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Wilson disease is a relatively rare genetic disorder that prevents the body from eliminating copper. The build-up of copper damages certain structures including the liver, nervous system, brain, kidneys and eyes. Wilson disease is fatal without medical treatment. There is no cure, but the condition can be managed.

Treatment options include medications, chelation therapy and avoiding foods high in copper.

Metabolism refers to the countless and ongoing chemical processes inside the body that allow life and normal functioning. Wilson disease is a rare genetic disorder, an 'inborn error of metabolism' that prevents the body from eliminating copper. The build-up of copper in the body damages certain organs including the liver, nervous system, brain, kidneys and eyes. In around half of cases, only the liver is affected. The copper begins to accumulate at birth, but symptoms appear most commonly during the teenage years. Without treatment, the copper poisoning is fatal. There is no cure, but Wilson disease can be managed.

Around one in 30,000 people have Wilson disease. It is caused by faults in a gene called ATP7B, which must be inherited from both parents for the disease to be expressed. Wilson disease is also known as hepatolenticular degeneration.

Symptoms of Wilson disease

Symptoms of Wilson disease can appear in individuals ranging from three years to over 50 years of age. Symptoms depend on which structures of the body are affected by copper poisoning, but can include:

- enlarged abdomen
- abdominal pain
- loss of appetite
- vomiting of blood
- splenomegaly (enlarged spleen)
- jaundice (yellowed eyes and skin)
- hand tremors
- stiffness and reduced movement of the extremities
- the movement of the body is jerky, slow and difficult to control
- speech difficulties
- personality changes
- symptoms of mental illness, such as depression or homicidal tendencies
- lack of menstruation (amenorrhoea)
- confusion
- dementia.

Cause of Wilson disease

Enzymes are special proteins that help to spark chemical reactions in the body. The removal of excess copper relies on a particular enzyme, but this enzyme is defective in people with Wilson disease. Copper deposits then build up inside the body and cause damage.

Wilson disease is inherited in an autosomal recessive manner. This means that in order for a person to be affected by Wilson disease, they need to inherit two faulty genes, one from each parent. If both parents carry a faulty gene for Wilson disease, each child has a one in four chance of inheriting both faulty genes and being affected by Wilson disease. If only one faulty gene is inherited, the child is a carrier but won’t develop any symptoms.

Most people with Wilson disease have no family history of the disorder.

Effects of copper on the body

Copper is common to a wide range of foods, but the human body needs only tiny amounts. The rest is excreted. In people with Wilson disease, the excess copper leaves the bloodstream and settles in various organs and structures, including the brain, spinal cord, eyes, liver and kidneys.

Copper is a toxic substance in large amounts, and causes damage to your tissues. The damaged tissues die and are replaced by scar tissue. As more and more tissue is replaced by scars, the affected organ loses its ability to function until it eventually fails.

Complications of Wilson disease

Without medical treatment, complications of Wilson disease can include:

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liver disease, such as hepatitis, cirrhosis or necrosis (death of the tissue)
increased susceptibility to infections
disorders of the spleen
anaemia
muscle atrophy
increased susceptibility to bone fractures
permanent physical disability
permanent intellectual disability
death.

Diagnosis of Wilson disease

Wilson disease is diagnosed using a number of tests, including:

- physical examination
- medical history
- eye examination to check for Kayser-Fleischer rings (brown rings outside the iris)
- blood tests
- urine tests
- genetic testing to detect mutations in the ATP7B gene
- magnetic resonance imaging (MRI)
- biopsy of affected organs, particularly the liver.

Treatment of Wilson disease

Without treatment, Wilson disease is fatal. The longer the copper poisoning continues, the harder it is to successfully treat, so early diagnosis is important. The aims of treatment are to reduce the amount of copper in the body and control the symptoms. Treatment must be lifelong. Death can occur in a matter of months if the treatment is stopped. Options may include:

- vitamin B6 to bolster the nervous system
- potassium supplements, taken before eating, to reduce the absorption of dietary copper
- zinc therapy to prevent the absorption of copper in the small intestine
- switching to a diet low in copper
- chelation therapy, which is the use of medications (such as penicillamine) that bind to copper and allow it to be excreted in the urine
- regular blood and urine tests to check copper levels so that treatment can be adjusted if necessary
- liver transplant in severe cases
- genetic counselling and testing for the family.

Low copper diet

Management of Wilson disease is lifelong. It is important to switch to a low copper diet. Some of the foods to avoid include:

- chocolate
- dried beans
- dried fruits
- mushrooms
- nuts
- offal such as liver
- peas
- shellfish
- whole wheat products.

Where to get help

- Your GP

References

- About Wilson’s disease [online information], Wilson’s Disease Association International, Brookfield, USA. More information here.

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More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing
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 palletes 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 varvus degrees of speech delay and some health problems.

- **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**
  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.

- Neurofibromatosis
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- Noonan syndrome
  Noonan 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.

- 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

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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.

**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.

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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

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Home

Related information on other websites

- Genetic support network victoria.

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Last updated: February 2018

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