
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 change, but the reason for the change is not known.
 - McCune-Albright syndrome is not an inherited disease.
 - The gene change that causes it occurs in the developing embryo.
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McCune-Albright syndrome is a genetic condition that affects bone growth, skin pigmentation and the body's hormone balance. Bone abnormalities such as 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 currently no cure for McCune-Albright syndrome.

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 spots with a jagged outline. Café au lait birthmarks are often the first signs of the condition and appear at or shortly after birth. They are light brown (or 'milk-coffee coloured') in people with lighter skin. In people with darker skin they may be harder to see
- 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
- 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's syndrome**.

Cushing's 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.

Read more about [Cushing's syndrome](#).

Causes of McCune-Albright syndrome

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

Despite hormonal irregularities, most 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 change 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 medicines known as bisphosphonates to improve bone density
- vitamin D supplements to reduce the risk of rickets
- regular and supervised exercise program to improve 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's 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.

Genetic counselling and McCune-Albright syndrome

If your child has been diagnosed with McCune-Albright syndrome, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand McCune-Albright syndrome and what causes it, and what a diagnosis means for your child's health and development. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by McCune-Albright syndrome.

Where to get help

- Your **GP (doctor)**
- **Orthopaedic surgeon**
- **Plastic surgeon**
- **Endocrinologist**
- **Genetic Support Network of Victoria (GSNV) Tel. (03) 8341 6315**
- **Victorian Clinical Genetics Services (VCGS) Tel. 1300 118 247**

This page has been produced in consultation with and approved by:

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