Heredity and Genetics[1]

No doubt the questions of why members of a species resemble each other or why family members may look similar or different have always interested people. No less intriguing are questions related to the mechanisms by which such similarities or differences are passed along or developed. An early idea put forward by the Greek philosopher Aristotle about inheritance or heredity was based on blood. This idea supposed that the blood of parents mixed and blended to result in their offsprings' characteristics. Some of the terms still used today have their origins in this idea: such terms are blood relatives, pureblood, blood lines. Later in history the development of microscopes and discovery of eggs and sperms also started new speculations:

- One idea stated the existence of a completely preformed individual, in very small size, inside a sperm. Once implanted into a female body, this new individual just grew in size.
- Others felt that this complete individual was in the egg, rather than the sperm.
- Another theory was that sperm and eggs contained sample cells from all body areas and that, combined in embryos, they just reproduced those cells into complete individuals.

Genetics and the cell theory

The development and refinement of microscopes and the emergence of the **cell theory** prepared the field of genetics for great advancements beginning in the early 1900's. The idea of **new cells always coming from existing cells** (which is the central dogma of the cell theory) brought forward the idea of life forms based on a continuation. That is, your cells originated from your parents, theirs from their parents and, in this fashion back into time. The questions of how far back and to what kinds of early life forms, often raise interesting speculations. Surprisingly, the most important beginnings in genetics did not originate with microscopic work or cellular studies. Instead, they began with simple breeding experiments in which the experimenter, a monk named Gregor Mendel, had no previous knowledge of genes, chromosomes or the processes of mitosis and meiosis and their roles in reproduction.

Variation – differences and similarities

The people in the photograph, and you, belong to one type or species of animal. Scientists call this species *Homo sapiens*. As you can see, the people have lots of things in common. For example, they have the same general body shape and their faces have similar features. However, even though they are all easily recognizable as humans, there are lots of small differences between them. People, like all living things, inherit their characteristics from their parents. Some of your characteristics come from your mother and some come from your father. This is why there are variations in every species of plant and animal. These variations are very important and have helped *Homo sapiens* in particular to evolve over millions of years into a very successful species.

Continuous variation *vs*. discontinuous variation

Different hair colour, body height, weight and skin colour are examples of different traits that we call variation. Since e.g. the skin colour can vary from all shades of black to white, we say this trait shows a **continuous variation** in human beings all over the world. In contrast traits showing a **discontinuous variation** also exist. Discontinuously varying traits are either present or not, e.g. you can either roll your tongue (see picture below) or you can't, there is nothing in between. It is a sort of "all or nothing" property.

[1] Sources: Ministry of Education, Saskatchewan; Biology first Oxford University Press, 1999; Biozone International Ltd (Cornelsen).

It is easy to follow the discontinuously varying traits you have and relate them to the entities that determine their inheritance: all traits are passed on from generation to generation by **genes** that are responsible for the appearance of these traits. Genes are entities in your DNA in the nucleus of the cells in your body. They are, of course, also present in sperm and egg cells and hence mix during fertilization, so that the offspring of parents are always composed of a selection of genes from the father and the mother. Discontinuously varying traits are often inherited by a single gene, whereas continuously varying traits are inherited by several genes.

Phenotype and genotype

Genes exist in varying forms that are called **alleles**. For each discontinuously varying trait human beings have two alleles in their DNA. Either the exact same alleles or two different alleles. Take a look at the **trait** handedness in the picture below. The **allele** for right handedness is abbreviated with a capital "R" and that for left handedness with a lower case "r". The capital letters always indicate the **dominant allele**, and the lower-case letters always indicate the **recessive allele**. If you inherited the alleles "RR" in your DNA you are definitely right-handed. If you inherited the alleles "Rr" in your DNA you are still right-handed, since the "R" allele dominates the "r" allele. Only in case you inherited the alleles "rr" you are left-handed. Look at the different traits depicted in the figure below and determine your **phenotype** and your **genotype**. Your **phenotype** is the collection of **traits** or **characteristics** you have. The **genotype** is the collection of all **genes** or rather **alleles**, dominant or recessive, in your DNA. In short: if you are right-handed your **phenotype** is said to be right-handed. Your **genotype** thus might be either "RR" or "Rr", not, however, "rr". If you are left-handed, your **phenotype** is said to be left-handed and your **genotype** has to be "rr". A genotype such as "RR" or "rr" is said to be **homozygous**, whereas a genotype such as "Rr" is said to be **heterozygous**.

