

A 12-year-old girl is brought to the clinic for "being sad." Her parents report that she has become withdrawn lately, and they often hear her crying in her room. When interviewed alone, the girl says that she is being bullied at school. The other students underwent growth spurts over the summer, and they have started to ridicule her height since school started. The girl has always been shorter than her peers but has noticed that she is shorter than even her 8-year-old brother. Her father is 190.5 cm (6 ft 3 in) tall, and her mother is 183 cm (6 ft) tall. On examination, the patient's height and weight are at the 5th and 55th percentiles, respectively. Blood pressure is 140/90 mm Hg in the right upper arm and 90/40 mm Hg in the lower extremities. Given the most likely underlying diagnosis, which of the following is this patient most at risk of developing?

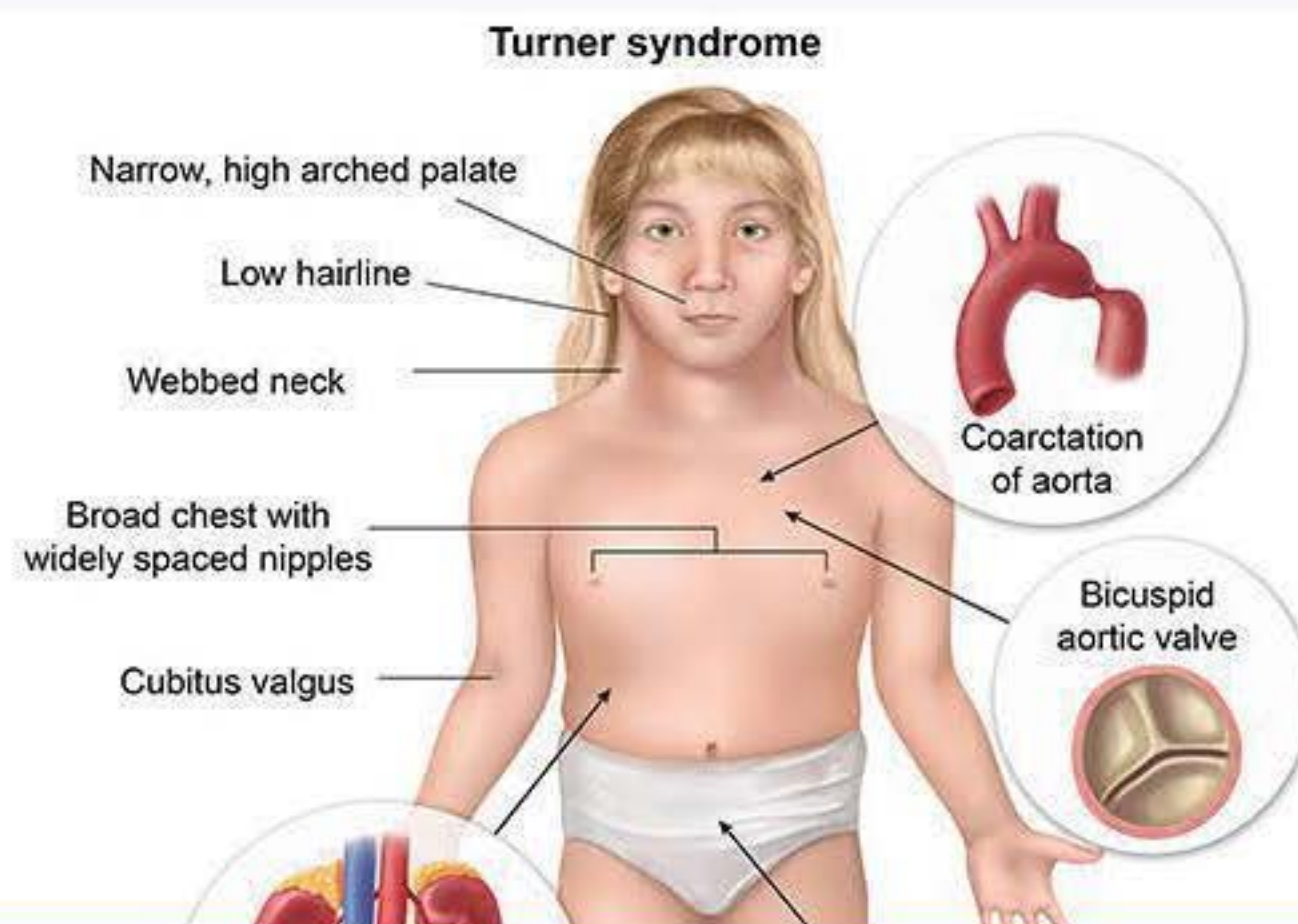
- ☐ A. Decreased bone mineral density
- ☐ B. Delayed cognitive development
- ☐ C. Malignant transformation of breast tissue
- ☐ D. Progressive decline of renal function
- ☐ E. Prolapse of mitral valve

