McCune-Albright syndrome

Summary

- McCune-Albright syndrome is a genetic disease that affects bone growth, skin pigmentation and hormone balance.
- The cause of McCune-Albright syndrome is a genetic mutation, but the reason for the mutation is not known.

McCune-Albright syndrome is a genetic condition that affects bone growth, skin pigmentation and the body’s hormone balance. Deformed, easily broken bones and premature sexual maturity are typical signs of the condition. Symptoms range in severity.

In some cases, the bone abnormalities and hormone problems are severe enough to be obvious in infancy. In other cases, the child seems healthy in all respects. The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict. There is no cure.

Symptoms of McCune-Albright syndrome

The symptoms and signs of McCune-Albright syndrome include:

- polyostotic fibrous dysplasia – the abnormal growth of two or more bones. Bones of the face, skull, arms and legs are commonly affected
- café au lait birthmarks, usually on the abdomen or back – these birthmarks are flat, oval-shaped and light brown (milk-coffee coloured) spots that have been described as having a ‘Coast of Maine’ (jagged) outline. Café au lait birthmarks are frequently the first signs of the condition and appear at or shortly after birth
- premature sexual maturity. This can affect girls and boys, but is much more common in girls
- onset of the menstrual period before the growth of breasts or pubic hair
- ovarian cysts, which are thought to trigger early menstruation
- premature sexual maturity in boys – this is less common but may still occur
- symptoms of an overactive thyroid gland (hyperthyroidism).

Complications of McCune-Albright syndrome

Some of the complications of McCune-Albright syndrome can include:

- brittle, warped bones (osteoporosis)
- repeated fractures
- rickets
- blindness or deafness due to abnormal growth of skull bones
- adrenal gland tumours
- osteitis fibrosa cystica – soft bones that are prone to developing cysts
- mobility problems or disabilities caused by abnormal bone growth or asymmetry of the skeleton
- in rare cases, excessive growth (gigantism) because of growth hormone imbalance – the face, hands and feet are commonly affected
- Cushing syndrome.

Cushing syndrome
The hormone cortisol is made by the adrenal glands, which are located on the kidneys. McCune-Albright syndrome may cause enlargement of the adrenal glands and the overproduction of cortisol. This causes a range of symptoms collectively known as Cushing’s syndrome.

Some of the symptoms may include:

- obesity
- weight gain around the abdomen
- wasting of the limbs
- a hump of fat high on the back
- round, red and puffy face
- thin, easily bruised skin
- slow healing of skin wounds
- skin ulcers
- arrested growth in childhood.

**Causes of McCune-Albright syndrome**

McCune-Albright syndrome is caused by a mutation in the gene called GNAS1. The mutation causes the GNAS1 gene to become more active than normal. The gene mutation is not inherited from the parents, nor is it present at the time of conception. The mutation actually occurs in an early cell division in the developing embryo. As a result, the mutation is present in some, but not all, cells of the person’s body. This is known as ‘mosaicism’.

Despite hormonal irregularities, the majority of women with McCune-Albright syndrome are fertile. A person with McCune-Albright syndrome cannot pass the condition on to their children. The cause of the genetic mutation is not known.

**Diagnosis of McCune-Albright syndrome**

The tests used to diagnose McCune-Albright syndrome may include:

- medical history
- physical examination
- blood tests to check hormone levels of the thyroid, parathyroid, adrenal glands and pituitary gland
- x-rays to examine the skeleton
- bone scan using an injection of radioactive material
- other scans, such as MRI
- genetic tests.

**Treatment for McCune-Albright syndrome**

There is no cure for McCune-Albright syndrome. Treatment may include:

- medication to manage pain
- surgery to bolster affected bones with grafts, pins, casts or a combination of procedures
- treatment with drugs known as bisphosphonates to encourage bone density
- vitamin D supplements to reduce the risk of rickets
- regular and supervised exercise program to encourage bone density
- physical therapy to maintain or improve joint mobility
- medication to reduce levels of the female sex hormone oestrogen
- surgery to remove ovarian cysts
• surgery to remove abnormal adrenal glands in the case of Cushing syndrome
• hormone therapy or surgery to treat a pituitary tumour
• hormone therapy or surgery to treat gigantism.

**Treatment team for people with McCune-Albright syndrome**

Treatment depends on the person’s age, general health and severity of the symptoms. Specialist doctors may include an orthopaedic surgeon to treat bone problems, a plastic surgeon to treat facial deformities and an endocrinologist to treat hormone imbalances.

**Where to get help**

• Your doctor
• Orthopaedic surgeon
• Plastic surgeon
• Endocrinologist

**Things to remember**

• McCune-Albright syndrome is a genetic disease that affects bone growth, skin pigmentation and hormone balance.
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