McCune-Albright Syndrome Characteristics and Presentation

McCune-Albright syndrome is characterized by increased hormone production. It results from a sporadic activating mutation in the GNAS gene. Only some cells in the body harbor the mutations, a phenomenon termed mosaicism. Patients may present with precocious puberty, Cushing syndrome, or hyperthyroidism. Café-au-lait spots might be seen, and fibrous dysplasia can occur.



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Characteristics

Increased Hormone Production

Up-arrow Harmonica

Endocrinopathy is a characteristic of McCune Albright Syndrome, which presents with increased hormone production. It may include hyperthyroidism, Cushing's syndrome, growth hormone excess, renal phosphate wasting, and precious puberty.

Sporadic

Sporadic-spear

This disorder is due to sporadic mutation during embryogenesis.

Mutation in GNAS Gene

Mutant in G-NASA Jeans

McCune Albright syndrome occurs due to a mutation in the GNAS1 gene, which normally acts to promote Gs-protein ?-subunit activation (Gs?). It activates adenylyl cyclase, and its mutation will trigger continuous cAMP signaling.

Mosaicism

Mosaic Pattern

McCune-Albright syndrome is sporadic and occurs in only a few body cells, termed mosaicism. This characteristic can present with a variety of phenotypes. If the mutation occurs in the germline, the whole body will be affected and the disease is lethal. If it occurs after fertilization (somatic mosaicism), then all cells that are descendants from a particular cell in early development might be affected.

Clinical Features

Precocious Puberty

Pubescent-baby

Precocious puberty occurs due to hyperfunctioning endocrinopathy in McCune-Albright syndrome. It is defined as an early onset puberty (before age 8 years in girls and before age 9 years in boys).

Cushing Syndrome

Cushion

Cushing syndrome is the rarest endocrinopathy seen in McCune-Albright syndrome. It is characterized by excessive production of cortisol, resulting in slowed growth and weight gain in the face and upper body. Early mortality has been related to opportunistic infections.

Hyperthyroidism

Hiker-thigh-droid

Hyperthyroidism is the second most common endocrinopathy seen after precocious puberty in

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McCune-Albright syndrome. It can be seen in more than 50% of patients. Symptoms include weight loss, tachycardia, tremor, sweating, and high blood pressure.

Cafe Au Lait Spots

Coffee-cow

A Cafe Au Lait Spot is a brown macula with jagged/irregular borders that is commonly seen on the lower back and buttocks. It presents unilaterally in McCune-Albright Syndrome.

Fibrous Dysplasia

Fiber-sack Disc

Fibrous Dysplasia is characterized by the replacement of bone with collagen and fibroblasts. It is found in multiple skeletal sites and is mostly unilateral. The most common sites are long bones of arms and legs, the ribs, and the face and skull (craniofacial area). Painless swelling in the ribs is often seen in patients. This condition weakens bones and increases the risk of fracture.

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