expressed in the phenotype'* indeed: *'The alleles I^A and I^B are both expressed in the phenotype'*. In contrast to in BvJ, the formulation *'expressed in the phenotype'* does not yield problems because the phenotype is defined as outer plus inner traits.

c) *'Hereditary trait' definitions*

Biologie voor Jou

Biologie voor Jou does not provide a definition for *hereditary traits*, though there is stated in the introduction of the chapter 'Heredity' that it is a goal to distinguish hereditary traits from non-hereditary traits. Indeed, a number of non-hereditary traits (n = 25) is mentioned in the books. Further, a number of possibly hereditary traits (n = 6) was found. These examples revealed important attributes of hereditary traits; four differences between hereditary and non-hereditary traits could be distinguished.

First of all, BvJ explicitly states that traits that are determined purely by environmental factors are non-hereditary[18] and a few examples of such environmentally determined, non-hereditary traits are mentioned in the books: *scar, nail length, leaves directed to the light, a skin with few wrinkles due to botox injections, extreme makeover* and *drooping geranium leaves.* Further, BvJ makes clear that congenital diseases can be hereditary or non-

[18] BvJ havo 4a, p. 184

hereditary. A congenital disease is only hereditary if and the disease has a genetic basis[19], thus skeletal disorder caused by maternal drugs intake is not regarded as hereditary congenital disease. Gender identity disorder is described as congenital, but *possibly hereditary* and homosexuality is described as possibly congenital because science has not reached consensus about whether these behaviours are caused by hormones or other factors.

In the vwo books, s*trong muscles due to training* is added to the list of environmentally induced non-hereditary traits, but here it is called a *modification.* It is used to illustrate that Lamarck's theory of acquired characteristics as evolutionary factors is not true because no changes in DNA are involved in these modifications[20].

Secondly, a practical aspect of inheritance is mentioned: it is explained that progeria (which is an autosomal dominant disease generally occurring after conception) is not hereditary because children with progeria die early[21]. Thus, it is not likely that the mutation responsible for progeria is inherited.

Third, in the chapter about behaviour, acquired behaviour (experience) is distinguished from innate behaviour (instinct). Instinct is regarded as largely hereditary why experience is regarded largely non-hereditary.

Fourth, a distinction is made between hereditary and epigenetic diseases. Hereditary diseases are explained as abnormalities in the DNA, while epigenetic diseases are explained as abnormalities in the activity of a gene. Such epigenetic changes or configurations can be reversed, but sometimes they are stable and hereditary. Thus, as it described that environmental influences *can* induce hereditary changes in an organism through the mechanism of epigenetics, epigenetic changes are not always hereditary, but they *can* be.

The other four trait examples marked as *possibly hereditary* do not yield further information about the demarcation between hereditary and non-hereditary: One example is '*Do you agree with the statement: 'Your whole genetic background. That is who you really are'?'.* This example is a question about a judgement and thus provides no information about BvJ's view on hereditary versus non-hereditary. The other three examples are about the ideas that the colour pattern of guppies, the canary song and eye pecking by are hereditary determined, however, BvJ mentions that these hypotheses have not been tested yet.

To summarize: Traits are regarded hereditary if they are not purely environmentally induced, i.e. thus only if some genetic base can be distinguished (though this need not to be a specific

[19] No comment is made whether this genetic error should be present in parents to be hereditary or whether such an error can also occur in the embryo stage. In fact, some hereditary syndromes, like the 22q11.2 deletion syndrome, are in most cases caused by a novel mutation occurring in the embryo stage (McDonald-McGinn, Emanuel, & Zackai, 2013).

[20] However, as epigenetic changes can be hereditary (Jablonka & Lamb, 2005), Lamarck could be true in some respects.

[21] BvJ vwo 5a, p. 118

gene). Epigenetic changes *can* be hereditary: they are hereditary only if the changes are stable and passed on via germ cells. Further, congenital diseases *can* be hereditary: they are hereditary only if they are caused by genetic defects. Finally, traits can only be hereditary if the bearer can pass them on to his or her children. It can be concluded that the definition of hereditary trait implicit in the examples of non-hereditary traits is in accordance with the earlier described scientific definitions (see paragraph 6.3b).

Nectar

Nectar does not provide a definition for *hereditary traits*, but four instances of explicitly non-hereditary traits were found in the books: lower birth weight, birth weight, brain damage due to oxygen deprivation at birth and smoking or not. These examples reveal important attributes of hereditary traits. In the first three cases, the examples have the function of distinguishing between hereditary and non-hereditary congenital traits. According to the writers, congenital diseases can be caused by hereditary factors or by non-hereditary factors like problems during pregnancy or birth (for example, oxygen deprivation during a complicated birth). An example of a congenital disease that is hereditary indeed, is the disease phenylketonuria (PKU) because this disease is based on information in the DNA.

The example of smoking or not has the function to distinguish between environmentally induced traits versus traits determined by the genotype. This distinction can already be found at the very beginning of the havo4 book; there it is described that the queen bee instinct of making honeycombs or pollen balls is innate, in contrast to learned. Among the three examples of possibly hereditary traits present in Nectar, one is about the genetic base of hand clasping; Nectar states that science has not reached consensus yet about the issue whether right or left thumb on top in hand clasping is learned or determined by genes.

The second example of a *possibly hereditary trait* is the cleft palate; Nectar just mentions without further explanation that this is possibly caused by a recessive allele. The third example is about the hypothesis that some ancestor probably had an X-chromosomal and an autosomal gene for colour vision. Both examples do not reveal much more than scientific uncertainty.

To summarize: Traits are regarded hereditary if they are based on information in the DNA as opposed to caused by environmental factors. Thus, congenital diseases *can* be hereditary: they are hereditary only if they are caused by genetic defects already present from conception. Finally, innate behaviour is hereditary as opposed to learned behaviour that is regarded non-hereditary. It can be concluded that the definition of hereditary trait implicit in the examples of non-hereditary traits is, though limited as no mention is made of epigenetic inheritance, in accordance with the earlier described scientific definitions.

c) *'Hereditary trait' relation to 'gene'*

In general, in both Nectar and BvJ, traits are regarded as caused by genes plus environmental factors like education, upbringing and friends. In addition, non-human influences like UV radiation and prevailing flu (Nectar) are mentioned. This relation is the same as that between *phenotype* and *genotype*. However, in some cases, BvJ and Nectar confuse alleles and traits, for example *'black and brown alleles'* (BvJ) or '*alleles yellow and smooth'* and '*the allele protruding teeth'* (Nectar).

