An Action-oriented Way to Learn Classical Genetics – Part II

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ABSTRACT

This article describes how to enable an action-oriented introduction of learners to classical genetics using the cotton reel model. In concrete terms, it deals with getting to know and understand Mendel's laws as well as the particularities of sex-linked inheritance.

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Introduction

The cotton reel model was presented in a previous article (Schlüter, 2013). Using this, the structure of a set of chromosomes can be explained, meiosis can be performed with a gaming orientation and the diversity of different chromosome combinations during egg cell and sperm formation can be discovered. At this point, further possibilities of using the cotton reel model are presented to teach different patterns or combinations of inheritance.

Extension of the Cotton Reel Model: Representation of Individual Genes

The basic structure of the model is not described further at this point. Relevant information is available in Schlüter (2013). For the representation of different inheritances, the cotton reel model must be extended to rubber bands which symbolize different genes or alleles. The number of genes considered while working on the model depends on the diversity of the different inheritances that the teacher would like to address. One inheritance, but also up to four different inheritances can be analyzed.

In the example presented below, reference is made to four different genes, and thus, also to four different inheritances (Table 1). Ideally, monogenic inheritances should be simulated. This is a problem if, in doing so, we want to refer to clear, easily identifiable characteristics in humans. In some cases, a monogenic inheritance had been assumed, which however, turned out to be incorrect; in other cases, the inheritance was represented in a simplified way in literature, although in a way not corresponding to reality (McDonald, 2010). According to this, it is pointed out that the inheritances described in this model for tongue rolling, the shape of earlobes and the shape of the hairline do not correspond to a simple monogenic inheritance (McDonald, 2010). For purposes of clarity, they are, however, represented as such.

Working with the Model: The "Cotton Reel Game"

The "game" or "play" task for the students is to create at random the sets of chromosomes/cotton reels of several egg cells and sperm cells from the sets of chromosomes/cotton reels of the parents, so as to "procreate" four children. The procedure regarding the simulation of meiosis and germ cell formation is described by Schlüter (2013). Once the set of chromosomes/cotton reels of the first child has been determined, it is recorded in color on the work sheet (Figure 1). Here, the color of the cotton reels is much less in the foreground than that of the rubber bands. Subsequently, the initial sets of chromosomes of the parents are rebuilt in order to procreate another child, until the cotton reel family has four children and the sets of chromosomes of all children are documented on the work sheet.

Analysis of the Model Work

First, the students determine the phenotype of all members of the cotton reel family. For the parents, they indicate the phenotype on the work sheet (Figure 1) in the left column of the tabular overview "Inheritance of Different Characteristics." Here, they only have to mark with a cross if the mother and the father are tongue rollers or not, if their earlobe is free or attached, etc. The corresponding genotype is already given at this point. Table 2 provides an overview of the genotypes and phenotypes of the parents.

The students indicate the phenotypes of the four children on the work sheet directly next to the corresponding sets of chromosomes/cotton reels, also by marking with a cross the appropriate distinction of characteristics. The students subsequently count for each characteristic (e.g., tongue rolling, shape of the earlobes, etc.) how often the different distinctions occur in the four children of their cotton reel family. They record these figures on the work sheet in the Table "Inheritance of Different Characteristics." For this, they use the first white small box (A) of the three successive small boxes respectively. Moreover, they analyze how often the three characteristics (tongue rolling, shape of the earlobe, haemophiliac) occur in girls and boys. For these three characteristics, they also indicate the incidence of the different genotypes.

Exactly the same information is subsequently provided for all cotton reel children produced in the class. This information is entered into the second, light grey small box (B). Now, we not only have the data of four but, for example, of 40 cotton reel children (in 10 student groups in the class). The higher the number of descendants, the easier it is to derive the ideal ratio of the different distinctions of characteristics from it.

The ideal ratio of the different distinctions of characteristics is subsequently determined by means of crossing schemes (see below). This ratio is entered in the third, dark-grey small box (C) and related to the previous small box (B), the number of distinctions of characteristics in all cotton reel children in the class. The following inheritances can be dealt with by working with the cotton reel model.

Mendel's First Law

Mendel's first law can be derived from the tongue rolling characteristic (gene on the brown chromosome/cotton reel pair). The students find out on their own that all children are tongue-rollers, although only one parent owns the characteristic of tongue rolling.

