

Sickle Cell Anemia is an autosomal recessive genetic disorder that can cause shortness of breath, fatigue and delayed growth and development. Sickling occurs because of a mutation in the hemoglobin gene. A couple is trying to have their second child. Their first child has sickle cell anemia. They want to know the possibility of their second child getting the disease. The father's parents are both healthy but the mother's mother has sickle cell anemia. What are their chances of having a second child with sickle cell anemia? Draw a pedigree for this family and advise them from the perspective of a genetic counselor on their odds with respect to their second child.

Cystic Fibrosis is an autosomal recessive genetic disorder that affects the lungs and digestive system of about 30,000 children and adults in the United States. Carriers show no symptom of the disease but can pass it on to their offspring. A couple Jane and Mark want to know their probability of having a child with CF. Jane has a history of CF in her family and suspects that she is a carrier of the trait. Mark has no history of CF in his family and is unsure if he is a carrier of the trait. Draw a pedigree and use Punnett squares to show the different possibilities for Jane and Mark. Present them with your findings.

Huntington's disease (HD) is a genetic disorder that affects muscle coordination and leads to cognitive decline and mental problems. It typically becomes noticeable in mid-adult life. The disease is caused by an autosomal dominant mutation. For a couple who has already had their 4 children (2 boys & 2 girls), the husband had a late onset of Huntington's in his 40's. His mother (their child's grandma) also had a late onset of the disease. Genetic testing was not as advanced when his mom died and so they were not sure what to do. The couple now wants to test their children but wants to know what the probabilities are before they begin genetic testing. Draw a pedigree of the family and give them advice accordingly. (Assume that the wife has no history and she is not a carrier)

Down's syndrome also known as trisomy 21 is caused when a person inherits 3 copies of chromosome 21. This happens when the chromosomes fail to separate during meiosis. People with Down's syndrome usually experience varying level of mental delays, and are prone to heart defects. A woman with Down's syndrome wants to have babies with a normal man. What is the possibility that their child will inherit Down's syndrome. Make a Punnett square and advise this couple. (Be careful)

Hemophilia is a recessive sex linked disorder that prevents the body from forming blood clots. A woman who is a carrier of hemophilia and a normal man are given the chance to choose the gender of their child. The woman's grandfather and great grandfather had hemophilia. There is no known history of hemophilia in the man's family. As the genetic counselor would you advise this couple to do so and why? Create a Punnett Square to support your answer