Task 1: Read the text, list all new terms shown in **bold letters** and provide a short definition/explanation for these terms according to the text.

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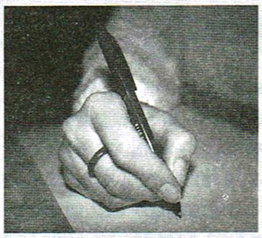
Task 2: Check out the traits listed in the figure below (Fig. 3) and determine your individual phenotype and genotype. Use the table below (Tab. 1) to document your results. In case of a dominant trait you have to write down the homozygous as well as the heterozygous genotype for this trait, since you do not know which one you really have.

Trait: Handedness

Dominant	Recessive
	
Phenotype: Right-handed	**Phenotype:** Left-handed
Allele: R	**Allele:** r

The trait of left or right handedness is genetically determined. Right-handed people have the dominant allele, while left handedness is recessive. People that consider themselves ambidextrous can assume they have the dominant allele for this trait.

Trait: Hand clasp

Dominant	Recessive
	
Phenotype: Left thumb on top	**Phenotype:** Right thumb on top
Allele: C	**Allele:** c

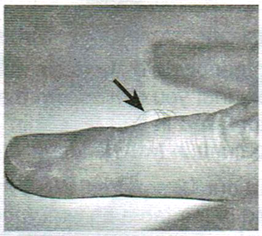
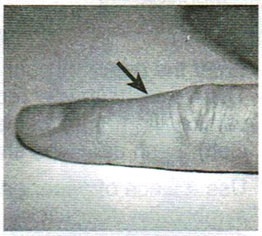
Like handedness, hand clasping shows dominance/recessiveness. When the hands are clasped together, either the left or the right thumb will naturally come to rest on top. The left thumb on top is the dominant trait (C), while the right thumb on top is recessive (c).

Trait: Dimpled chin

Dominant	Recessive
	
Phenotype: Chin cleft	**Phenotype:** No chin cleft
Allele: D	**Allele:** d

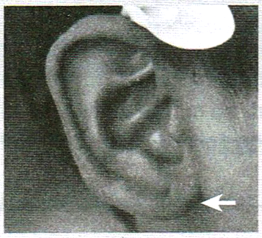
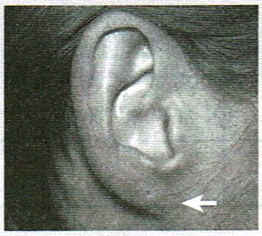
A cleft or dimple on the chin is inherited. A cleft is dominant (D), while the absence of a cleft is recessive (d), although this gene shows **variable penetrance**, probably as a result of modifier genes.

Trait: Middle digit hair

Dominant	Recessive
	
Phenotype: Hair on middle segment	**Phenotype:** No hair on mid segment
Allele: M	**Allele:** m

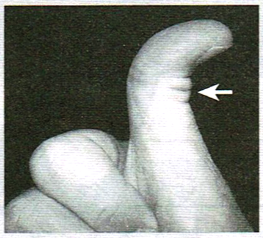
Some people have a dominant allele that causes hair to grow on the middle segment of their fingers. It may not be present on all fingers, and in some cases may be very fine and hard to see.

Trait: Ear lobe shape

Dominant	Recessive
	
Phenotype: Lobes free	**Phenotype:** Lobes attached
Allele: F	**Allele:** f

In people with only the recessive allele (homozygous recessive), ear lobes are attached to the side of the face. The presence of a dominant allele causes the ear lobe to hang freely.

Trait: Thumb hyperextension

Dominant	Recessive
	
Phenotype: 'Hitchhiker's thumb'	**Phenotype:** Normal thumb
Allele: H	**Allele:** h

Photos: RA

There is a gene that controls the trait known as 'hitchhiker's thumb' which is technically termed distal hyperextensibility. People with the dominant phenotype are able to curve their thumb backwards without assistance, so that it forms an arc shape.

Your traits:	Thumb	Ear lobes	Chin cleft	Middle digit hair	Handedness	Hand clasp
Phenotype:						
Genotype:						