Moreover, all cotton reel children have the same genotype: Rr or rR. If we mark the genotypes of the parents and those of the children on the work sheet in the table "Inheritance of Different Characteristics," it becomes evident that both parents are homozygous, whereas the children are all heterozygous. Mendel's first law (principle of uniformity) can, thus, be directly understood. "*If two individuals of the same species are crossbred who differ from each other by one characteristic, with the individuals being homozygous for the respective distinction of characteristics, their direct descendants (that is, the individuals of the filial generation) all show the same distinction of characteristics.*" Mendel's skill was to develop this rule without knowing anything about chromosomes, genes and alleles. Figure 2 shows the corresponding crossing scheme for Mendel's first law.

Mendel's Second Law

Mendel's second law can be developed by means of the earlobe shape characteristic (gene on the red chromosome/cotton reel pair). Regarding the phenotypes, the students find out that some of the children even have attached earlobes although both parents have free earlobes. However, these children remain in the minority (ideal ratio: 1 : 3).

It can be derived from the genotypes that both parents are heterozygous and that all three mentioned genotypes appear in the children. The ideal ratio of the genotypes of the children amounts to 1 (FF) : 2 (Ff/fF) : 1 (ff).

Mendel's second law (law of segregation) can be derived by analysing the phenotypes (Figure 3). It is reproduced in a slightly modified manner in the following, as the filial generation is analysed during the cotton reel game and, contrary to Mendel, not the grandchildren's generation. "*If two individuals of the same species are crossed who both show the same distinction of characteristics and are heterozygous in this respect, the distinctions of characteristics split at a ratio of 1:3 in the following generation.*"

Mendel's Third Law

Two characteristics must be considered for developing Mendel's third law: hairline shape in the forehead (gene on the green pair of chromosomes) and earlobe shape (gene on the red pair of chromosomes). As they fill in the table "Inheritance of Different Characteristics," the students see that both parents have the same phenotype: a widow's peak as well as free earlobes.

On the contrary, the children show four different combinations of characteristics: (1) widow's peak and free earlobes, (2) widow's peak and attached earlobes, (3) straight hairline and free earlobes, (4) straight hairline and attached earlobes. Here, the first mentioned combination of characteristics is most frequently observed, and the last mentioned one is least frequently observed. The ideal ratio of the mentioned phenotypes can again be determined by means of a crossing scheme: 9 : 3 : 3 : 1.

The genotypes of the parents clearly show that they are heterozygous for both characteristics. As the inheritance of both characteristics occurs dominant-recessively, Mendel's second law can again be used in both cases. This means that the proportion of children with widow's peak to those with straight hairline amounts to 3 : 1 (in 16 children 12 : 4). The ratio of children with free earlobes to those with attached earlobes is also 3 : 1. In altogether 16 children, 75% (= 12) theoretically have a widow's peak. Of these 12 children with widow's peak, 75% (= 9) theoretically have a free earlobe and only 25% (= 3) have an attached one. The same applies to the four children with straight hairline: 75% (= 3) have a free earlobe and 25% (= 1) have an attached one (Figure 4).

An additional question deals with the extent to which the set of chromosomes of the parents looks different when the two observed characteristics are linked, that is, when only the two dominant distinctions of characteristics (free earlobe and widow's peak) as well as the two recessive distinctions of characteristics (attached earlobe and straight hairline) are observed together, but there is no combination of dominant and recessive distinctions of characteristics. For this, the gene for hairline shape as well as the gene for earlobe shape must appear on the same (for example the red) pair of chromosomes. They are, thus, linked and inherited together. The allele combination must be such that the dominant alleles of both genes appear on the same (for example the light red) chromosome and the recessive alleles of both genes are on the other (for example the dark red) partner chromosome. Due to gene linkage, the distribution of the characteristics in the descendants corresponds to the expected result of Mendel's second law, that is, only two phenotypes are observed (Figure 5). If, on the contrary, the genes are not linked, four different phenotypes (Figure 4) appear, as already mentioned.

Based on the obtained results, Mendel's third law (in altered form) can be phrased as follows: "*If two individuals of the same species are crossbred, the distinctions of different characteristics are inherited independently from each other and appear in all combinations only if the genes are on different chromosome pairs.*" (Note: This statement applies as long as the parents are heterozygous in the considered characteristics. If, on the contrary, they are homozygous, the principle of uniformity applies and the descendants show the same phenotype – this, however, does not apply to the grandchildren's generation.)

