

Supplemental Material

CBE—Life Sciences Education

Timm *et al.*

SUPPLEMENTAL MATERIALS

Questionnaire

In this part it is your task to analyze the given pedigrees and to work on the corresponding tasks. Please always pay attention to the task and indicate at the end of each task how difficult you found the processing of the respective task.^[1]

Task 3a

Analyze the pedigree on the next page regarding the mode of inheritance.^[2]

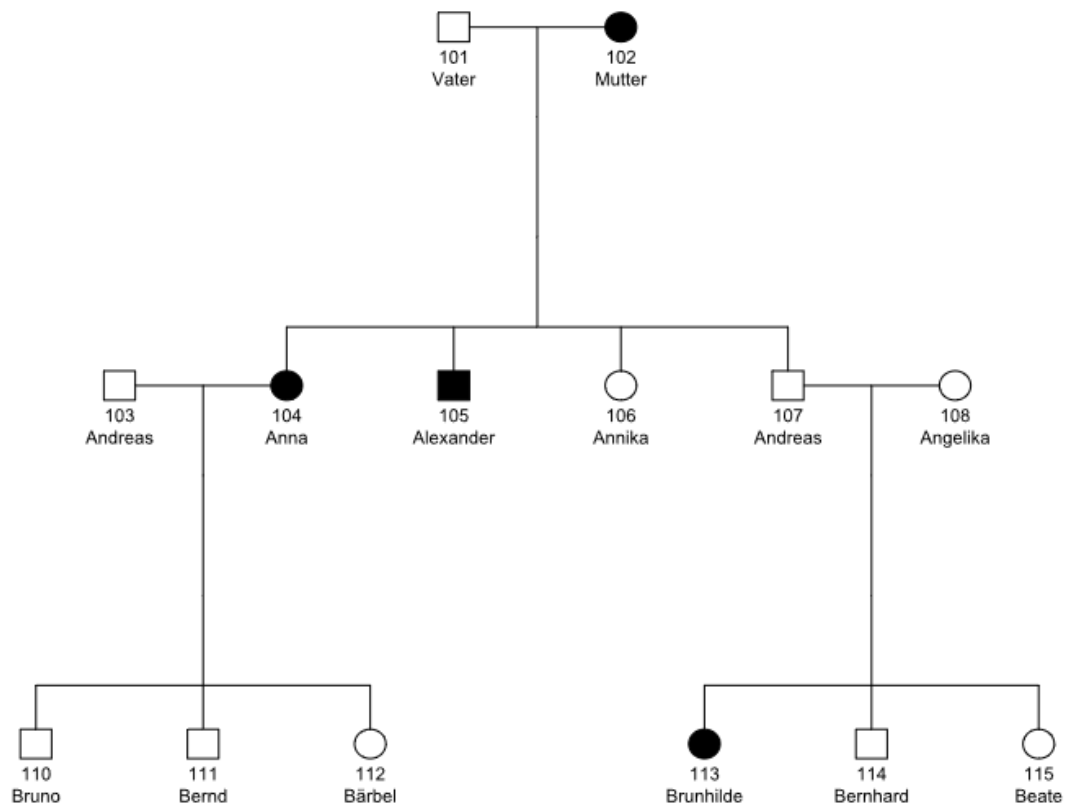
Describe your approach as coherently and completely as possible.^[3]

Note: Each person in the family tree is assigned a number. Please always use these numbers when referring to individual persons.

