Williams OB Chapter 13: Genetics

**CREOG Learning objectives:**

1. Counsel patients about genetics

- Solicit a family pedigree

- Describe and perform pre-pregnancy counseling and testing

- Describe and perform antepartum genetic testing and counseling

- Perform prenatal genetic testing

- Refer for diagnostic testing

2. Describe patterns of inheritance

- Mendelian modes (eg autosomal dominant, autosomal recessive, X-linked)

- Non-Mendelian modes (eg mitochondrial, imprinting, polygenic)

**Practice Questions**

1. A patient presents to her primary care physician to establish care. The patient’s family history is significant for a genetic disorder. When a family pedigree is drawn, there is male-to-male transmission and a lack of gender bias for this genetic condition. Additionally, 50% of the affected siblings also have it. What is the most likely mode of inheritance?

A. Autosomal dominant

B. Autosomal recessive

C. Imprinting

D. Mitochondrial inheritance

E. X-linked recessive

Source: True Learn

2. A 30-year old nulliparous woman presents for her first prenatal care visit at 8 weeks gestation. Both she and her partner are of Ashkenazi Jewish ancestry. They deny consanguinity, and there is no family history of any genetic disorders. Which of the following genetic disorders is most associated with Ashkenazi Jewish ancestry?

A. Canavan disease

B. Cystic fibrosis

C. Familial dysautonomia

D. Fragile X syndrome

E. Tay-sachs disease

Source: True Learn

3. Your patient is a 24-year-old G1 woman with an intrauterine pregnancy at 12 weeks’ gestation. She presents for her new obstetric appointment. Her medical history is significant for type 1 diabetes mellitus, which is under good control with insulin. She has never had surgery. The patient is a known carrier for the X-linked recessive condition hemophilia A. Which of the following Is correct regarding X-linked disorders?

A. A male with an affected allele cannot pass the condition to their sons, but all daughters of affected fathers will be affected with the condition and can pass it on to their children.

B. X-linked recessive patterns manifest by skipping generations as the affected are usually children of unaffected carriers.

C. In the X-linked inheritance pattern, fathers cannot pass the condition to their children; while mothers will pass the disease to all of their children assuming complete penetrance.

D. A male with an affected allele on his single X chromosome is hemizygous and cannot transmit the disorder to their male offspring

E. A male with an affected allele on his single X chromosome is hemizygous and all his daughters would not be obligate carriers.

Source: True Learn

High-Yield Resources:

1) ACOG Technology Assessment #14: Modern Genetics in Obstetrics and Gynecology

<https://www.acog.org/clinical/clinical-guidance/technology-assessment/articles/2018/09/modern-genetics-in-obstetrics-and-gynecology>

2) NCBI Classic Mendelian Genetics: <https://www.ncbi.nlm.nih.gov/books/NBK132145/#:~:text=There%20are%20five%20basic%20modes,%2Dlinked%20recessive%2C%20and%20mitochondrial>

Answers

1. A 2. B 3. D