'Gene for'

Both BvJ and Nectar describe disease examples with 'gene for *x*', for example *'gene for haemophilia'* or *'allele for red-green colour-blindness'*. Further, alternative alleles for '*dimpled cheek vs. no dimpled cheek*' and for *'tail vs. no tail'* are described. For the problems with such formulations, see paragraph 3.4 of the theoretical basis. Only from the DNA chapter (in the havo5 and vwo5 book) onwards, BvJ explicitly mentions that diseases are caused by the absence or malfunctioning of a gene, for example that albinism is caused by a gene defect, that the Schinzel-Giedeon syndrome is caused by a mutation in the SETBP-1 gene and that hemochromatosis is caused by a mutation in an HFE allele. However, the absence of malfunctioning of genes or alleles is not connected to dominance or recessiveness, which still leaves a gap in coherent understanding of the precise development of diseases. [22]

In the Nectar books, it is explained that Turner's syndrome is caused by monosomy and that progeria is caused by a mutation in the LMNA-gene. However, in a number of other cases, disease examples are described with 'gene for *x*', for example *'the allele for red-green colour-blindness'*. Further, in some instances, Nectar makes the mistake of ascribing the normal equivalent of a disease to the unbroken equivalent of the broken allele, for example alleles for *'normally functioning cholesterol receptors'* and *'alleles for normally functioning muscles'*. In the DNA chapter (only in the vwo6 book), Nectar explains that diseases are caused by defective alleles and after that, all diseases are correctly described as being the result of defective or missing genes or alleles. Such a formulation also occurs twice in the vwo 5 book, where it is explained that hypohydrotic ectodermal dysplasia is caused by a mutation in the EDA-1 gene and that red-green colour-blindness is caused by defective alleles. However, also in the Nectar books, the absence of malfunctioning of genes or alleles is not connected to dominance or recessiveness.

Monogenetic vs. polygenetic traits and pleiotropy.

Already in the definition of *gene,* the Nectar havo book makes clear that a gene is a stretch of DNA with the information for a trait. This definition implies that the relation between

[22] After all, many traits, including blood types and eye colours are dominant or recessive because of the absence or malfunctioning of a gene or allele (Dupré, 2012).

genes and traits is one to one. The BvJ books add to this that a gene can also code for part of a trait, thus the relation between respectively genes and traits is many (or one) to one. The Nectar vwo book defines *gene* as a coding sequence with the information for making a protein and thus avoids the question about the quantitative relation between genes and traits. However, in none of the definitions, there is room for the interpretation that one gene can code for many (parts of) traits, or *pleiotropy* (see paragraph 3.4).

It should be noted that BvJ states early in the 'Heredity' chapter that for many traits, more than one gene is involved, however, BvJ limits itself to the use of one gene for a trait when the concept of allele pairs is introduced. Only in the vwo book, BvJ rectifies this at the end of the chapter by stating that traits can also be polygenetic: '*In this theme there is looked especially at hereditary traits which are determined by one gene and sometimes the reality is a little simplified. Often hereditary traits come into being because two or (many) more gene pairs determine together one hereditary trait.*'[23]

It is interesting that the BvJ vwo book also makes mention of alternative splicing, the mechanism which enables a gene to code for different proteins.

The *Nectar* books also start with examples of one gene coding for one trait, but later they pay more attention to polygenetic inheritance: the vwo book dedicates a whole paragraph and havo book a subparagraph to polygenetic inheritance. In the havo book, it is mentioned that at least eye colour, skin colour, nose shape and hair colour are inherited polygenetic and that these traits are therefore very variable. The vwo book mentions that body length, skin colour, eczema and high blood pressure are polygenetic and adds that the variability of traits is not only determined by (multiple) genes, but also by environmental influences like sunlight. It is surprising, however, that though the havo5 book introduces the polygenetic base of eye colour; the formulation '(single) gene for eye colour' is used again later in the book.

[23] BvJ vwo 4b, p. 47

7.2 Comparison of hereditary trait examples with scientific use in C1 to C6 categories

In this paragraph, 'hereditary trait' attributes from the C1 to C6 categories as explicitly mentioned in the BvJ or Nectar books are listed. Further, the presence of the scientific side of the C1 to C6 categories in the examples is quantified.

a) Attributes as explicitly mentioned in BvJ and Nectar books

Biologie voor Jou

In the final assignment of the BvJ 'Heredity' chapter, it is written that heredity has been treated on different organizational levels (**C2**). Already early in the introductory chapter, these organizational levels are described, from small to large: molecule, organelle, cell, tissue, organ, organ system, organism, population, biocenosis (biotic community), ecosystem and biosphere. In addition, it is mentioned that organisms have emergent characteristics, which are characteristics dependent on but not present on lower organizational levels. An example of this is walking, which depends on the muscle system, the bone system and the nerve system, however, the systems on their own are not sufficient for the ability of walking. In BvJ, organizational levels play an explicit role throughout the books.

Already early in the 'Heredity' chapter, BvJ explains that the phenotype is not determined only by the genotype, but also by environmental factors and that how much each factor contributes to traits differs per trait (**C3**).

Later, it is mentioned (though in a non-compulsory part of the books) that certain traits can change during the lifetime of an organism (**C5**). This is illustrated with the fact that a black-haired person can become grey. In a second example, it is mentioned that a dominant allele for a trait can come to expression at later age, for example the allele for baldness.

C4 is explicitly mentioned in the vwo book, though not in the 'Heredity' chapter: In the introductory chapter, it is explained that certain gene sets are involved in the architecture of a fruit fly. The activity of these genes on molecular level has results on cell level, resulting in cell differentiation and the growth and development of tissues and organs.