Now, based on your previous considerations, determine the mode of inheritance shown.^[4]

- autosomal-dominant
- autosomal-recessive
- X-gonosomal-dominant
- X-gonosomal-recessive

Pedigree Task 3a



[5]

Task 3b

Analyze the pedigree on the next page regarding the mode of inheritance.^[2]

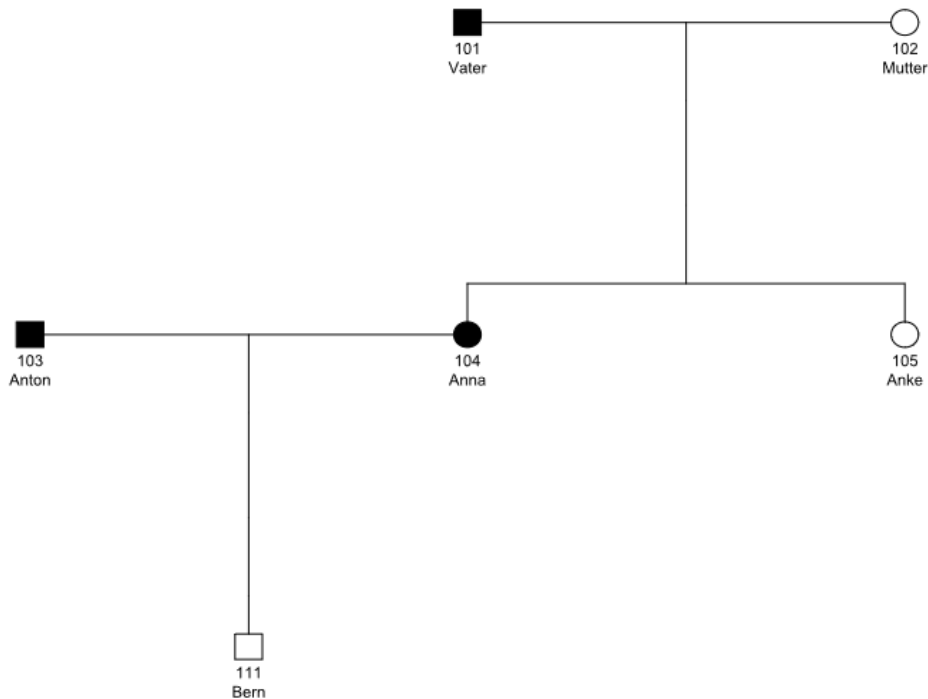
Describe your approach as coherently and completely as possible.^[3]

Note: Each person in the family tree is assigned a number. Please always use these numbers when referring to individual persons.

Now, based on your previous considerations, determine the mode of inheritance shown.^[4]

- autosomal-dominant
- autosomal-recessive
- X-gonosomal-dominant
- X-gonosomal-recessive

Pedigree Task 3b



[5]

Remarks

[1] In adapting the instrument for younger students, we have made minor linguistic changes. This included switching from addressing younger students as “Du” to using the polite form “Sie” with older students.

Changes that affect the content level have been marked individually (see Notes 2 and 4).

[2] For the beginning genetics students we slightly modified the instructions to emphasize the usual goal of pedigree analysis, to determine the mode of inheritance. The instruction there was as follows:

Analyze the pedigree on the next page to determine the mode of inheritance at hand.

[3] Hackling and Lawrence (1988) included a similar prompt.

[4] We have adjusted the answer options somewhat for the beginning genetics students in case they lack the technical terms related to X-linked inheritance. The response options for this group were:

- autosomal dominant mode of inheritance
- autosomal recessive mode of inheritance
- sex-linked (X-chromosomal) dominant mode of inheritance
- sex-linked (X-chromosomal) recessive mode of inheritance

[5] Both pedigrees were generated semi-automatically based on matrices (Surmann, 2017) and plotted with the R package kinship2 (Therneau and Sinnwell, 2015).

Table of variables and categories including code descriptions and examples

Table 1: *Variables and categories used to characterize students' reasoning on pedigree problems.*

