McCune-Albright syndrome

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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.
- The cause of McCune-Albright syndrome is a genetic mutation, but the reason for the mutation is not known.

References


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More information

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A-Z of genetic conditions

Ambiguous genitalia
The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.

Angelman syndrome
The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.

Ankylosing spondylitis
Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.

Barrett's oesophagus
Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.

Bipolar disorder
Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.

Central nervous system birth defects
Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.

Charcot-Marie-Tooth disease (CMT)
Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

Cleft palate and cleft lip
Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

Congenital adrenal hyperplasia (CAH)
CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

Creutzfeldt-Jakob disease (CJD)
Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

Cri du chat syndrome
Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.

Cystic fibrosis (CF)
When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.

Digestive tract birth defects
Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

Down syndrome
With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

Dwarfism
Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

Eczema (atopic dermatitis)
Eczema can vary in severity, and symptoms may flare up or subside from day to day.

Essential tremor
Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

Fragile X syndrome

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The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic...

- **Friedreich's ataxia**
  
  To the casual observer, a person with Friedreich ataxia may seem to be drunk...

- **Genetic factors and cholesterol**
  
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol...

- **Haemochromatosis**
  
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses...

- **Haemophilia**
  
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed...

- **Hair**
  
  Human hair grows one centimetre every month...

- **Hearing problems in children**
  
  The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development...

- **Huntington's disease**
  
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age...

- **Kabuki syndrome**
  
  Kabuki syndrome affects males and females equally and there is no cure...

- **Kennedy's disease**
  
  Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs...

- **Kidneys - medullary cystic kidney disease**
  
  Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys...

- **Kidneys - polycystic kidney disease (PKD)**
  
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds...

- **Klinefelter syndrome**
  
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur...

- **Leukodystrophy**
  
  Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions...

- **Long QT syndrome**
  
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement...

- **Marfan syndrome**
  
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious...

- **McCune-Albright syndrome**
  
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict...

- **Muscular dystrophy**
  
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs...

- **Neonatal syndrome**
  
  Neonatal syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features...

- **Osteoporosis in children**
  
  Osteoporosis in children is rare and usually caused by an underlying medical condition...

- **Phenylketonuria (PKU)**
  
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods...

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- Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

- Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

- Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

- Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

- Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- Thalassaemia
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- Tourette syndrome
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

- Treacher Collins syndrome
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

- Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

- Turner's syndrome
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

- Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

- Von Willebrand disease
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

- Williams syndrome
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

- Wilson disease
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

Genes and genetics explained
- Genes and genetics explained
  Children inherit physical characteristics such as eye colour from their parents through their genes.

- Gene therapy
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

- Genetic disorders
  genetic disorder is caused by an altered or faulty gene or set of genes.

- Genetic services in Victoria
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

Genetic testing
- DNA profiling
  DNA profiling is a way of establishing identity and is used in a variety of ways.
Egg freezing

You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances...

Genetic services in Victoria

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Genetic testing for inherited cancer

A predisposition to certain cancers can be inherited via altered genes...

Newborn bloodspot screening

Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions...

Pregnancy tests – chorionic villus sampling

Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities...

Pregnancy tests - maternal serum screening

Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis...

Related Information

- Eczema (atopic dermatitis)
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- Spinal muscular atrophy (SMA)
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- Raynaud's phenomenon
  Raynaud's phenomenon can be a sign of a more serious underlying condition, so see your doctor if you experience it...

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- All Doctors

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