Sex-linked Inheritance

The particularities of sex-linked inheritances can be addressed by means of the characteristic "haemophiliac" or "non-haemophiliac" (gene on the blue chromosome/cotton reel pair). The different sizes of the chromosomes/cotton reels in the model clearly show that the Y chromosome is significantly smaller than the X chromosome and therefore provides less space for genes. Diverse genes which are present on the X chromosome are, therefore, missing on the Y chromosome, thus also the gene for coagulation factor VIII.

With the cotton reel model, the genotypes of the parents were selected as to make the following aspects become visible in the offspring production: (1) Healthy parents can have "ill" children. In the present inheritance, the ideal ratio of healthy to ill children is 3 : 1. (2) More boys than girls are affected by haemophilia. In the inheritance taken as an example, only the boys (theoretically half of the boys) are haemophiliac. (3) Women can be carriers of haemophilia and, still, be phenotypically healthy. In the inheritance taken as an example, all girls are phenotypically healthy; however, half of them are theoretically carriers of the defective gene. The following ideal ratio can be determined by means of a crossing scheme: ill boys: healthy boys : healthy girls (not carriers): healthy girls (carriers) = 1 : 1 : 1 : 1. Ill girls are not observed in the inheritance taken as an example. They could only be observed if the father was haemophiliac and the mother was a carrier (Figure 6).

A sex-linked inheritance can be derived from an uneven distribution of the distinction of characteristics in girls and boys. It is true that such an uneven distribution is directly identifiable in the described inheritance taken as an example; however, it would not be visible phenotypically as soon as the father is haemophiliac (no matter whether the mother is a carrier or not). The uneven distribution of the distinction of characteristics in the sexes becomes visible, however, as soon as entire phylogenetic trees are analyzed.

No sex linkage can be observed in the inheritance of the characteristics "tongue rolling" and "shape of the earlobe". All "procreated" girls and boys are tongue rollers. In the ideal case, half of the children with free earlobes (theoretically 75%) are girls and the other half are boys. The same applies to children with attached earlobes (theoretically 25%).

Table 1. Overview of the Different Genes and Alleles on the Chromosomes/Cotton Reels as Well as of the Inheritances that Can Be Deduced by Means of the Allele Composition

		Maternal allele		Paternal allele		
Color of the chromosome/ cotton reel	**Characteristic/ gene for …**	**Expression of the characteristic / allele for …**	**Color of the allele/ rubber band**	**Expression of the characteristic / allele for …**	**Color of the allele/ rubber band**	**Inheritance**
Pair of chromosomes no. 1/brown (autosomes)						
light brown	**Tongue rolling**	tongue rolling *(dominant)*	purple	no tongue rolling *(recessive)*	red	**Mendel's first law**
dark brown		tongue rolling *(dominant)*	purple	no tongue rolling *(recessive)*	red	
Pair of chromosomes no. 2/red (autosomes)						
light red	**Shape of the earlobe**	free earlobe *(dominant)*	blue	free earlobe *(dominant)*	blue	**Mendel's second law**
dark red		attached earlobe *(recessive)*	yellow	attached earlobe *(recessive)*	yellow	
Pair of chromosomes no. 3/green (autosomes)						
light green	**Shape of the hairline (in the forehead)**	widow's peak *(dominant)*	brown	widow's peak *(dominant)*	brown	**Mendel's third law**
dark green		straight hairline *(recessive)*	white	straight hairline *(recessive)*	white	
Pair of chromosomes no. 4/blue (sex chromosomes/heterosomes)						
light blue	**Coagulation factor VIII**	defective coagulation factor *(recessive)*	red	working coagulation factor *(dominant)*	green	**sex-linked inheritance**
dark blue		working coagulation factor *(dominant)*	green	missing gene due to small size of the Y chromosome		

Table 2. Genotypes and Phenotypes of the Parents

Characteristic	**Mother**		**Father**	
	Genotype (rubber band colors and possible abbreviations for the alleles)	**Phenotype**	**Genotype** (rubber band colors and possible abbreviations for the alleles)	**Phenotype**
Tongue rolling	purple-purple → RR	tongue rolling	red-red → rr	no tongue rolling
Shape of the earlobe	blue-yellow → Ff	free earlobe	blue-yellow → Ff	free earlobe
Shape of the hairline	brown-white → Ww	widow's peak	brown-white → Ww	widow's peak
Blood coagulation	red-green → ***x***X	working blood coagulation	green-… → Xy	working blood coagulation

For the genotype, the allele combination is indicated in the form of rubber band colors and abbreviations. Upper case letters correspond to a dominant, lower case letters to a recessive distinction of characteristics. In detail, the letters mean: R = tongue rolling, r = no tongue rolling, F = free earlobe, f = attached earlobe, W = widow's peak, w = straight hairline, X = allele for working gene product on X chromosome, x = allele for defective gene product on X chromosome, y = missing gene on Y chromosome.

