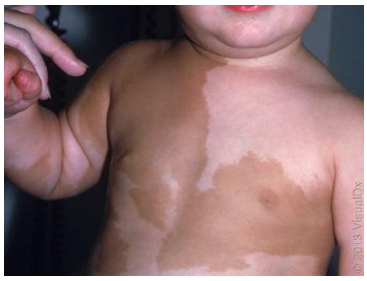
McCune-Albright Syndrome

What is McCune-Albright Syndrome?

McCune-Albright Syndrome encompasses three main components but can have several more features. The three main components are early onset of puberty, abnormal skin pigmentation called café-au-lait spots, and a bone disorder called fibrous dysplasia of the bone. Other features include other endocrine hormone abnormalities like increased secretion of thyroid hormones, growth hormone or cortisol. Other organs like the liver and heart can also be affected. Both boys and girls can be affected by this syndrome.

The three main components of the disorder are outlined in more detail below:

- Early puberty: This typically effects girls more commonly. Girls can experience vaginal bleeding (like a period) or breast development as young as age 2. This is typically caused by an increase in the hormone estrogen which is secreted by cysts that develop on the ovaries. Boys can also experience early puberty but this is rarer. Symptoms of early puberty in boys can include enlargement of the penis or testicles, and more wrinkles or rugae in the scrotum.
- Café-au-lait spots: These are light brown patches of skin that have an irregular, jagged border. This border is often compared to the coast of Maine given the jagged appearance. These spots may just appear on one side of the body.



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- Fibrous dysplasia of the bone: This term refers to the replacement of healthy bone with scar-like bone. This can cause the bones to be weak and fracture. People with this disorder may have uneven growth or a deformed appearance to the bone. These lesions are often on one side of the body.

What causes McCune-Albright Syndrome?

There is a mutation in the *GNAS* gene. This gene is needed for building a larger protein called a G protein. This mutation causes the G protein to be excessively active which leads to the over production of hormones and abnormal bone growth.

How does a person acquire McCune-Albright Syndrome?

This is not a syndrome that is passed down in families. It is the result of a sporadic or random mutation.

How is McCune-Albright Syndrome diagnosed?

If your child's doctor has a clinical suspicion for this syndrome, they may check your child's hormone levels and order imaging of the bones to see if fibrous dysplasia is present. There are genetic tests available but typically the diagnosis is made by identifying classic physical exam and laboratory findings in the child.

How is McCune-Albright Syndrome treated?

Each individual aspect of McCune-Albright Syndrome is treated separately. At this time, there is no gene therapy to address the underlying *GNAS* mutation.

- The goal of treatment in precocious puberty in young girls is to decrease estrogen levels. A type of drug called an aromatase inhibitor can be used. If central precocious puberty develops, meaning the brain starts secreting puberty hormones (LH and FSH) which trigger the ovaries to secrete more estrogen, then a class of medications called a GnRH agonist is used which puts a pause on pubertal development.
- Given how rare early puberty is in young men with MAS, the treatment plan is not as well defined. Typically a class of medications called anti-androgens is used. Aromatase inhibitors may also be used. These medications decrease different puberty hormone levels. If a young boy is experiencing central precocious puberty, then a GnRH agonist would be used similar to in young girls.
- If a patient has excess secretion of other hormones (thyroid hormone, growth hormone or cortisol), these are addressed on an individual basis.
- There is no specific treatment for the fibrous dysplasia. Surgery may be needed to address a fracture or abnormally shaped bone. A medication called a bisphosphonate may be used if the patient is experiencing significant bone pain. Adequate calcium, vitamin D, and phosphorus are important to maintain good bone health.