

ESSAY 5

Answers must be written out in paragraph form. Outline form is not acceptable. Labeled diagrams may be used to supplement discussion, but a diagram without a written explanation will not receive credit. You must cite the source of all outside information you include. Include the page number of information from the course textbook or the web address of information found online.



Figure 1. Long big toe (LBT) and short big toe (SBT) phenotypes

The genetic basis for the difference in length of the big toe relative to the second toe in *Homo sapiens* remains unknown. In an investigation of the prevalence and inheritance pattern of relative toe length among inhabitants of Rivers State, Nigeria, a total of 101 families comprising of 101 sets of parents (fathers and mothers) and their offspring were sampled (Aigbogun, Alabi, Didia, & Ordu, 2019). The observed big toe patterns were described as long big toe (LBT), representing a big toe longer than or equal in length to the second toe, or short big toe (SBT), representing a big toe shorter than the second toe (Figure 1). The offspring phenotypes were tabulated alongside the parental combination patterns (i.e., when both parents had LBT, when both parents had SBT, and a combination of LBT and SBT). Chi-square analysis tested the association between sex and big toe length, as well as conformance to a simple Mendelian dominant-recessive pattern. Results are shown in Table 1. Chi-square analysis for cross set I (when both parents had LBT) is shown in Table 2.

TABLE 1. DISTRIBUTION OF BIG TOE LENGTH WITH RESPECT TO PARENTAL PHENOTYPE COMBINATION

Cross Set	Parental Phenotype Combination		F ₁ Male Phenotypes			F ₁ Female Phenotypes		
	Father's Phenotype	Mother's Phenotype	LBT	SBT	Total	LBT	SBT	Total
I	LBT	LBT	24	6	30	28	6	34
II	SBT	SBT	1	8	9	2	6	8
III	LBT	SBT	7	5	12	8	5	13
IV	SBT	LBT	7	10	17	7	5	12

TABLE 2. MENDELIAN CHI-SQUARE TEST FOR FREQUENCY OF BIG TOE LENGTH PATTERN FROM CROSS SET I

Phenotype	Observed	Expected	χ^2
LBT	52	48	0.33
SBT	12	16	1.00
Total	64	64	1.33

- (a) **Explain** how differences in the DNA base pair sequence of a gene can lead to different relative toe length phenotypes. **Describe** how an individual with a heterozygous genotype for big toe length can produce both gametes with the LBT allele and gametes with the SBT allele.
- (b) Based on the expected values in Table 2, **state** the null hypothesis for cross set I. **Construct** a Punnett square for cross set I to illustrate the hereditary basis for the null hypothesis. **Justify** the use of a chi-square test to analyze the mode of inheritance of big toe length.
- (c) **Evaluate** the null hypothesis for cross set I based on the chi-square test in Table 2. Identify the appropriate statistical values AND comparison that **provide support** for your evaluation. Use the data in Table 1 to **justify** the claim that big toe length is an autosomal trait.
- (d) Glucose-6-phosphate dehydrogenase (G6PD) deficiency is a recessive disorder caused by a mutation in the *G6PD* gene on the *Xq28* region of the long arm of the X chromosome (Gigarel et al., 2004). Symptoms include the destruction of red blood cells (hemolytic anemia) in response to the ingestion of certain foods.

A man with SBT and wild-type G6PD production marries a woman with LBT and wild-type G6PD production despite the fact that her father had SBT and G6PD deficiency. Assuming that big toe length is caused by a single autosomal gene where LBT is dominant to SBT, **predict** the possible genotype combinations with respect to big toe length and G6PD production in the woman's egg cells. **Calculate** the probability of each phenotype combination with respect to big toe length and G6PD production in the offspring that arise from this marriage and enter the probabilities on the table provided (Table 3). **Provide support** for your calculation.

References

- Aigbogun, E. O., Alabi, A. S., Didia, B. C., & Ordu, K. S. (2019). Morton's toe: Prevalence and inheritance pattern among Nigerians. *International Journal of Applied and Basic Medical Research*, 9(2), 89–94.
- Gigarel, N., Frydman, N., Burlet, P., Kerbrat, V., Steffann, J., Frydman, R., ... Ray, P. F. (2004). Single cell co-amplification of polymorphic markers for the indirect preimplantation genetic diagnosis of hemophilia A, X-linked adrenoleukodystrophy, X-linked hydrocephalus and incontinentia pigmenti loci on *Xq28*. *Human Genetics*, 114(3), 298–305.

TABLE 3. PROBABILITY OF BIG TOE LENGTH AND G6PD PRODUCTION PHENOTYPES IN MALE AND FEMALE OFFSPRING

Male Offspring			Female Offspring		
Phenotype		Probability Among Male Offspring	Phenotype		Probability Among Female Offspring
Big Toe Length	G6PD Production		Big Toe Length	G6PD Production	
LBT	Wild-type		LBT	Wild-type	
LBT	Deficiency		LBT	Deficiency	
SBT	Wild-type		SBT	Wild-type	
SBT	Deficiency		SBT	Deficiency	
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