Figure 1. Work Sheet

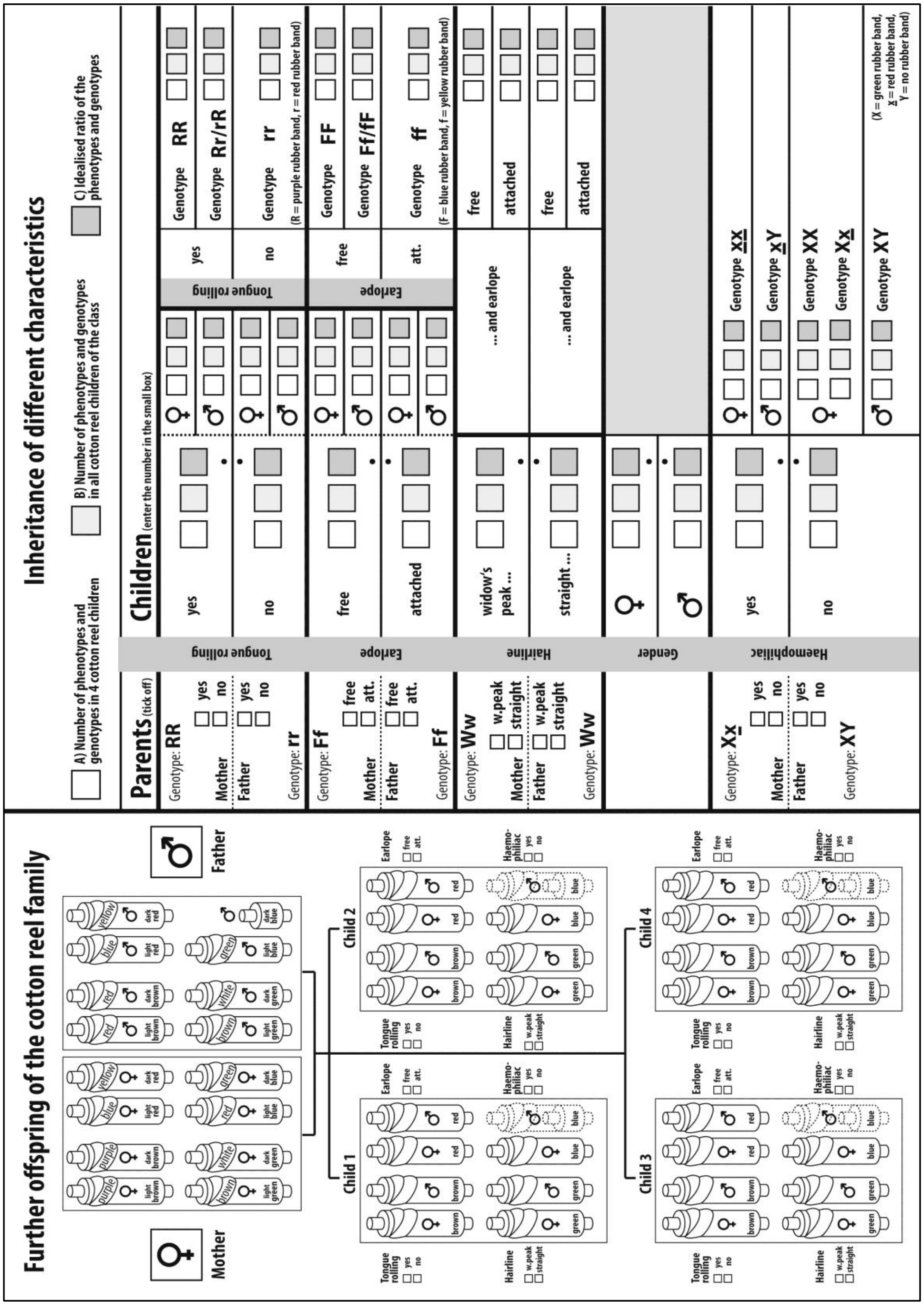
