

## McCune-Albright syndrome

McCune-Albright syndrome is a genetic disease 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 disease.

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 normal and 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

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
- Premature sexual maturity 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).

### Possible complications

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.

## A gene mutation

McCune-Albright syndrome is caused by a mutation in the gene known as GNAS1. The mutated gene is found in some, not all, cells of the person's body. This is known as 'mosaicism'. The mutation happens spontaneously during the development of the baby in the womb, which means the condition is not inherited from the parents.

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

## Diagnosis methods

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 options

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

- Drugs 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
- Drugs 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

Treatment depends on the patient's age, general health and severity of the symptoms. Specialist doctors may include:

- **Orthopaedic surgeon** – to treat bone problems.
- **Plastic surgeon** – to treat facial deformities.
- **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.
- The cause is a genetic mutation, but the reason for the mutation is not known.

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Genetic Health Services Victoria

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