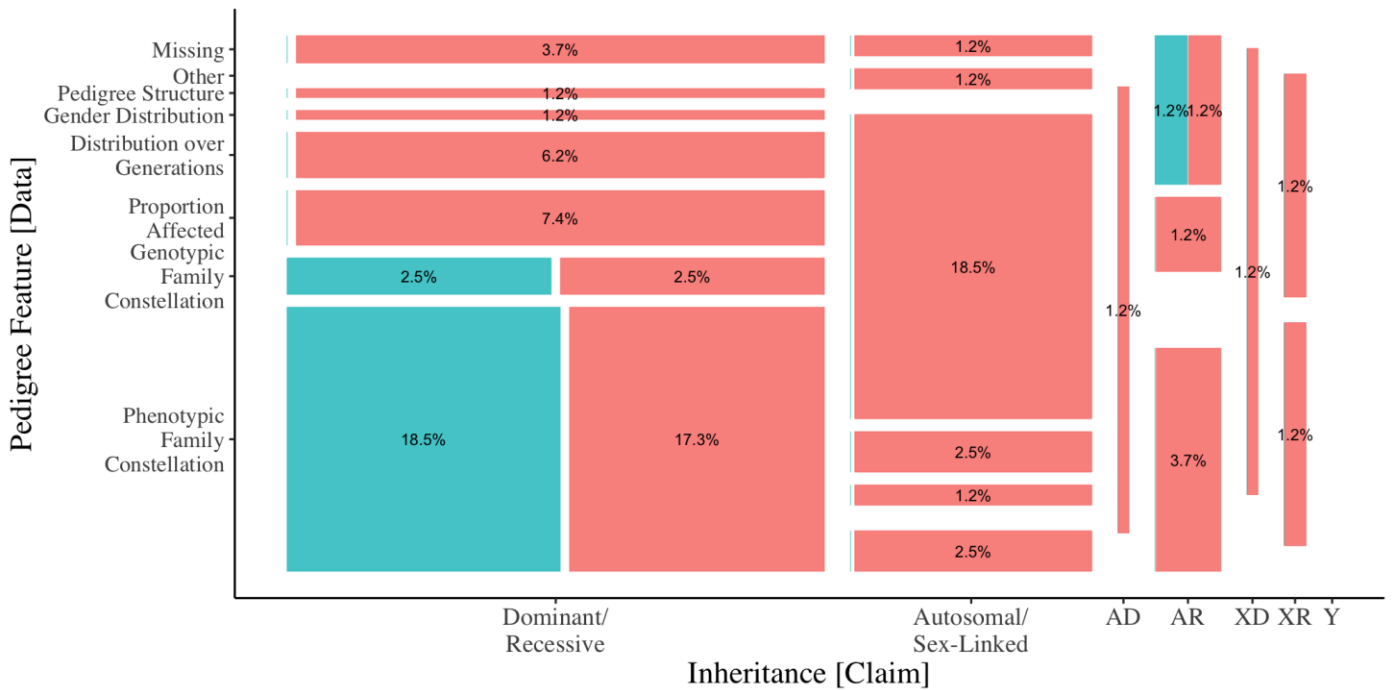
Variable	Categories	Code Description	Example
Type of Statement	Description	Description of a concrete family constellation or a pedigree feature.	“Two men [101, 103] and one woman [104] are affected.” [1016: 41]
	Allegation	Unsubstantiated statement/assertion that goes beyond the descriptive of given information.	“The mode of inheritance relates to sex.” [1035: 34]
	Clue	A specific family constellation or a pedigree feature is taken as an indication of a determination (i.e., regarding mode of inheritance or a genotype).	“Again, both sexes are affected, which again indicates autosomal inheritance.” [1012: 41]
	Proof	A specific family constellation or a pedigree feature is taken as proof of a determination (i.e., regarding mode of inheritance or a genotype).	“If you look at the first and second daughter generations, you see that parents 107 and 108 have a child with the trait (113), so the inheritance must be recessive.” [1013: 35]
	Other	Statement without concrete reference to the present pedigree.	“The first thing I did was to look at the pedigree to see how many generations are shown.” [1038: 34]
Pedigree Feature	Phenotypic Family Constellation	(Phenotypic) description of a concrete person or family constellation within the pedigree.	“Father 101 is trait carrier. Mother 102 healthy. Daughter 104 ill.” [1002: 39]
	Genotypic Family Constellation	(Genotypic) description of a concrete person or family constellation within the pedigree.	“Dominant does not work either, because there is no dominant allele in the parents [107, 108]. Thus it is only autosomal recessive.” [D17: 43]
	Proportion of Affected Persons	Indication of the proportion of affected persons in relation to the total number of persons.	“Since very few people are affected, the inheritance is recessive.” [1003: 34]
	Gender Ratio Among Affected	Indication of the gender ratio of those affected.	“Since males and females are affected, the disease can only be autosomal.” [D19: 41]
	Distribution of Affected over Generations	Description of the trait's occurrence as a function of the generation to which it	“This pedigree cannot be dominant either, since the trait is not present in every generation.” [B6: 46]

Variable	Categories	Code Description	Example
		belongs or the generation sequence.	
	Pedigree Size or Structure	Basic features of the pedigree are mentioned, such as the number of generations represented.	“- 3 generations” [1005: 36].
	Other	None of the previous categories can be clearly assigned, but a concrete reference to the pedigree is given.	“The first thing that stands out is that the trait is recessive because it was inherited only conditionally.” [B14: 40]
	Missing	There is no specific reference to the pedigree.	“The mode of inheritance shown is autosomal recessive,” [D4: 40]
Claim	Autosomal Dominant	The statement refers to autosomal dominant inheritance.	“The pedigree cannot be autosomal dominant, since neither (110, 111, 112) are affected. In a dominant inheritance, this should have been the case.” [D8: 43]
	Autosomal Recessive	The statement refers to autosomal recessive inheritance.	“So the pedigree can only be autosomal recessive.” [B6: 42]
	X-linked Dominant	The statement refers to X-linked dominant inheritance.	“Gonosomal dominant can be excluded on individuals 101, 102, & 104.” [B15: 41]
	X-linked Recessive	The statement refers to X-linked recessive inheritance.	“The inheritance can also not be X-gonosomal recessive. This can be seen from the fact that person 113 is trait carrier and thus has two X chromosomes which inherit the trait recessively. However, this is not possible because person 107, as her father, can only inherit one X chromosome to her, which is dominant and person 113 would then also not be a trait carrier.” [B6: 41]
	Autosomal	The statement refers to autosomal inheritance.	“The mode of inheritance is autosomal, as both sexes are affected.” [1009: 40]
	X-linked	The statement refers to X-linked inheritance.	“The mother [102] is trait carrier, therefore the trait is X-chromosomal.” [1006: 34]
	Y-linked	The statement refers to Y-linked inheritance.	“and since a girl is affected, y-gonosomality can also be excluded.” [D2: 43]

Variable	Categories	Code Description	Example
	Dominant	The statement refers to dominant inheritance.	“Since individuals 103 and 104 are both ill but have a healthy child [111], the inheritance is dominant.” [1012: 42]
	Recessive	The statement refers to recessive inheritance.	“Since very few people are affected, the inheritance is recessive.” [1003: 34]
	Other	The statement does not refer to a specific mode of inheritance or a type of inheritance.	“On the pedigree one learns that in the filial generation only the dominant sex was ever inherited.” [1034: 40]
Orientation	Confirmation	The statement has a confirming character.	“3. However, it [the inheritance] must be recessive because 107 and 108 (both not diseased) have a diseased child [113].” [D22: 43]
	Refutation	The statement has a rejecting character.	“1. The process cannot be gonosomal dominant because 101 and 102 have a son [105] affected by the trait and a daughter [106] unaffected by the trait” [D22: 41]
Conclusiveness	Conclusive	The statement is conclusive from a scientific perspective and considering the given pedigree.	“Evidence [of recessive inheritance] is that individuals 107 and 108 have a sick child [113] even though they are healthy.” [1012: 37]
	Indeterminable	The statement cannot be evaluated as either conclusive or inconclusive.	“2. Looking at the points above [individual generations, traits, and sex], I found that the trait was inherited recessively.” [1024: 35]
	Inconclusive	The statement is not conclusive from a scientific perspective and considering the given pedigree.	“The mode of inheritance is autosomal, as both sexes are affected.” [1009: 40]