Nectar

In Nectar, though only in the vwo version, the phenotype definition makes clear that there are also non-outer traits (**C1**). This notion does not occur in the havo definition.

Secondly, it is explicitly described in both the havo and vwo version that the environment plays a role in the establishment of traits (**C3**). The vwo book even dedicates a whole paragraph (four pages) to the nature-nurture debate.

Nectar describes in the vwo book that, despite the genome differences between humans, there are great similarities between their characteristics, for example that everyone has a

heart and lungs. Thus it is made clear that there are also traits without variance within a population (**C4**).

Further, the authors of *Nectar* write already early in the vwo Heredity chapter that traits can change during your lifetime (**C5**). Another example is the fact that children with freckles can become adults with a nearly uniform coloured skin. However, though the allele for freckles, melanin and UV-radiation are discussed as factors contributing to freckles in the vwo book, the question of why freckles predominantly manifest in ten year olds is left open.

b) Attributes as explicitly mentioned in BvJ and Nectar books

All mentioned *hereditary trait* and *phenotype* (as used in the synonym version) examples in Biologie voor Jou and Nectar have been analysed using the framework with the 6 categories as described in paragraph 6.2. In this section, it is presented what percentage of the examples in each *Nectar* and *BvJ* do exhibit the scientific side of each category. For each textbook, the percentage of positive C1 – C6 instances is calculated and displayed in figure 1 as percentage of the total number of traits (havo + vwo) found in the textbooks. Thus examples occurring in both havo and vwo book are counted twice. As examples could exhibit the scientific side of either none or more than one category, the sum of percentages of examples exhibiting scientific sides of categories is more than 100%. The trait 'Experiencing more happiness' for example, is a non-outer trait (C1), it is described on multiple organizational levels like serotonin levels and brain functioning (C2), it is described as partially environment-dependent (C3), but it is not universal in a population (C4) and it is not described as a changeable trait (C5) or as endophenotype (C6). Thus, the trait 'Experiencing more happiness' is counted as a positive example for C1, C2 and C3, but not in the categories for C4, C5 and C6.

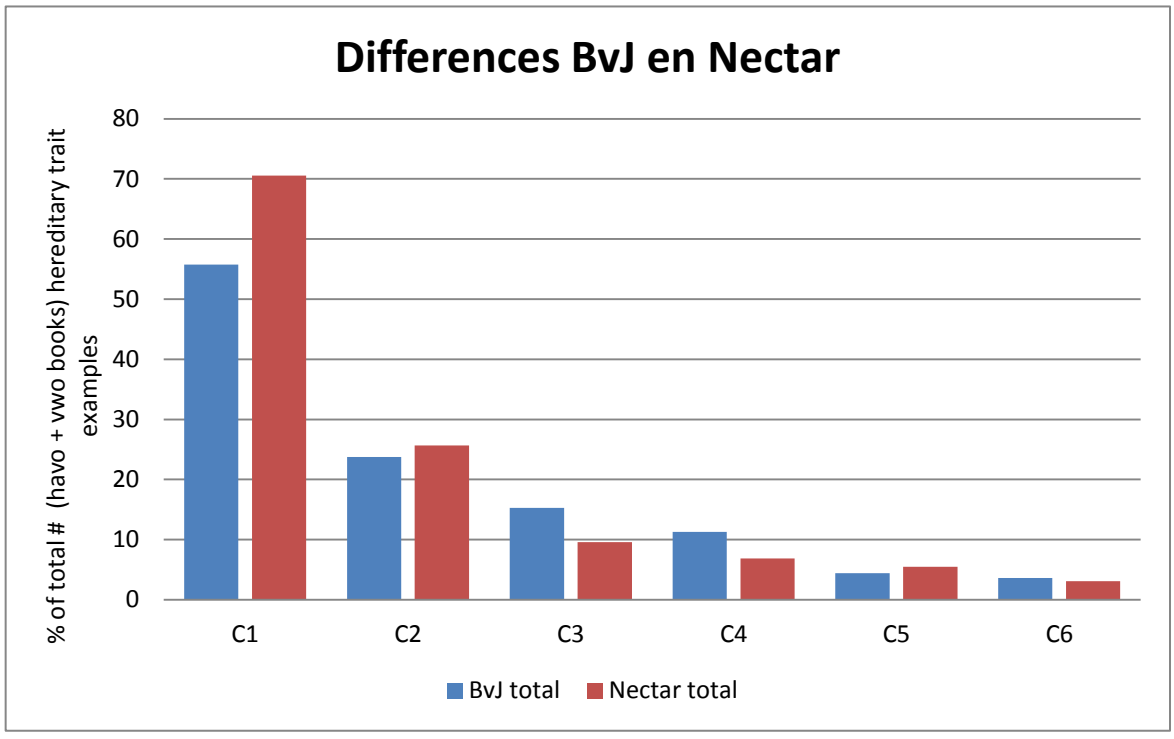

Figure 1 Traits in the C1 – C6 categories

The blue bar at the left of the figure, marked with ‘C1’, for example, means that 56% of all BvJ examples of hereditary traits were non-outer traits. The red bar marked with ‘C5’ means that 5% of all hereditary trait examples in Nectar were described as changeable.

There were slight differences between the havo and vwo versions of respectively BvJ and Nectar, which are displayed in figure 2 and 3.

