Case Studies-Genetic Counselor

***Read each Scenario and put yourself in the place of a genetic counsellor. Use punnet Squares/pedigree charts as evidence to advise each family. Create a chart to share your advice with the class/family. You may do additional research if you want to find out more about the disorder.***

1. Sickle 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. The 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.
2. 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 punnet squares to show the different possibilities for Jane and mark. Present them with your findings.
3. 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 children, the husband had a late onset of hungtinton'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 then advice accordingly. (assume that the wife has no history and she is not a carrier)
4. Color blindedness is a sex linked recessive trait. There is no actual blindness but there is a deficiency of color vision. A couple with a color blind man and a woman of normal vision has a child they have two daughters one that is colorblind and one that isn’t. What is the probability that the non color blind daughter will marry and have a son that is color blind? Draw a pedigree to show this. What advice can you give this couple for their future offspring?
5. A couple has been trying to have kids for years. Both parents sought genetic counseling and the father found out that he has a genetic condition called Y chromosome infertility which causes a lack of sperm production, or very little sperm production. What advice do you have for this couple as a genetic counselor? Draw a pedigree for this family.
6. Neurofibromatosis-1 is an inherited disorder in which nerve tissue tumors form in the skin, nerves and brain. It is an autosomal Dominant disorder. A man and woman, Peter and Pam want to have children both have a history of NF-1 in their family. Peter’s mom and Pam’s dad died from NF-1. Peter and Pam met at a support group for people who have lost relatives to NF-1. Peter and Pam want to have children but are worried about the chance of having a child with NF-1. Draw a pedigree and calculate the risk if having a child with NF-1 and advise peter and Pam accordingly.
7. **Adrenoleukodystrophy (ALD)** is X-Linked recessive disorder, it causes rapid degeneration to a vegetative state. Lorenzo’s parents come to you wanting to know their chances of having another child with ALD. Given what you already know about ALD from Lorenzo’s oil. Draw a pedigree of the family and tell them their probability of having a second child with ALD.
8. Fragile X syndrome is the most common form of inherited mental retardation in males and is also a significant cause of mental retardation in females. It is inherited in an X linked Dominant pattern. However the symptoms are sometimes milder in females than males especially if the females are heterozygous. A woman with fragile X Syndrome wants to have kids with a normal man. They seek genetic counseling. The woman with fragile X inherited it from her Mother. Her and her spouse want to know the probability of passing fragile X on to their daughter or son.
9. Down 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 syndrome usually experience varying level of mental retardation, and are prone to heart defects. A woman with Down syndrome wants to have babies with a normal man. What is the possibility that their child will inherit Down syndrome? Make a punnet square and advise this couple.
10. 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. As a genetic counselor would you advise this couple to do so and why?

***Genetic Disorders***

Name\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Period\_\_\_\_

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| **Genetic Disorder** | Facts (Type of inheritance, Symptoms of the disorder, who is more likely to get it) |
| Down’s Syndrome |  |
| Sickle Cell Anemia | 1. Autosomal recessive/ codominant 2. Shortness of breath, fatigue, delayed growth and development 3. Anyone can inherit it |
| Hemophilia |  |
| Cystic Fibrosis |  |
| Huntington’s Disease |  |
| Color Blindedness |  |
| Y Chromosome Infertility |  |
| Neurofibromatosis-1 |  |
| Adrenoleukodystrophy (ALD) |  |
| Fragile X syndrome |  |