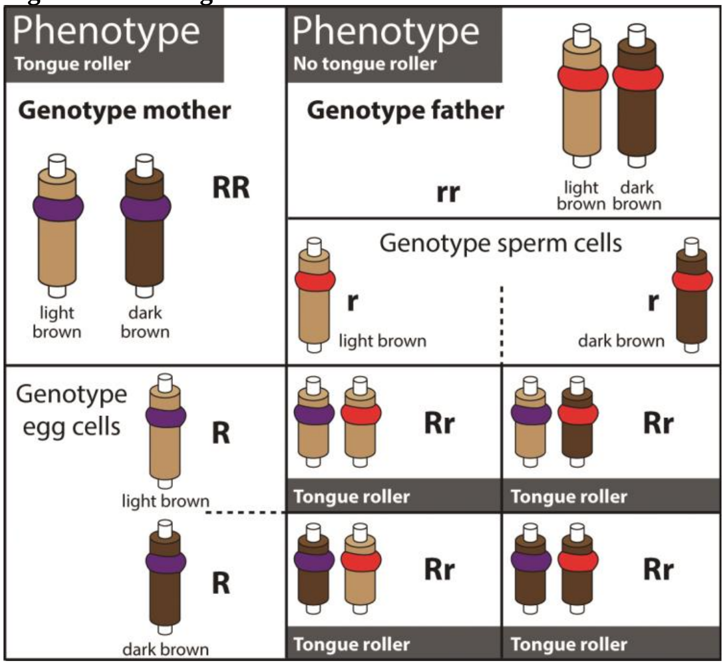
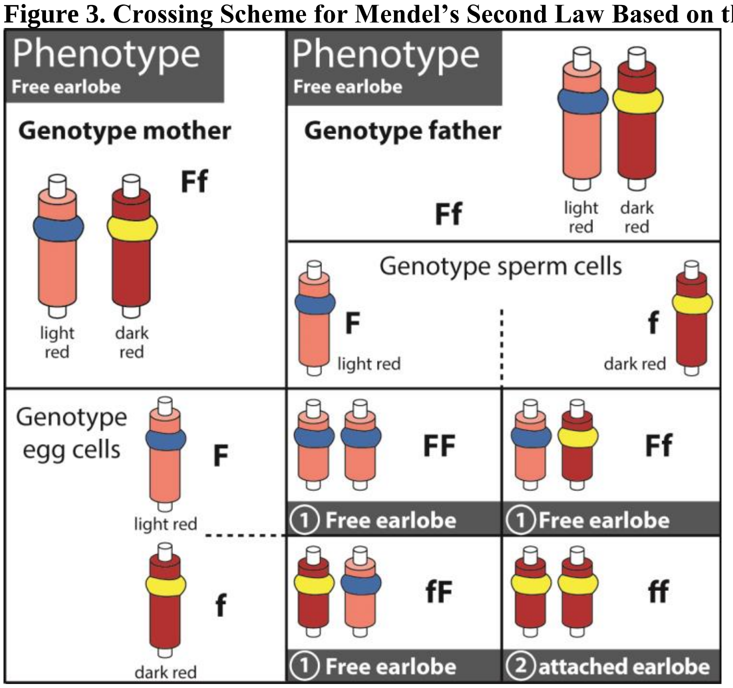