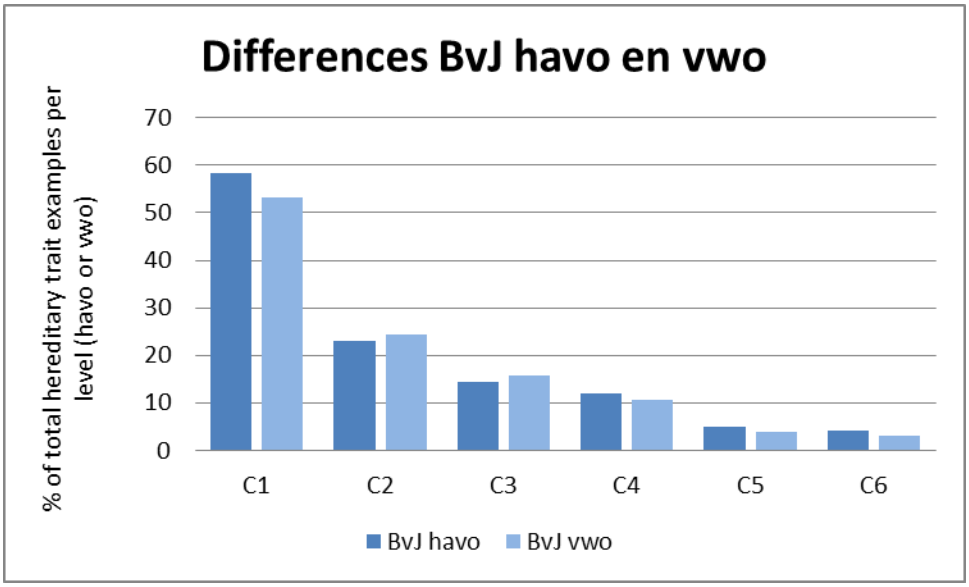

Figure 3 Different percentages between havo and vwo.

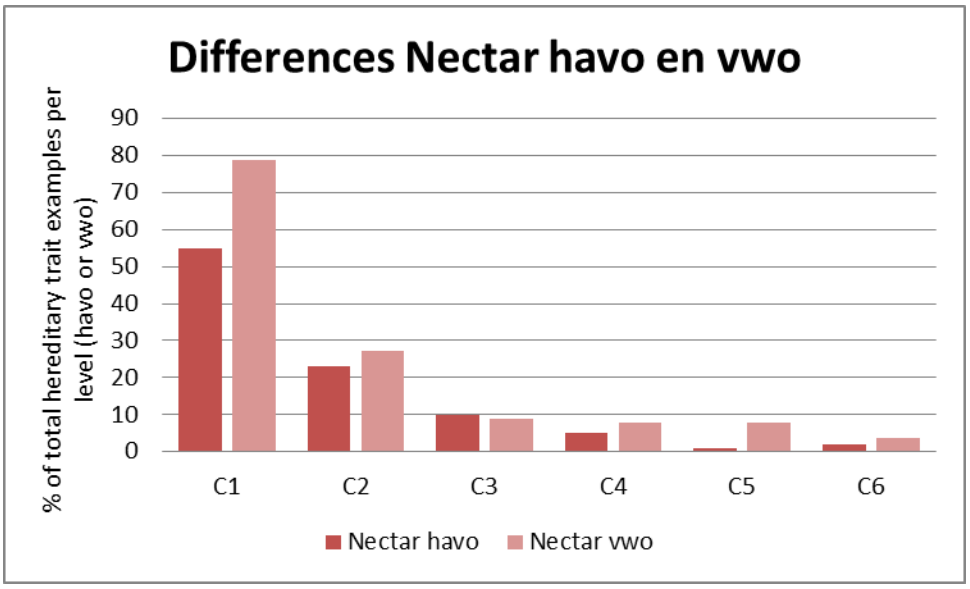

Figure 2 Different percentages between havo and vwo.

For example, in figure 3, the dark red bar means that 55% of the Nectar havo examples are non-outer while this is the case for 79% of all vwo examples (pink bar).

In the next part, it is described per category, how many examples and thus what percentage of all traits was counted as exhibiting the scientific side of the category.

C1

Outer visible traits vs. also internal traits

It was found that *Biologie voor Jou* as well as *Nectar* do not restrict themselves to outer traits, but also mention diseases or anomalies, internal regulation bodies or characteristics, structural elements and psychological/behavioural characteristics as *hereditary trait*s. 56% (*n*=277) of the Biologie voor Jou examples and 71% (*n*=206) of the Nectar examples are non-outer traits. In some instances, these traits were called *phenotype*, which has been discussed paragraph 7.1.

C2

Traits on organism level vs. also on other levels

In many instances, the *Nectar* and the *BvJ* textbooks describe hereditary traits on multiple organizational levels: *Biologie voor Jou* does this for 24% (*n*=118) of the mentioned traits while *Nectar* does this for 26% (*n*=75). As already indicated in 7.1 and 7.2, only *BvJ* explicitly mentions that traits have entities on different organizational levels. In *BvJ*, organizational levels play an explicit role throughout the books.

C3

Environment-independent traits vs. partially environment-dependent traits

Both BvJ and Nectar mention partially environment-dependent traits as examples of traits. 15% (*n*=76) of the BvJ trait examples are explicitly environment-dependent, like sense of happiness, drinking alcohol, sporting behaviour, becoming a queen bee instead of a working bee, playing the piano and personality. The example 'sense of happiness' is even already introduced at the beginning of the chapter 'Heredity' in both the havo and vwo version. 49 out of the 76 environment-dependent traits in BvJ are psychological or behavioural traits, and the other are traits are traits like alcohol drinking and becoming a queen bee. In both books, no simple 'Mendelian' traits like eye colour are described as partially environment-dependent. In all cases, Mendelian traits are described as '*gene for eye colour'*, *'allele for wrinkled pea'*, etcetera.

Traits with the formulations 'two genes *play an important role in*' or 'genes are *of interest in the case of*' are not regarded as partially environment-dependent traits as, though the mentioned formulations can indicate that the environment also plays a role, they can also indicate that multiple traits influence the specific trait. In some cases, a degree of how strong the genetic base of a trait is compared to the environmental influences was mentioned, mostly in the context of twin research outcomes. These cases, including 'weight' and 'height' (vwo), were all counted as partially environment-dependent.

Further, it is indicated in that the courtship ritual of a stickleback fish is largely determined by hereditary factors, while the piano playing of a girl is largely determined by learning processes. Thus according to the BvJ books, the genetic base for the courtship ritual is much stronger than the genetic base for piano playing. However, there is an inconsistency as innate behaviour like sucking on

a breast is mentioned as fixed in the genotype, while on the next page, it is stated that sucking on a breast is *largely* determined by hereditary factors.