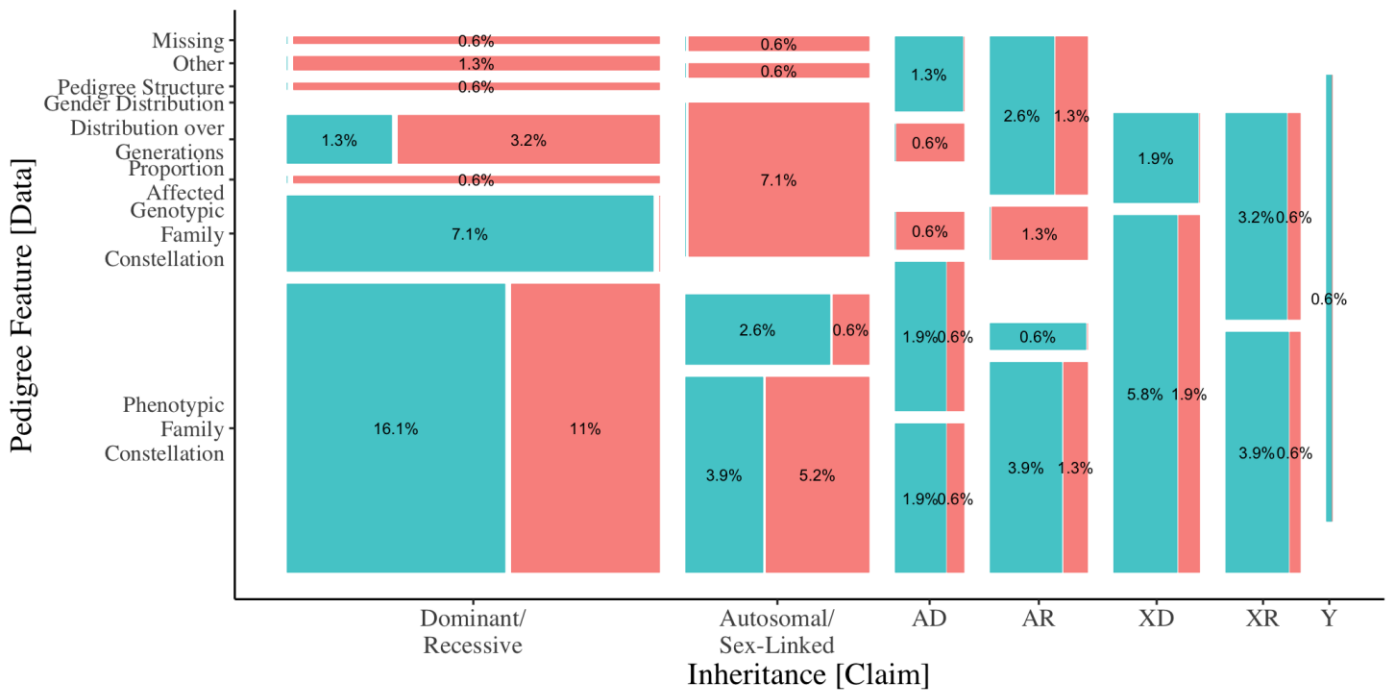
Relationship between argument structure and conclusiveness

(a) Arguments of Beginning Genetics Students



Percentages are based on n = 81 arguments regarding the mode of inheritance.

(b) Arguments of Advanced Genetics Students



Percentages are based on n = 155 arguments regarding the mode of inheritance.

■ Conclusive
 ■ Inconclusive or Indeterminable

Figure 1: Mosaic plot showing the structure and conclusiveness of students' arguments. The relationship between the inheritance mentioned in the argument, the data used to prove that claim and the arguments consistency is shown separately for each group. We suggest using the color coding of Figure 2 to allocate the kind of evidence represented by the individual tiles. A larger area indicates a more frequent occurrence. **AD**: autosomal dominant inheritance; **AR**: autosomal recessive inheritance; **XD**: X-linked dominant inheritance; **XR**: X-linked recessive inheritance; **Y**: Y-linked inheritance