Figure 2. Crossing Scheme for Mendel's First Law Based on the Example of Tongue Rolling Inheritance



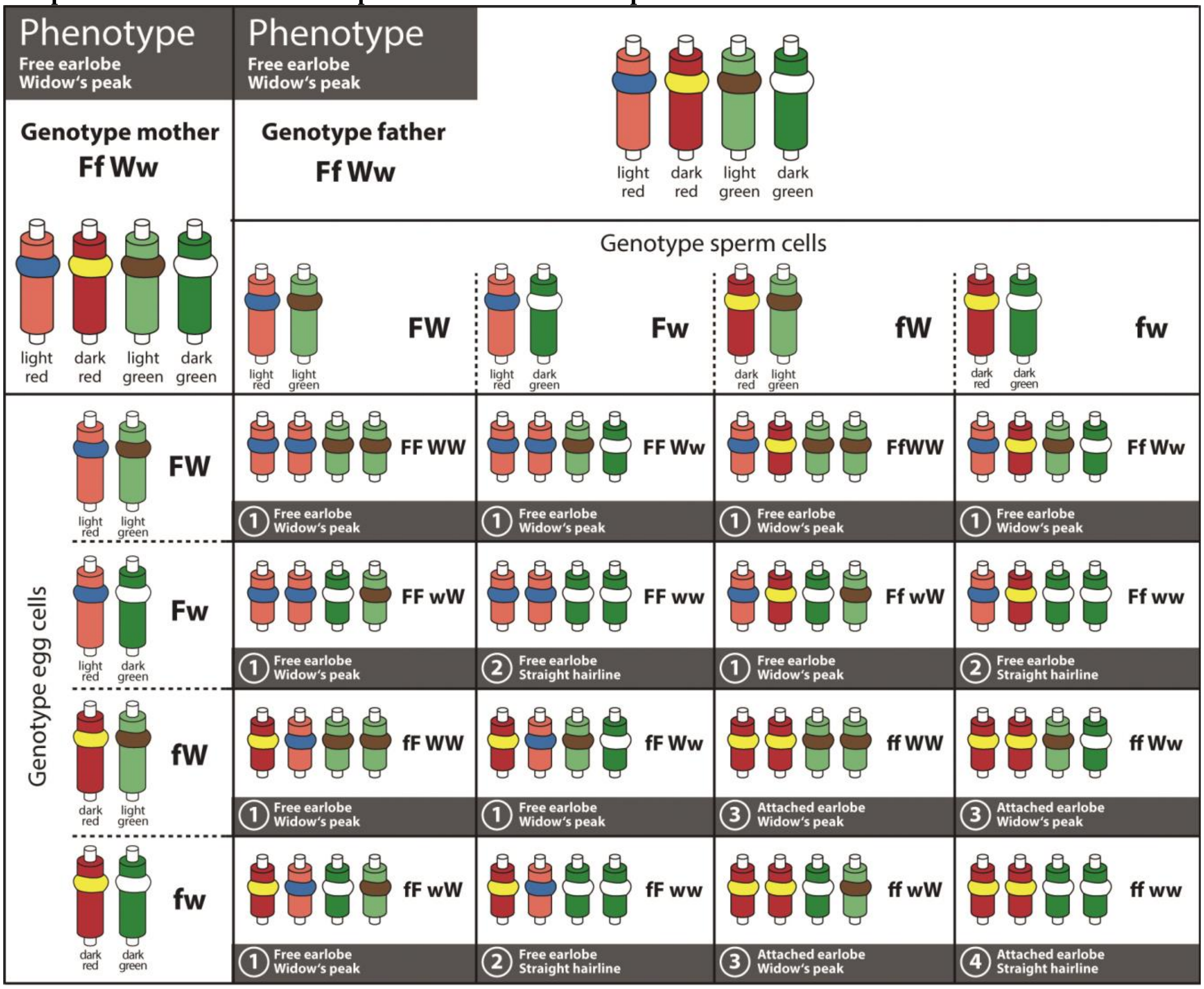
R = allele for tongue rolling, r = allele for no tongue rolling, lower case letter = allele for recessive distinction of characteristics, upper case letter = allele for dominant distinction of characteristics.

Figure 3. Crossing Scheme for Mendel's Second Law Based on the Example of Earlobe Shape Inheritance



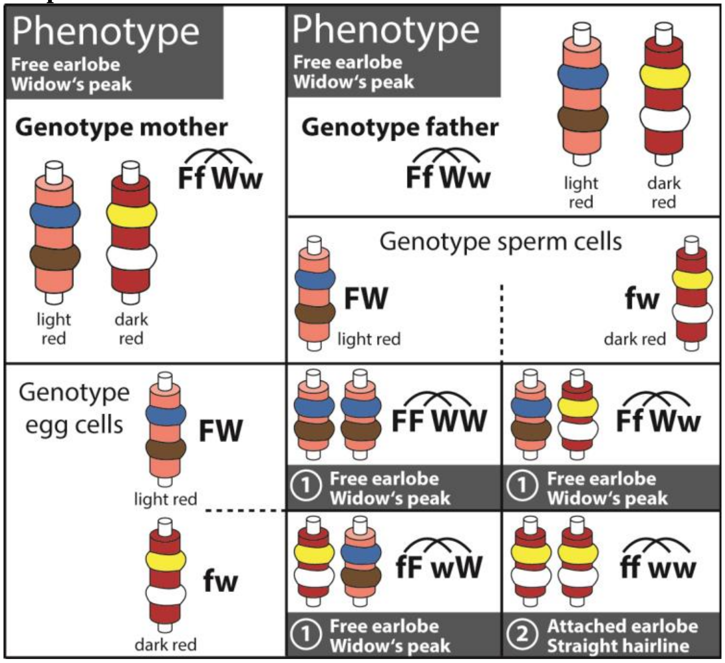
F = allele for free earlobe, f = allele for attached earlobe, lower case letter = allele for recessive distinction of characteristics, upper case letter = allele for dominant distinction of characteristics.

