

An 8-year-old female presents to your office because her mother has noticed "several physical changes." She denies headaches, emesis or visual disturbances. Her medical history is significant for two bone fractures which occurred at 6 and 7 years of age. Her family history is unremarkable. She has a healthy 15-year-old brother. She has a moonlike face. Physical examination reveals two light-brown spots with irregular contours on the back of the shoulders and left side of the neck. There is a bruise on the right arm, and elevation of the breasts with budding and enlargement of the areola. There is coarse, dark pubic hair along the labia and the pubic junction. The eye examination is normal. What is the most likely diagnosis?

- ☐ A. Von Recklinghausen disease
- ☐ B. McCune-Albright syndrome
- ☐ C. Peutz-Jeghers syndrome
- ☐ D. Sturge-Weber disease
- ☐ E. Cushing's disease
- ☐ F. Adrenal tumor



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- ☐ A. Von Recklinghausen disease [5%]
- ☒ B. McCune-Albright syndrome [50%]
- ☐ C. Peutz-Jeghers syndrome [2%]
- ☐ D. Sturge-Weber disease [2%]
- ☐ E. Cushing's disease [26%]
- ☐ F. Adrenal tumor [15%]

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

McCune-Albright syndrome is a rare condition characterized by precocious puberty, cafe au lait spots and multiple bone defects (polyostotic fibrous dysplasia). It is responsible for 5% of the cases of female precocious puberty, and may be associated with other endocrine disorders, such as hyperthyroidism, prolactin- or GH-secreting pituitary adenomas, and adrenal hypercortisolism. McCune-Albright syndrome is sporadic and has been recently attributed to a defect in the G-protein cAMP-kinase function in the affected tissue, thereby resulting in autonomous activity of that tissue.

**(Choice A)** Unlike in Von Recklinghausen syndrome, the cafe au lait spots of McCune-Albright syndrome are large, have irregular borders and are not associated with axillary or genital freckles.



- ☐ D. Sturge-Weber disease [2%]
- ☐ E. Cushing's disease [26%]
- ☐ F. Adrenal tumor [15%]

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McCune-Albright syndrome is a rare condition characterized by precocious puberty, cafe au lait spots and multiple bone defects (polyostotic fibrous dysplasia). It is responsible for 5% of the cases of female precocious puberty, and may be associated with other endocrine disorders, such as hyperthyroidism, prolactin- or GH-secreting pituitary adenomas, and adrenal hypercortisolism. McCune-Albright syndrome is sporadic and has been recently attributed to a defect in the G-protein cAMP-kinase function in the affected tissue, thereby resulting in autonomous activity of that tissue.

**(Choice A)** Unlike in Von Recklinghausen syndrome, the cafe au lait spots of McCune-Albright syndrome are large, have irregular borders and are not associated with axillary or genital freckles.

**(Choice C)** Peutz-Jeghers syndrome is characterized by gastrointestinal tract polyposis and mucocutaneous pigmentation. It may also involve the development of an estrogen-secreting tumor, leading to precocious puberty.

**(Choice D)** Sturge-Weber disease is a sporadic phakomatosis characterized by mental retardation, seizures, visual impairment and a characteristic port-wine stain over the territory of the trigeminal nerve.

**(Choice E)** This patient's moonlike facies and bruises indicate Cushing's syndrome, which occurs in association with McCune-Albright syndrome. Cushing's disease consists of hypercortisolism secondary to an ACTH-secreting pituitary tumor.

**(Choice F)** Adrenal tumors result most commonly in heterosexual precocious puberty (i.e., premature development of male secondary sexual characteristics in a female).

**Educational Objective:**

Remember the 3 P's of McCune-Albright syndrome: **precocious puberty**, **pigmentation** (cafe au lait spots) and **polyostotic fibrous dysplasia**.

Time Spent: N/A

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