In the analysed **Nectar** books, 10% (n=28) of the trait examples are described as partially environment-dependent traits, for example aggressiveness, fear, drinking alcohol, character, creativity, shyness, numeracy skills and playing the piano. 8 out of 11 (havo) and 6 out of 17 (vwo) environment-dependent traits in Nectar are psychological or behavioural traits. In the havo books, the others are playing the piano well, hairdressing and cholesterol level, while in the vwo books; these are skin colour, eye colour, hair colour, asthma, blood pressure and cholesterol level. It is worth noting that in the vwo book, the complex interaction of genetic and environmental factors is described also for seemingly simple single-gene Mendelian traits in the case of *eye colour, hair colour and skin*. It is even stated that *'it is no much use to draw a crossing design of eye colour'*. In the vwo4 book, it is even described that the gender of chickens (which obviously seems to be genetically determined) highly depends on the environmental temperature during the egg stage. Thus here it is made explicit that environment-independence of traits is at least very rare. However, in the vwo book it is stated that at least blood type is determined exclusively by the genotype.[24]

C4

Traits with variance within a population vs. traits without variance within a population

Both BvJ and Nectar mention traits for which no variance is known as examples. In BvJ 11% (*n*=56), these are traits like sharp cat teeth, oxygen transport, searching behaviour for dogs, the skulking behaviour of cats and the synthesis of hormones.

In Nectar, 7% (*n*=20) of the mentioned traits are traits which do not vary within a population, for example a nose, blood proteins, cytochrome B, cell receptors and the urge of baby's to start walking around their first year.

In none of the both biology methods, these traits are coupled with the term 'allele', which is correct since traits without variance in a population are coded by genes but not by alternative alleles.

C5

Traits which remain the same vs. traits which change

Both BvJ and Nectar mention traits which can change during the lifetime of an organism as examples. Only traits which are specifically described as changing or changeable were counted.

In BvJ, only 4% (*n*=22) of all examples are explicitly mentioned as changing or changeable, for example alcohol drinking, wheat plant height, skin tanning and the production of proteins. In a non-compulsory part of the books, hair colour is described as a changing trait. Thus, no examples of changing Mendelian traits are provided. It should be noted here that the examples height and weight, which are described in the books as partially environment-dependent, also could have been

[24] This is not exactly true, as blood type sometimes can change (Loghem, 1963; Sandler, Langeberg, & Landy, 2012), thus also environmental factors can play a role.

described as changing or changeable during the lifetime of organisms, however, BvJ has missed this opportunity.

In Nectar, only 5% (*n*=16) of all examples are explicitly mentioned as changing or changeable, for example eyesight, IQ, cholesterol level, freckles, hair colour and skin colour. In addition, eye colour, which is a seemingly single-gene Mendelian trait (though it is in truth very complex), is described as a trait which can change. This example is also described as partially environment-dependent, thus here Nectar has seized the opportunity to connect the attributes of C3 and C5.

C6

Traits on organism level vs. traits on lower organizational levels which indicate the chance of or susceptibility to certain traits or diseases

Both BvJ and Nectar mention **traits which indicate resistance or susceptibility as examples**.

In BvJ, 4% (*n*= 18) of all examples are such traits, for example predisposition to diabetes, resistance against mosaic virus and resistance against penicillin. However, in none of these examples it is clear that these traits are in fact measured with biomarkers. No specific biological entities indicating susceptibility or resistance except 'gene for' are mentioned. In fact, the microarray used to measure mRNA expression is discussed, but it is not linked to the trait concept.

In Nectar, 3% (*n*= 8) of all examples are traits which indicate resistance or susceptibility, for example susceptibility to gluttony, drought resistance, resistance to the white fly and susceptibility to breast cancer as measurable by analysing mutations in the BRCA-1 gene. Only the last example is described as a biomarker, though not with the specific term *'biomarker'*. However, because the gene sequence of the BRCA-1 gene is not an endophenotype because it is DNA itself, this example is not counted in this C6 category.

7.3 Reliability

The interrater reliability for the raters was found to be Kappa = 0,627 ($p < .0005$), 95% CI (0.437, 0.817). This is a substantial agreement (Landis & Koch, 1977).

8. Conclusion

In this section, the research question *'To what extent do biology textbooks cover the current scientific definitions, hyponyms and attributes of the concepts hereditary trait and phenotype?'* is answered. However, first the answers to the sub-questions will be evaluated.

a) Subquestions 1 – 2

The first subquestion was about what attributes and hyponyms of the concepts *hereditary trait* and phenotype could be distinguished in actual science. These attributes and hyponyms have been inventoried in paragraph 6.1.

The second subquestion was about what definitions of the concepts *hereditary trait* and phenotype can be distinguished in actual science and what attributes or hyponyms they do reveal. The definitions and the attributes and hyponyms implicit in them are listed in paragraph 6.3.

In paragraph 6.2, the found attributes and hyponyms have been categorized in a framework. In each of the six categories, the attributes and hyponyms have been described and contrasted with textbook descriptions and students' understanding as described in paragraph 3.5.

b) Subquestions 3 – 4

The third subquestion was about which of the attributes and hyponyms as described in the paragraph 6.2 framework are represented in secondary education biology textbooks and to what extent. It was hypothesized that the Dutch textbooks would count only outer visible, physical, Mendelian traits as *hereditary trait* or *phenotype* and fail to mention (C1 – C6):

1. Non-outer traits (hyponym)
2. Multiple organizational levels of traits (attribute)
3. Partially environment-dependence of traits (attribute)
4. Traits without variance in a population (hyponym)
5. Traits which change during the lifetime of an organism (hyponym)
6. Traits which indicate susceptibility or resistance (hyponym)

As described in 7.2, it was found that *Biologie voor Jou* as well as *Nectar* do not purely restrict themselves to static, simple traits with clearly visible variation within species. Traits revealing the scientific side of all categories (C1 – C6) were mentioned, though most categories had only few representatives in the books. In short, more than 60% of the examples were non-outer traits (C1), about 25% of the examples were described on multiple organizational levels, 10-15% of the examples were described as partially environment-

dependent (C3) and C4 – C6 hyponyms occurred in less than 10% of the examples. However, traits which indicate susceptibility or resistance (C6) were never labelled with the word 'biomarker' or endophenotype'.

