

A 15-year-old girl is brought to the physician for evaluation of amenorrhea. She has never menstruated although her mother and sisters had menarche around age 13. The girl has a history of difficulty with identifying different odors but no prior sinus infections or other illnesses. She takes no medications. Examination shows short stature and no pubic or axillary hair. She has no breast development. Ultrasound confirms a uterus and 2 normal-appearing ovaries. Serum follicle-stimulating hormone level is 2 mU/mL (normal 4-30 mU/mL), and luteinizing hormone is 3 mU/mL (normal 5-25 mU/mL). Karyotype result is most likely to show which of the following?

- ☐ A. 45 XO
- ☐ B. 45 YO
- ☐ C. 46 XX
- ☐ D. 46 XY
- ☐ E. 47 XXY

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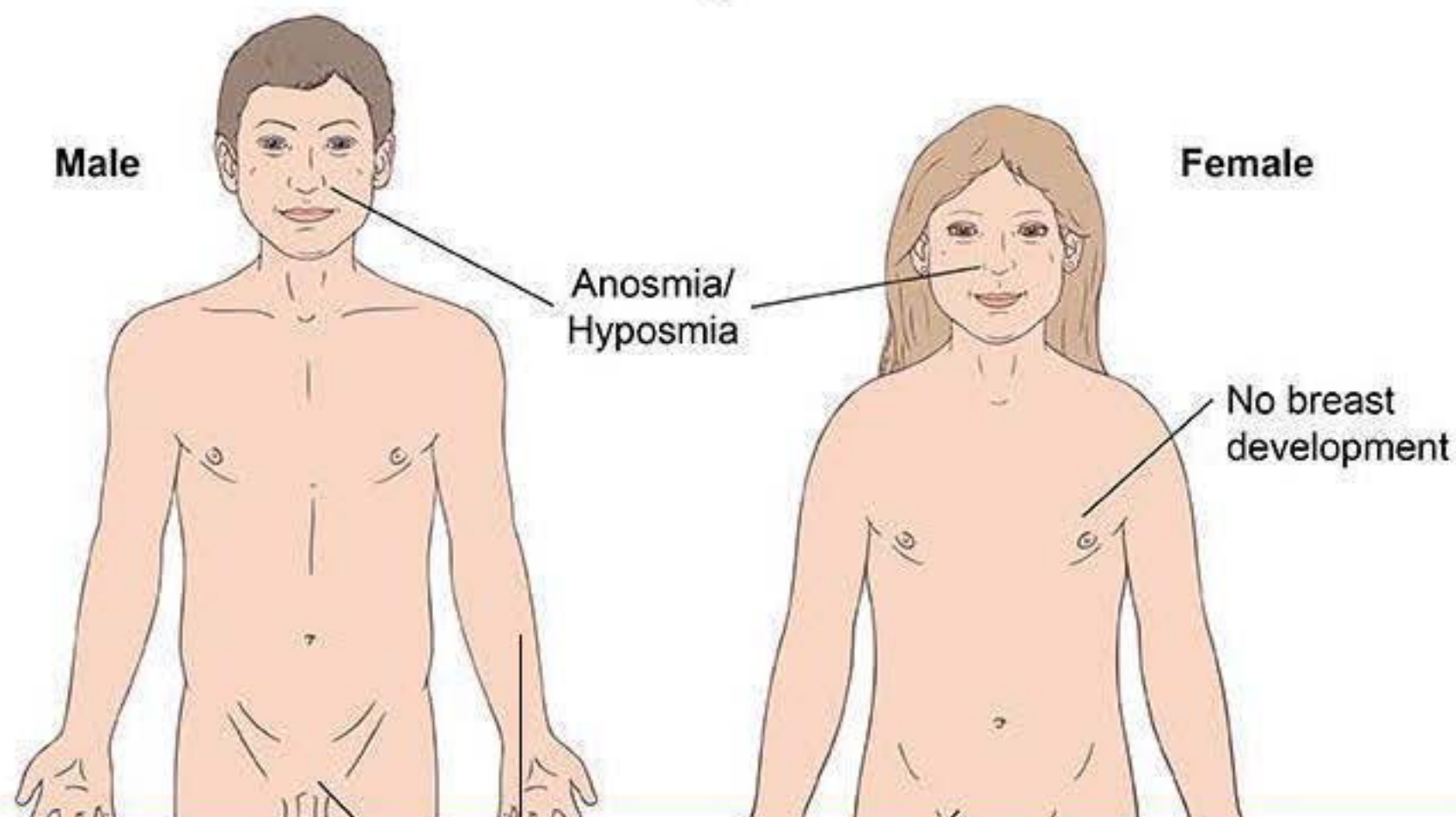
A 15-year-old girl is brought to the physician for evaluation of amenorrhea. She has never menstruated although her mother and sisters had menarche around age 13. The girl has a history of difficulty with identifying different odors but no prior sinus infections or other illnesses. She takes no medications. Examination shows short stature and no pubic or axillary hair. She has no breast development. Ultrasound confirms a uterus and 2 normal-appearing ovaries. Serum follicle-stimulating hormone level is 2 mU/mL (normal 4-30 mU/mL), and luteinizing hormone is 3 mU/mL (normal 5-25 mU/mL). Karyotype result is most likely to show which of the following?

- ☐ A. 45 XO [17%]
- ☐ B. 45 YO [0%]
- ☒ C. 46 XX [72%]
- ☐ D. 46 XY [4%]
- ☐ E. 47 XXY [7%]

[Proceed to Next Item](#)**Explanation:**

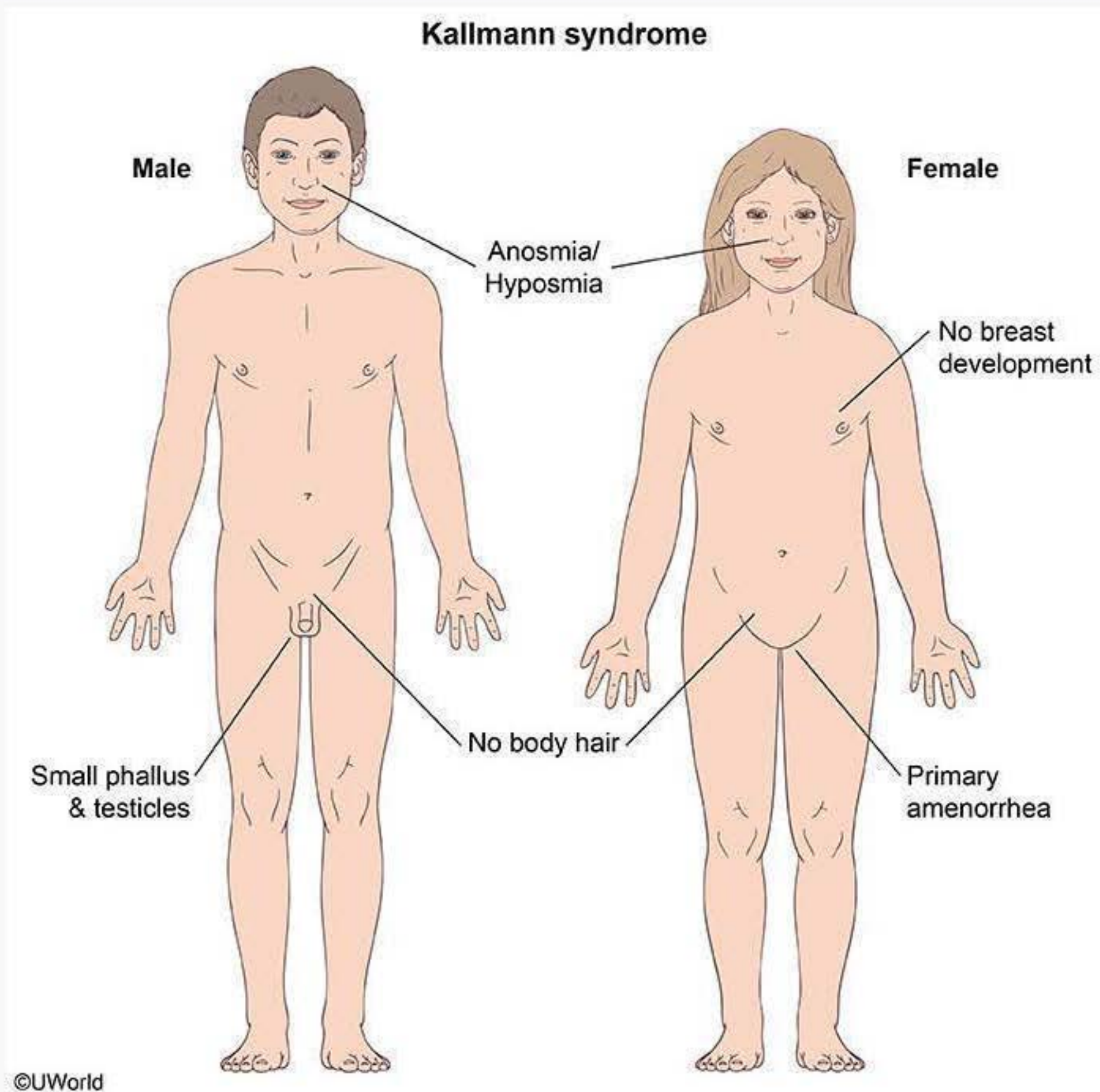
User Id: [REDACTED]

Kallmann syndrome



Explanation:

User Id: [REDACTED]



Kallmann syndrome is an X-linked recessive disorder of migration of fetal gonadotropin-releasing hormone (GnRH) and olfactory neurons, resulting in hypogonadotropic hypogonadism and rhinencephalon hypoplasia. Affected boys and girls have **normal genotype** and internal reproductive organs. However, the congenital **absence of GnRH secretion** results in **short stature** and **delayed or absent puberty**. Girls, such as this patient, may have primary amenorrhea and absent breast

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Typical laboratory findings include low follicle-stimulating hormone and luteinizing hormone levels. Early diagnosis is important as hormonal treatment can help facilitate development of secondary sex characteristics, build and maintain bone and muscle mass, and improve fertility.

(Choice A) Patients with Turner syndrome have the **genotype 45 XO**. These patients have primary amenorrhea, but the follicle-stimulating hormone level is generally elevated due to primary ovarian failure.

(Choice B) A 45 YO fetus is not a viable genotype.

(Choice D) Patients with androgen insensitivity or 5- α -reductase deficiency will have a female phenotype with a normal male genotype (46 XY). Patients with these disorders will not have ovaries.

(Choice E) Patients with 47 XXY have **Klinefelter syndrome**; they have a male phenotype that includes small testes.

Educational objective:

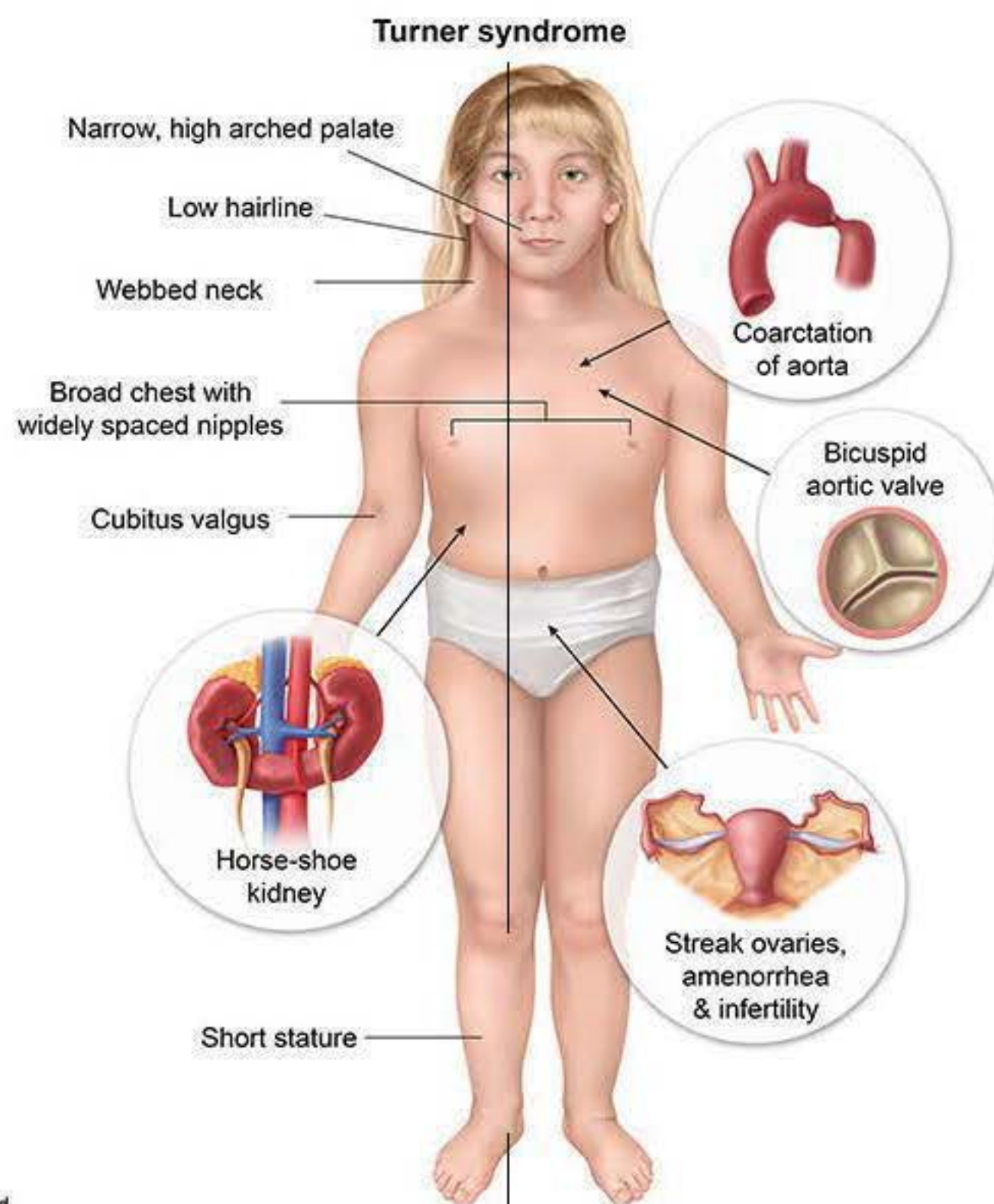
Patients with Kallmann syndrome present with delayed/absent puberty and anosmia. The karyotype will be consistent with their male or female phenotype. Follicle-stimulating and luteinizing hormone levels are low, consistent with gonadotropin-releasing hormone deficiency.

References:

1. **Diagnosis and evaluation of hypogonadism.**

Media Exhibit

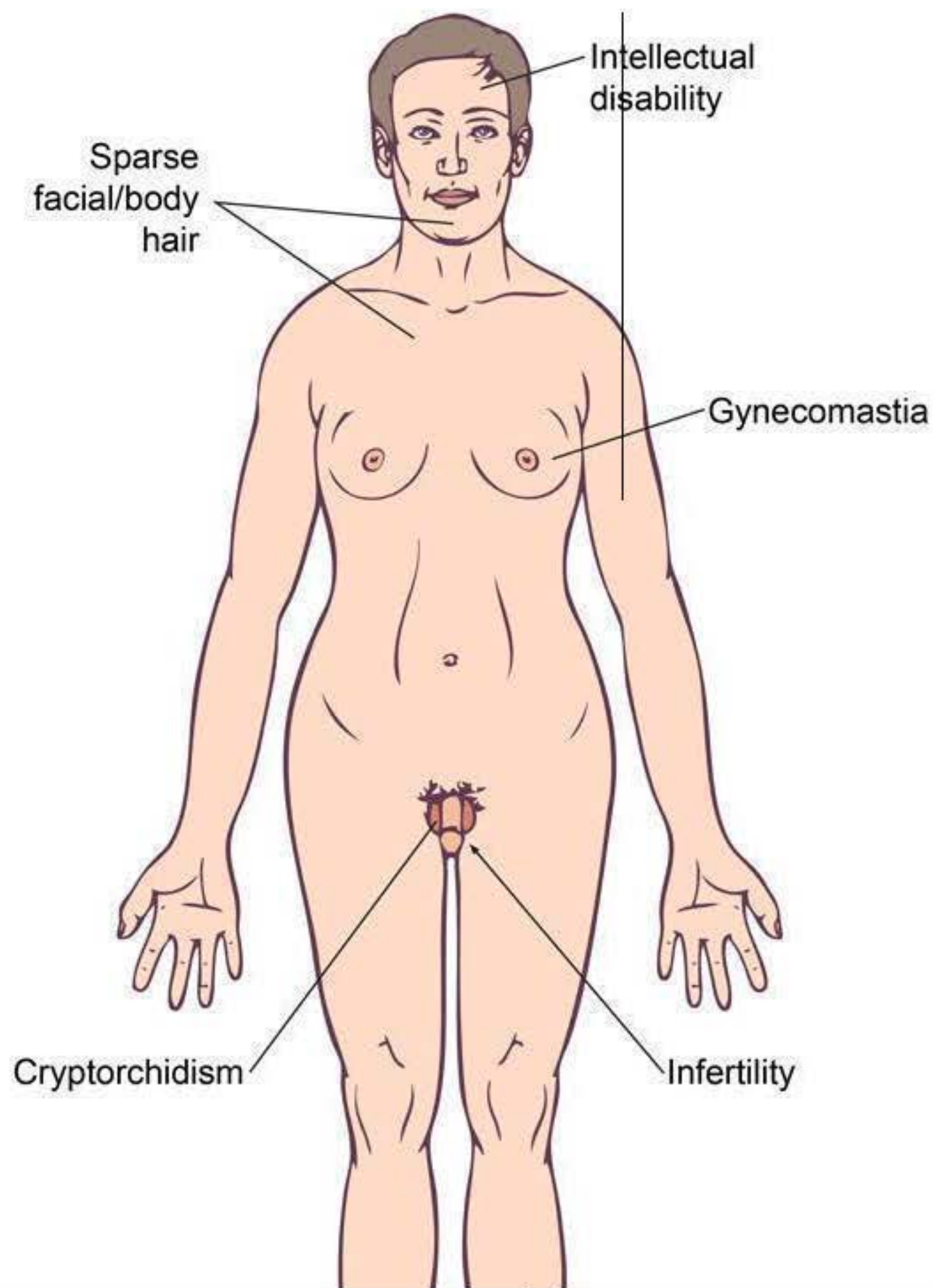
syndrome



Media Exhibit

ter syndrome

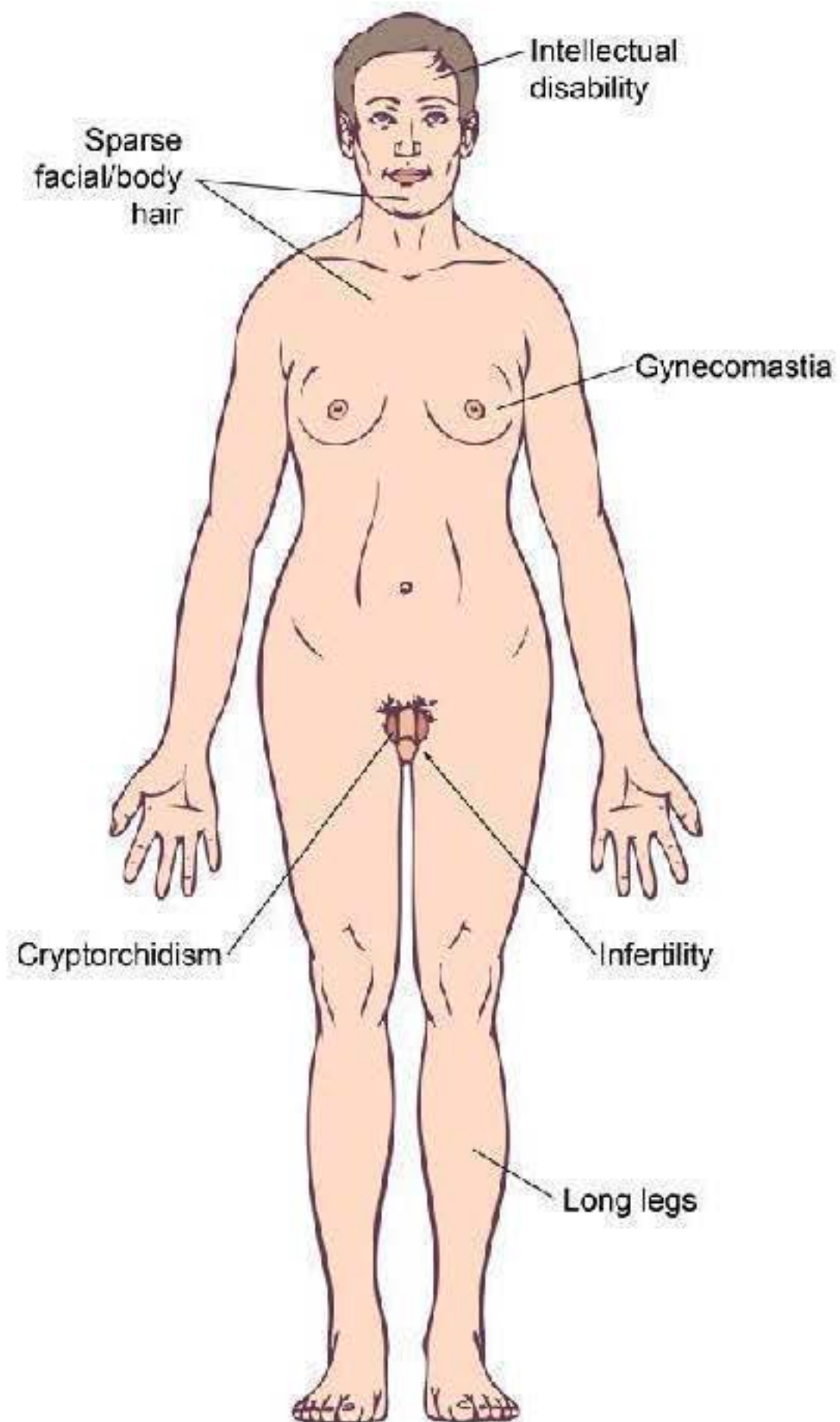
Klinefelter syndrome (47,XXY)



Media Exhibit

ter syndrome

Klinefelter syndrome (47,XXY)



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