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- ☒ A. Decreased bone mineral density [52%]
- ☐ B. Delayed cognitive development [8%]
- ☐ C. Malignant transformation of breast tissue [1%]
- ☐ D. Progressive decline of renal function [20%]
- ☐ E. Prolapse of mitral valve [19%]

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

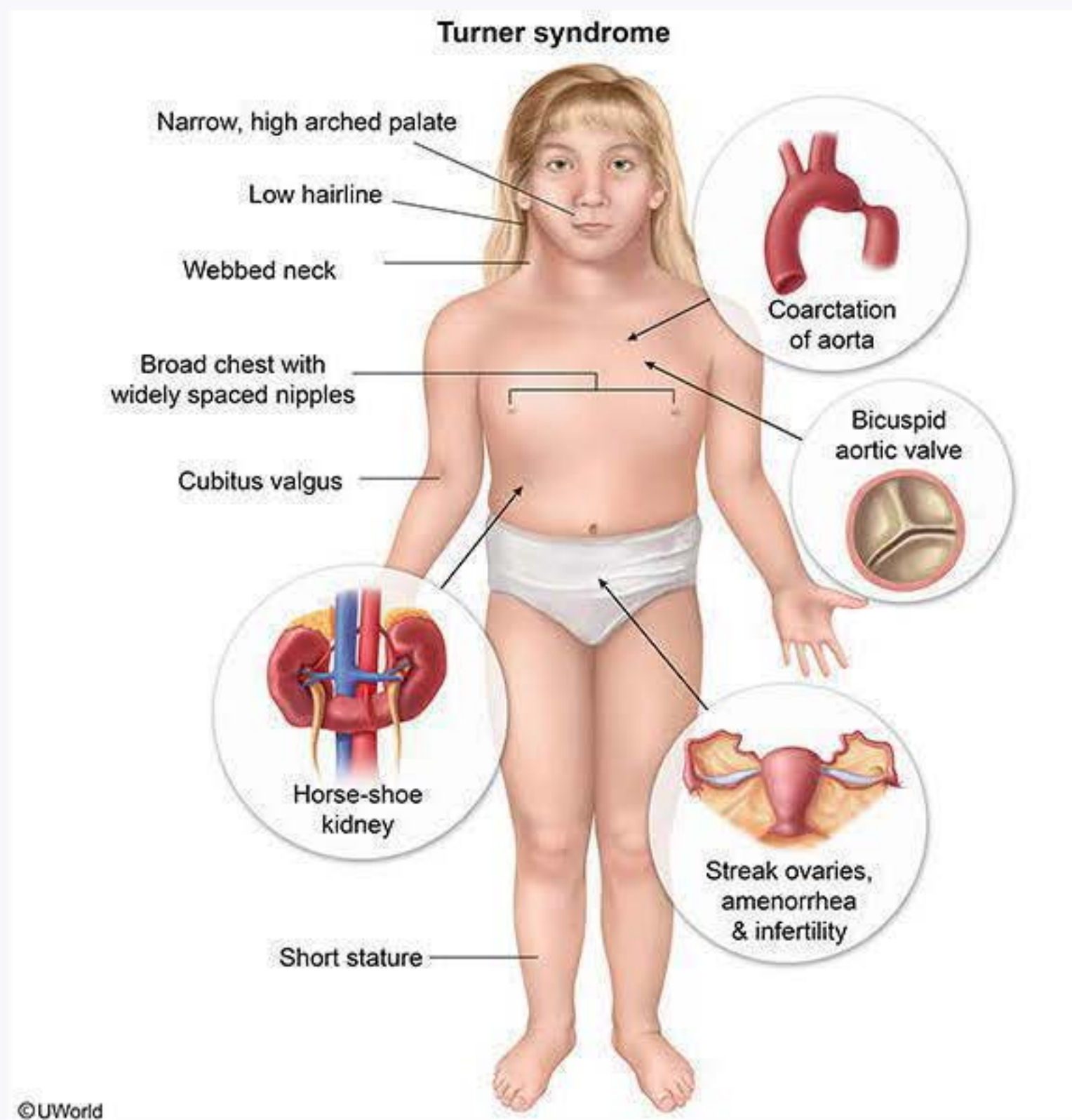
User Id: [REDACTED]



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Explanation:

User Id: [REDACTED]



This patient has a short stature and signs of aortic coarctation (higher blood pressures in upper versus lower extremities). The most likely diagnosis is **Turner syndrome (TS)**, a chromosomal abnormality due to loss of part or all of one X chromosome, most commonly with a **karyotype of 45,X**.

Patients with TS typically have **ovarian dysgenesis**, leading to "streak ovaries" (small amounts of tissue with little to no follicles) and premature ovarian failure. Because ovaries normally produce estrogen, patients with TS are **estrogen deficient**, which leads to minimal or no thelarche (breast development). Estrogen also normally serves to

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(Choice B) Most patients with TS have normal cognitive abilities. There is an increased risk of impaired nonverbal skills (eg, mathematics), attention-deficit disorder, and problems with executive functioning.

(Choice C) Patients with TS are at increased risk for certain malignancies (eg, meningioma, gonadoblastoma) but are at decreased risk for breast cancer. Because elevated estrogen levels are typically a risk factor for breast cancer, the decreased estrogen levels in patients with TS are likely protective.

(Choice D) Patients with TS are at increased risk of congenital renal abnormalities, including **horseshoe kidney** (fusion of kidneys). Although these structural abnormalities may increase the risk of urinary tract infections, renal function is typically normal in patients with TS.

(Choice E) Approximately half of all patients with TS will have cardiac abnormalities, most commonly a bicuspid aortic valve or coarctation of the aorta. Mitral valve prolapse is significantly less common in patients with TS and is seen more frequently in those with Marfan syndrome and Ehlers-Danlos syndrome.

Educational objective:

Short stature is the most common clinical finding in patients with Turner syndrome. These patients are at increased risk of osteoporosis due to estrogen deficiency from ovarian dysgenesis.

References:

1. **Efficacy of estrogen replacement therapy (ERT) on uterine growth and acquisition of bone mass in patients with Turner syndrome.**

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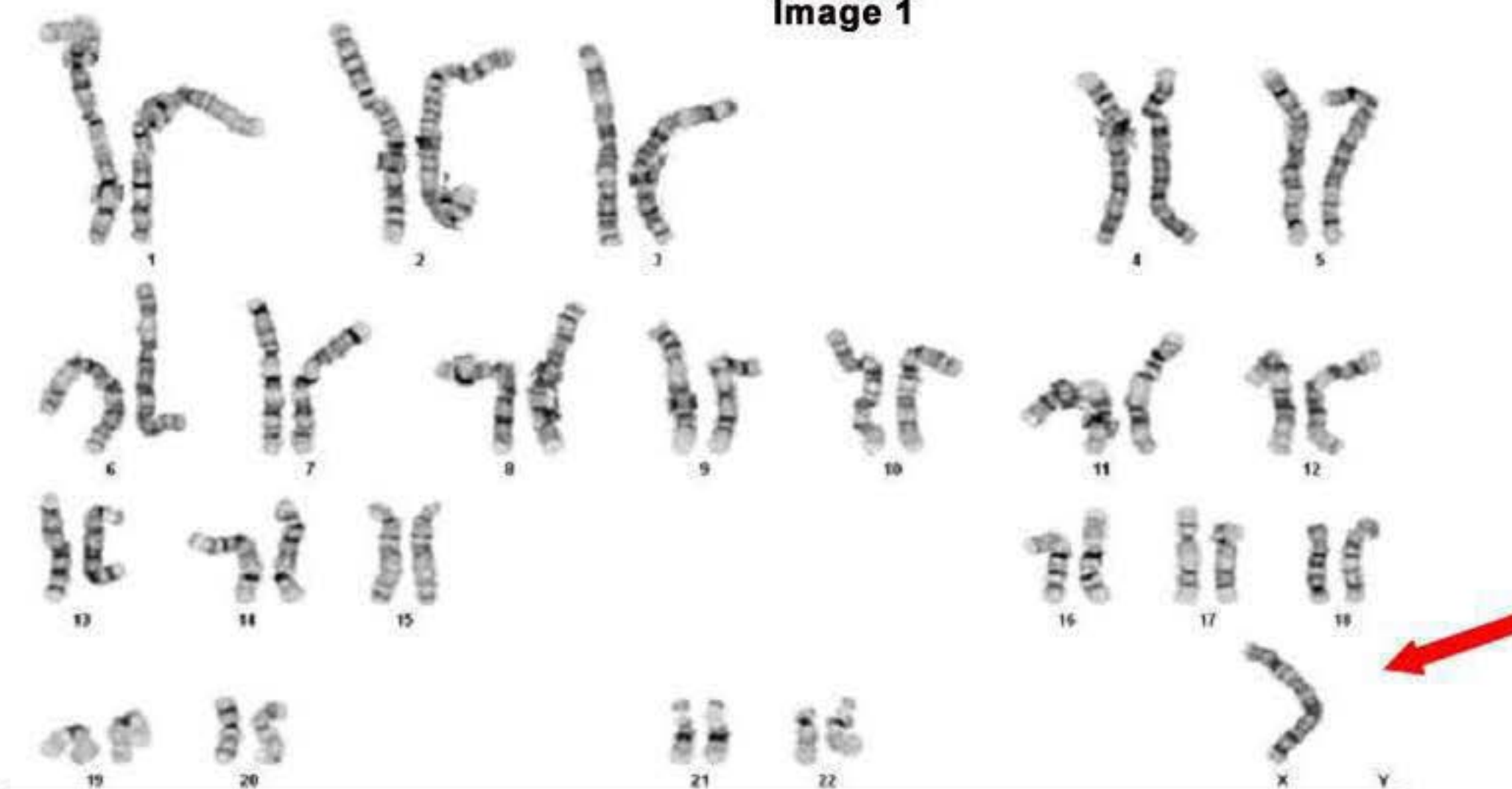
References:

1. **Efficacy of estrogen replacement therapy (ERT) on uterine growth and acquisition of bone mass in patients with Turner syndrome.**
2. **Bone health in children and adolescent with Turner syndrome.**

Media Exhibit

syndrome karyotype

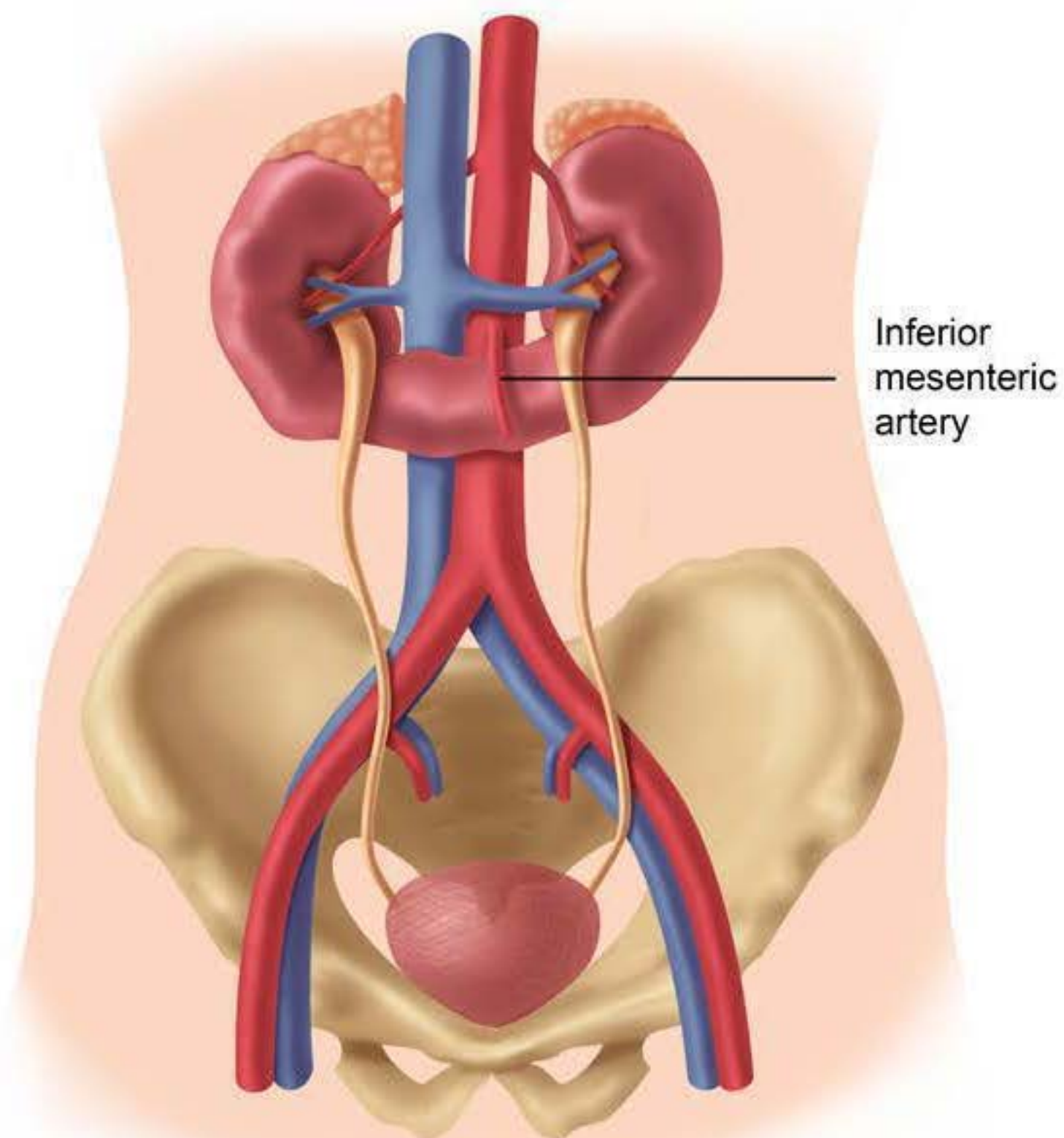
Image 1



Media Exhibit

hoe kidney

Horseshoe Kidney



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