In addition, the C2 and C3 attributes and the C5 hyponym (though the fact that some traits can change is described only in a non-compulsory part of the 'Heredity' chapter) were explicitly mentioned in both the havo and the vwo version of *BvJ*, while C4 was mentioned explicitly only in the vwo version. Further, in both the havo and the vwo version of *Nectar*, C3 is explicitly mentioned. C1, C4 and C5 were explicitly mentioned in the vwo version.

These findings can be summarized in table 1 below (checkmarks indicate that the category occurs in the textbook, minus marks indicate that the category does not occur in the textbook):

Table 1 Occurence of the scientific side of the C1 – C6 categories in BvJ and Nectar books.

Cate-gory	BvJ havo		BvJ vwo		Nectar havo		Nectar vwo	
	Explicit in text	Implicit in examples	Explicit in text	In examples	Explicit in text	In examples	Explicit in text	In examples
1	-	√ 59%	-	√ 53%	-	√ 55%	√	√ 79%
2	√	√ 23%	√	√ 24%	-	√ 23%	-	√ 27%
3	√	√ 15%	√	√ 16%	√	√ 11%	√	√ 9%
4	-	√ 12%	√	√ 11%	-	√ 5%	√	√ 8%
5	√[25]	√ 5%	√[26]	√ 4%	-	√ 1%	√	√ 8%
6	-	√ 4%	-	√ 3%	-	√ 2%	-	√ 3%

The fourth subquestion was about what definitions are used in secondary education biology textbooks for *hereditary trait* and *phenotype.* In paragraph 7.1, these definitions are analysed and it was found that the Nectar *phenotype* definition about only outer traits does match scientific definitions and that the BvJ definition does not. The *hereditary trait definitions* of both methods do match scientific definitions indeed. However, both BvJ and Nectar use *phenotype* also for specific traits while they do not explain this conceptual variation. In addition, many cases of unpunctual language use (like '*the allele protruding teeth*') have been found in the books.

[25] In non-compulsory part of the textbook
[26] In non-compulsory part of the textbook

c) Main research question

In answer to the main research question, *'To what extent do biology textbooks cover the current scientific definitions, hyponyms and attributes of the concepts hereditary trait and phenotype?'*, it can thus be concluded that biology textbooks cover the current scientific definitions and conceptual hyponyms and attributes of the concepts hereditary trait and phenotype to unsatisfactory extent. While some attributes and hyponyms of *phenotype* and *hereditary trait* regarded important in scientific texts are richly represented in textbook examples and explicitly mentioned, other attributes and hyponyms are not explicitly described and only in a small percentage of the hereditary trait examples represented in the textbooks.

Further, the *phenotype* definition of the BvJ books does not match scientific definitions. Third, the presence of conceptual variation in the use of *phenotype* and unpunctual language is a barrier to correct and complete covering of the scientific definitions, hyponyms and attributes of the concepts hereditary trait and phenotype.

In the Discussion chapter, the likelihood and nature of problems that students are likely to experience regarding the *hereditary trait* and *phenotype* concepts are further explained.

9. Discussion

In this section, the results of this research are interpreted in the first paragraph named 'Implications'. Further, the limitations of this research are listed and finally, suggestions are posed for future research.

9.1 Implications

The nature of problems that students will experience regarding the *hereditary trait* and *phenotype* concepts can be divided in four aspects:

1) problems due to limited textbook attention to certain *hereditary trait* hyponyms or attributes
2) problems due to the unusual *phenotype* definition in BvJ
3) problems due to conceptual variation regarding the concept *phenotype*
4) problems due to unpunctual language use

These causes of these problems have been discussed in the Conclusion section. As already discussed in the Introduction, there are four types of possible problems:

a) unclear definitions can lead to problems in understanding related genetic concepts like 'gene' and 'adaptation'
b) unclear definitions can lead to rote learning as opposed to meaningful learning
c) unclear, incorrect or limited understanding of genetics can lead to problems in practical situations
d) unclear, incorrect or limited information about a concept can lead to inability to use the concept in new situations

The problems themselves are worked out piece by piece below.

1) Problems due to limited textbook attention to certain hereditary trait hyponyms or attributes

To analyze the problems as formulated in the first aspect, it is important to have a closer look at the results about the six categories. For each category, it was indicated what percentage of the examples exhibited the scientific side of the category and whether it was explicitly mentioned in the textbook. Learning psychology theories indicate that the provision of many examples with a specific attribute results in recognition of the attribute and inclusion in the prototype (see paragraph 3.4). It is likely that the same is valid for hyponyms. In the textbooks, many traits are non-outer traits, thus for C1, this means that

(despite the fact that the hyponym 'non-outer traits' is not explicitly mentioned in most books) it is very likely that students using the books will recognize that not all traits are outer, but that there are also internal traits. It is important to note, however, that still about half of the examples of hereditary traits provided in the 'Heredity' chapter are outer, discrete, Mendelian traits. Especially in questions about familial inheritance of traits and calculations of risks of getting diseased, such traits are used. In the BvJ introduction of the paragraph 'Monohybrid crossings' for example, it is stated that in genetics, crossbreeding is a general method to study the inheritance of traits. Thus, it is still possible that students associate 'inheritance' or 'hereditary' with Mendelian traits and with the accessory Punnett squares. Connected with this is the fact that it is possible that the context in which the hereditary trait or phenotype examples are provided, influences whether pupils think of them as hereditary traits or just as traits without thinking of genetics. For example, it is well possible that students reading the chapter Behaviour do not realize that the traits they encounter are indeed hereditary traits and make a connection to their genetic knowledge. Thus even though many non-Mendelian traits are provided, the context of these traits could prevent students from recognizing important attributes and hyponyms for their conceptual understanding of *hereditary trait* and *phenotype.* It should be noted that Mendelian traits are not *simple* traits or outdated traits, but the problem is that it is not explained what proteins are involved in the pathway from the gene to the phenotype and that often, multiple genes are involved (Fogle, 1987). The wrinkled pea skin, for example, is caused by an insertion in the gene coding for the SBEI protein involved in the manufacture of amylopectin resulting in a lower starch concentration, which causes the wrinkled pea skin (Offner, 2011).

