

Name: _____

Probabilities of Inheritance

Many genetic diseases follow a Mendelian pattern of inheritance. For the following scenarios, research the disease and determine the gene that is mutated and which protein is affected? Then determine the probability of inheritance. The probability should be expressed as a percentage.

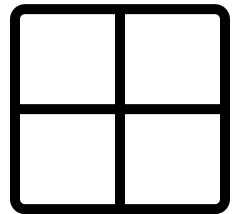
1. Smith-Lemli-Opitz Syndrome (SLOS) is a cholesterol synthesis disorder that follows an autosomal recessive pattern of inheritance. Megan and James had a child that was diagnosed with SLOS shortly after birth. They want to have another child so they went in for genetic testing. They were both determined to be heterozygous for the disorder. What is the probability that they will have another child with SLOS?

Symptoms: _____

Which gene is mutated? _____ Probability of inheritance: _____

Name of affected protein? _____

Describe the relationship between the affected protein and the disease symptoms:



2. Cystic Fibrosis is a genetic condition that affects a person's ability to clear mucous from their lungs. It is inherited in an autosomal recessive pattern. Morgan has cystic fibrosis but her partner Kevin does not. He is not a carrier either. What is the probability of their child having cystic fibrosis? What is the probability that their child is a carrier?

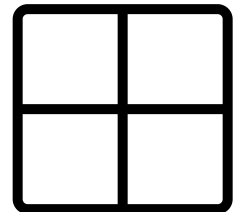
Symptoms: _____

Which gene is mutated? _____

Name of affected protein: _____

Probability of inheriting disease? _____ Probability of being a carrier? _____

Describe the relationship between the affected protein and the disease symptoms:



3. Autosomal dominant polycystic kidney disease (ADPKD) is an inherited disease that causes renal failure. Carrie, the mother, is heterozygous for ADPKD and the father, Damon, is homozygous recessive. What is the probability that they will have a child with ADPKD?

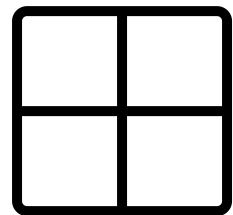
Define “renal”:

Symptoms:

Which gene is mutated?

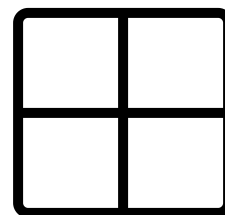
Probability of inheritance?

Describe the relationship between the affected protein and the disease symptoms:



X-Linked disorders occur when the gene mutated is on the X chromosome. In order to predict X-linked disorders, the X and Y chromosome need to be part of the Punnett Square (see example where D=disorder.)

	X^D	X^d
X^D	$X^D X^D$	$X^D X^d$
Y	$X^D Y$	$X^d Y$



4. Beta-propeller protein-associated neurodegeneration (BPAN) is a disorder that damages the nervous system. This disorder is X-linked dominant, which means that this disease is more prevalent in females. If the mother is heterozygous for BPAN and the father is recessive, what is the probability that they will have a child with BPAN?

Symptoms:

Which gene is mutated?

Probability of inheritance?

Describe the relationship between the affected protein and the disease symptoms: