





10. Secondary sex characteristics arise during puberty. What causes these developmental changes?
  
  
  
  
  
  
  
  
  
  
11. List two examples of secondary sex characteristics in females and two in males.
  
  
  
  
  
  
  
  
  
  
12. Are there secondary sex characteristics that could be the basis of a reliable sex determination test? Explain your answer.
  
  
  
  
  
  
  
  
  
  
13. Regarding DSDs ...
  - a. What does the abbreviation DSD stand for?
  
  
  
  
  
  
  
  
  
  
  - b. How common are DSDs?

**Answer the following questions using the table of DSDs.**

14. *CYP21A2* gene mutations
  - a. By what mechanism can a mutation in *CYP21A2* change testosterone levels?
  
  
  
  
  
  
  
  
  
  
  - b. What changes in phenotype are typical in people with a *CYP21A2* mutation?
  
  
  
  
  
  
  
  
  
  
15. *SRY* gene mutations
  - a. How can an error in chromosome segregation during meiosis affect inheritance of the *SRY* gene?

- b. What is the effect of a nonfunctional *SRY* gene on the development of primary sex characteristics?
- c. Which sex is typically assigned at birth to an individual with a mutation that results in a nonfunctional *SRY* gene?

## 16. XO

- a. How can an error in chromosome segregation result in Turner syndrome?
- b. Can an individual with Turner syndrome have biological children?

17. *AR* gene mutation

- a. Which phenotypes may arise in an individual with a mutation in the *AR* gene?
- b. In humans, the *AR* gene is located on the X chromosome. If a person who is XY has a complete deletion of the *AR* gene, *do you think* giving them testosterone injections during puberty would cause an increase in expression of male secondary sex characteristics compared to not receiving injections? Explain your answer.

## 18. XXY

- a. Which of the following terms describes the process of homologous chromosomes failing to separate correctly during meiosis?
- |                                |                  |
|--------------------------------|------------------|
| A. Chromatid adhesion syndrome | C. Ametaphasia   |
| B. Nondisjunction              | D. Anti-anaphase |
- b. If a person with Klinefelter's syndrome wanted to look more like an average male, what medical treatment could help accomplish this?



**CASE STUDIES**

Click the “**Case Studies**” tab, read about the two athletes and the history of sex verification tests, and then use the information to answer questions 21-22.

21. Click the "**Sprinter**" tab.

a. Which four tests have historically been performed to determine an athlete’s biological sex?

b. What is a genotype that would explain the sprinter’s phenotype? Explain.

22. Click the "**Swimmer**" tab.

Under current regulations, would the swimmer be allowed to compete? Explain.

**EXTENSION ACTIVITY DSD TABLE**

The table on the following page provides additional information on the DSDs shown at the bottom of the Human Development tab. Complete the table to indicate whether a “sex verification test” of a person with each listed DSD would show Barr bodies or the *SRY* gene, whether their testosterone level would be low (typical of a female) or high (typical of a male), and whether they are likely to menstruate or be fertile (able to have biological children without in vitro fertilization or other medical interventions).

Chromosomes	Genetic mutation	Syndrome	Internal reproductive anatomy	External genitalia	Secondary sex characteristics	Sex typically assigned at birth	Barr body (Y/N)	SRY gene (Y/N)	Testosterone (Low/High)	Menstruation (Y/N)	Fertile (Y/N)
46,XX	None	None	Female	Female	Female	Female					
46,XX	<i>CYP21A2</i> mutation or deletion	Congenital adrenal hyperplasia	Female	Ambiguous	Male but reduced	Female					
46,XY	<i>SRY</i> mutation or deletion	Swyer syndrome	Female but nonfunctional ovaries	Female	Absent or female, but reduced	Female					
45,X	Complete or partial loss of X chromosome	Turner syndrome	Female but absent or nonfunctional ovaries	Female	Absent or female, but reduced	Female					
46,XY	Androgen receptor (AR) deletion	Complete androgen insensitivity syndrome	Shortened vaginal canal and testes, no fallopian tubes or uterus.	Female	Female	Female					
47,XXY	Extra X chromosome	Klinefelter syndrome	Male	Male	Male but reduced. Breast growth may occur	Male					
46,XX	<i>SRY</i> gene translocation to X	XX male syndrome	Male	Male but small testes	Male but reduced	Male					
46,XY	<i>SRD5A2</i> mutation or deletion	5-alpha reductase deficiency	Male	Female or ambiguous	Male	Female					No
46,XY	Mutation or deletion of the genes for AMH or AMH receptor ( <i>AMHR2</i> )	Persistent Müllerian duct syndrome	Male reproductive organs plus uterus and fallopian tubes	Male, but one or both testes may be undescended	Male	Male					No
46,XY	None	None	Male	Male genitalia	Male	Male					

**EXTENSION ACTIVITY**

Four factors are considered when determining a person’s biological sex: 1) chromosomes, 2) hormones, 3) primary sex characteristics that are features essential for sexual reproduction (internal reproductive structures and external genitalia), and 4) secondary sex characteristics, features that typically appear during puberty and are different between the sexes, but are not directly involved in reproduction (for example, females typically have enlarged breasts and widened hips; and males typically have increased facial and body hair, increased muscle mass, and a larger Adam’s apple). Given that the top male athletes outcompete the top female athletes by about 10% in most sports, provide a justification for how each of these four factors is or is not likely to contribute to performance differences between sexes.

Factor that helps determine biological sex	Likely to affect athletic performance? (Y/N)	Justification
Chromosomes		
Hormones		
Primary sex characteristics		
Secondary sex characteristics		