For C2, the fact that provision of many examples with specific attributes leads to recognition of the attribute would mean that both students using the Nectar and the BvJ book do both recognize the fact that traits have entities on different organizational levels. This can be stated with even more certainty for BvJ, because in that method, organizational levels are explicitly introduced and referred to in the books. However, also for this category it should be noted that students could have struggles to apply this attribute also to Mendelian traits. After all, Mendelian traits are often described with the formulation 'gene for *x*' in the textbooks without mentioning biological entities in-between.

It is already also likely that students using BvJ or Nectar recognize the attribute that (most) traits are partially environmentally dependent (C3). In both BvJ and Nectar, this attribute is also explicitly mentioned and repeated. Again, it could be that students have struggles to apply this attribute also to Mendelian traits because Mendelian traits are often described with the formulation 'gene for *x*' in the textbooks without mentioning the influence of environmental factors.

As in the havo versions of both BvJ and Nectar, the hyponym 'traits without variance within a population' is not explicitly mentioned and as only about 10% of the BvJ examples and

about 5% of the Nectar examples are such traits, it is not likely that students will recognize this hyponym. In the vwo versions, C4 is explicitly mentioned and it is thus possible that vwo students do recognize this hyponym indeed.

Only very few examples (less than 10%) of changeable traits (C5) are provided in the BvJ and Nectar books. This attribute is only mentioned explicitly in the Nectar vwo version and in a non-compulsory part of the BvJ books. Therefore, it is likely that only vwo students using Nectar will recognize that certain traits can change.

Finally, only very few examples of susceptibility or resistance (C6) are mentioned in the BvJ and Nectar books. In none of the books, the term 'biomarker' or 'endophenotype' is used, thus it is likely that students do not recognize this hyponym.

It can be concluded that it is thus possible that most students associate 'hereditary traits' with simple Mendelian traits without thinking of organizational levels, non-outer traits or environmental factors (let alone the scientific side of C4 to C6). Returning to the theoretical problems as described in the Introduction section of this paper, this has the theoretical implication that students' genetic knowledge is not coherent and thus less meaningful than desired (**problem b**). This could result in situations that students do not see the connection between genetics and evolution because education about inheritance mainly focuses on dichotomous (either/or) traits on organism level instead of on normal distributed phenotypic variation within or between populations, which is the substrate for selection (Dougherty, 2009). Further, students might be unable to use their knowledge about hereditary traits in practical situations (**problem c**), for example, it is possible that pupils will think too deterministic and with a confused understanding of risk when they think about the inheritance of diseases (Gifford, 2000; Dougherty, 2009). It is well possible that they regard this chance as computable with so-called Punnett squares, which are central in using pedigree problems often used in genetic textbook examples and tasks (Gericke, Hagberg, Santos, Joaquim, & El-Hani, 2014). A limited view on what factors ultimately influence whether a family trait will occur or not will probably discourage couples to get a baby when they consider the chance of passing on the trait to their baby[27] (Thomas, 2000). It is possible that students do not realize that environmental factors like diet can prevent or lessen a disease because they think genetic traits are unchangeable (Gifford, 2000).

2) Problems due to the unusual *phenotype* definition in BvJ

As described in the Results and Conclusion sections, the BvJ definition of *phenotype* is about outer traits, which is very problematic.[28] As showed in 7.1, understanding gene expression, the genotype-phenotype relation, dominance and evolution are seriously threatened by the definition of the phenotype as the whole of outer traits (**problem a** and **b**).

[27] EO broadcast http://gemi.st/EO_101215216 *Ik kom bij je slapen* – Lindsay's dilemma (2014). [TV programme] Nederland 3: EO.

[28] This definition could be influenced by some scientific definitions which state that the phenotype is *measurable* or by the Greek etymology suggesting that the phenotype is that what *appears*.

Secondly, as the definition only covers part of the traits resulting from the genotype, it is possible that students (like already hypothesized in the introduction) cannot use the concept well in new situations involving non-outer traits (**problem d**).

Finally, it is probable that students using BvJ do not know the difference between the carrier status for a disease and a disease that does not have outer visible symptoms because of phenotype definition. As already described in the introduction, Chamany, Allen, & Tanner (2008) found that conflation of the carrier status and the diseased status can yield serious problems (**problem c**).

3) Problems due to conceptual variation regarding the concept *phenotype*

Further, it is a problem that the term phenotype is used in both BvJ and Nectar both for the whole of traits and for individual traits. Using different semantic versions of a concept is called conceptual variation, and Gericke et al. (2014) have showed that without explaining this conceptual variation, using it is very dangerous for students' conceptual understanding (**problem b**). It could hinder students in using the concept in novel situations (problem **d**).

The conceptual variation of the term phenotype in education could be defended by the notion that the meaning of terms is always under construction, subject of the process of building meaning (Lewis & Lynn in: Slisko & Dykstra, 1997). However, Slisko and Dykstra, 1997 stress that the disadvantages of conceptual variation outweigh the advantages because it can also lead to confusion of linguistic and logical structures in the network of related terms (**problem a**).

4) Problems due to unpunctual language use

In a number of cases, unpunctual language use is detected in genetic terminology. For example, alleles and traits are sometimes used interchangeably in both books, which is clearly wrong. These descriptions can contribute to a sloppy use of genetic terms, which is dangerous for conceptual understanding (**problem b**).

Secondly, as found in respect to category 5, BvJ and Nectar use formulations of 'genes for diseases' which misrepresents the molecular base of diseases (most diseases are caused by malfunctioning or absent genes). Further, the healthy equivalent of a disease is ascribed to the 'other allele' of the gene, which is not in correspondence with the fact that most traits are coded by multiple genes and with the principle of pleiotropy and alternative splicing (Fogle, 1987). The phrase 'gene for' is also harmful for a complete understanding of polygenetic inheritance, dominance, recessiveness and codominance as these terms are regard traits which do not have a one-to-one relation with a gene (**problem a**). Thus also topics like genetic engineering are likely to be misunderstood when the formulation 'gene for' is maintained (Fogle, 1987).