Figure 4. Crossing Scheme for Mendel's Third Law Based on the Example of the Inheritance of Two Independent Characteristics: Shape of the Earlobe and Shape of the Hairline



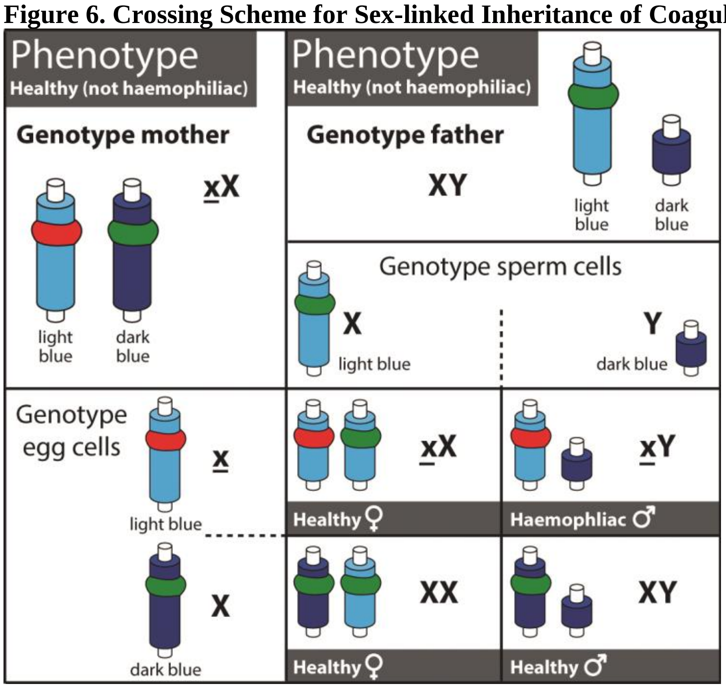
F = allele for free earlobe, f = allele for attached earlobe, W = allele for widow's peak, w = allele for straight hairline, lower case letter = allele for recessive distinction of characteristics, upper case letter = allele for dominant distinction of characteristics.

Figure 5. Crossing Scheme for the Inheritance of Two Dependent Characteristics: Shape of the Earlobe and Shape of the Hairline



F = allele for free earlobe, f = allele for attached earlobe, W = allele for widow's peak, w = allele for straight hairline, lower case letter = allele for recessive distinction of characteristics, upper case letter = allele for dominant distinction of characteristics.

Figure 6. Crossing Scheme for Sex-linked Inheritance of Coagulation Factor VIII



X = allele for working coagulation factor, x = allele for defective coagulation factor, y=missing gene/allele on the Y chromosome, lower case letter=allele for recessive distinction of characteristics, upper case letter=allele for dominant distinction of characteristics.

Summary

As presented, the cotton reel model enables students to understand the transmission of different characteristics from the parents to the children in a playful way. It thereby enables an action-oriented introduction into the topic of inheritance which is otherwise rather overly theoretical. Moreover, the model enables a better understanding of the relationship between the inheritance of individual genes/alleles and their location in the double set of chromosomes. For this purpose, different modes of representation are selected. It starts with an action-oriented representation dealing with the cotton reel model and the procreation of several cotton reel children. This is followed by the iconic representation, in which entire sets of chromosomes as well as individual pairs of chromosomes are illustrated with the genes/alleles located on them. At the end comes the symbolic representation, which refers to the abbreviation of alleles which are often found isolated in crossing schemes. Thus, three different modes of representation are addressed, by means of which content can be presented according to Bruner (1966). These three proposed different ways of representation (representation triad) on the same topic should enable a better understanding of the contents.

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