Thus, returning to the research problem which is about whether the use of the concepts *hereditary* trait and *phenotype* in education could be problematic to genetic understanding, it can be answered that this is definitely the case.

Incomplete and incorrect understanding of the concepts *hereditary* trait and *phenotype* could thus contribute to the danger Dougherty summarizes with the words:

'Taken together, current [genetics, DvD] 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.'[29]

These are serious threats to students' present and future responsibility as genetic literate 21-century citizens, thus it is advisable for textbook writers, curriculum writers and teachers to be aware of i) the attributes and hyponyms as described in the category framework, ii) good and equivocal definitions of *phenotype* and iii) the importance to avoid unpunctual language use.

9.2 Limitations

The first limitation of the research is that the vwo6 books of the BvJ method have not been analyzed since these are not already available. However, as these books will draw on the topics nutrition, transport, gas exchange and excretion, protection and balance, it is not likely that this book will contain many examples of hereditary traits. For comparison, the Nectar chapter about excretion contains three examples and the chapter about protection contains four examples of hereditary traits.

Second, it is not tested whether students indeed do not recognize the attributes and hyponyms as described in the scientific side of the category framework and whether they indeed experience problems when they read the concepts *phenotype* and *hereditary trait* or use them in writing.

Third, it is an obstacle that this paper has dealt with the English terms *phenotype* and *hereditary trait* without evidence that the Dutch equivalents *fenotype* and *erfelijke eigenschap* have exactly the same meaning. It is possible that there are differences between the languages in the use of these terms, which might have influenced the outcomes.

Fourth, it has not been tested whether the categories, as extracted from dictionaries, articles and books, are indeed important in science. Their presence in scientific texts does not imply that they are also of crucial importance in scientific research.

[29] Dougherty, *Closing the Gap,* p. 8

Fifth, only one recent dictionary has been used for inventorizing the current definitions of *hereditary trait* and *phenotype.* It is possible that that dictionary is not completely representative because it is from a specific country or from a specific author who does not know the full range of use of the terms in modern science.

Sixth, it has not been tested what students get from the media about genetics. It is well possible that students do see TV-programmes about genetics at home or watch instructional video clips in the classroom which contain more information and reveal more of the attributes and hyponyms of *hereditary trait* and *phenotype* than what is presented in the textbooks.

9.3 Further research

In further research, it should be tested whether secondary school students indeed do not recognize the six categories. In such research, also assumptions like the one that students do not capture attributes or hyponyms provided the low number of examples with information about them, should be tested.

In addition, the importance of the categories and the actual use of the terms *hereditary trait* and *phenotype* in actual science should be tested. For example, it should be researched whether it is important that endophenotypes are recognized as hereditary traits and how important these endophenotypes are in biological or medical research. This will also indicate whether it is important to include the categories in education.

10. Literature

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This section is not part of the official SEC research requirements and should therefore not be graded.

Appendix I: Recommendations

Undoubtedly, the question has now arisen how the concepts 'phenotype' and 'hereditary trait' should be used in education. This question has multiple answers. First of all, as the concept 'genome' is used in the textbooks without the relational opposite 'phenome', it would be a good first step to include the term 'phenome'. This is a term that describes the totality of traits (or 'phenotypes' of an organism and it already plays a role in science in the field of phenomics. Though 'phenome' is sometimes defined as the totality of 'phenes', it better not to introduce the term 'phene' as this would only cause confusion and because that term does not often occur in scientific texts. Further, it should be explained in the books that 'phenotype' originally was coined as a descriptor of the totality of traits, but that it is now used also for individual traits. Together with that, it should be concluded that 'phenotype' thus can be synonymous with both 'phenome' and 'trait' and that it is depends on the context which of these versions is used. Another option would be elimination of the term 'phenotype' from the textbooks, however, as the term is still used often in research, this would only be ignorance of the conceptual variation of this term in science and advance problems with the term 'phenotype' to higher education where the term is still used.

Further, the formulation 'expressed in the phenotype' should either be abandoned or be defined very clearly as gene expression. If the formulation would still be used, it is crucial to explain why genes are sometimes not expressed and to connect this with the principle of epigenetics.

Third, the principles of multiallely and pleiotropy should be introduced already early in the genetics or heredity chapters. Start with many-to-many gene-trait relations. Simplification to one gene coding for one trait should be avoided because this is a simple lie. Instead, it should be made explicit that traits seemingly coded for by one gene are in fact defects that are caused by the absence or malfunctioning of a specific part of the DNA. Connected with this, dominance, recessiveness and codominance should be explained better, for example with help of the websites http://genetics.thetech.org/ask/ask227 and http://learn.genetics.utah.edu/content/inheritance/patterns/. In addition, mutations could be classified in amorph, hypomorph, hypermorph, antimorph and neomorph to make dominance, recessiveness and natural selection more clear.

Fourth, genes should not be pictured as discrete part of the DNA of as beads on a string. Alternative splicing and methylation should be introduced to explain why it is hard to speak of a 'gene'. Further, avoid formulations of 'genes are of interest' or 'play an important role' or explain why they are of interest or play an important role.

In general, teachers and textbook writers should make use of the History and Philosophy of Science (HPS) to explain conceptual variation (Genseberger, 1989b). They should show that Johannsen already had the many-to-many understanding and that developments in research

shifted the focus from outer visible traits to internal traits. Further, genetics education should be language-oriented, which is the principle of paying attention to and talking with students about concepts and their definitions. This is a good way to avoid unpunctual and unclear language use and problems resulting from these.

As some researchers use the terms *phenotype* and *hereditary traits* too loosely, for example the incorrect formulation that laboratory animals not showing a difference with control animals 'do not have a phenotype' (Crusio, 2002) also science itself should consider what rules should be established for the terms *phenotype* and *hereditary traits.*