CPA FACT SHEET

Genetic Conditions

Cerebral Palsy ALLIANCE

What are genetic conditions?

Some genetic conditions are inherited, while others are caused by random faults in one or more genes in the person's body.

A gene is a part of a cell in your body that provides instructions for life. Before you are born, it tells your cells what colour your eyes should be, whether your hair should be straight or curly, and how your heart should develop.

Once you are born, your genes keep providing instructions all through your life. But they now have to compete with all the other influences around you, such as what you eat, what you drink, what education you are offered and more.

Types of genetic conditions

There are many different genetic conditions, most of which are rare, including Down Syndrome, Angelman syndrome, muscular dystrophy and Rett syndrome. Genetic disorders can be classified like this:

Single gene conditions, where the person inherits a single abnormal or mutated gene. Examples are muscular dystrophies, Huntington's disease and phenylketonuria.

Complex gene conditions, where the person inherits two or more mutated genes which sometimes combine with environmental or lifestyle factors to cause a disease. These diseases tend to run in families. Examples are some forms of spina bifida and other neural tube defects, some forms of cerebral palsy and some forms of hydrocephalus.

Chromosomal conditions, where the person has damaged, missing or extra chromosomes. Examples are Turner syndrome, cri-du-chat syndrome and Klinefelter Down Syndrome.

Causes

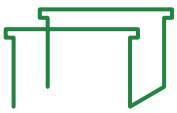
Some genetic conditions are inherited from one or both parents. The parent might or might not have known that they had a faulty gene or chromosome.

Other genetic conditions occur because a problem occurs with one or more genes or chromosomes during conception and fertilisation.

Symptoms

Some babies are born with obvious signs of a genetic condition, such as a physical deformity or a neurological problem. For many others, genetic conditions only start showing as the child is growing up, or in the teenage or adult years.

When a genetic condition begins in infancy, the signs that something is wrong are often first noticed by the baby's parents. Their fears may be confirmed during routine checks of whether the child is meeting normal developmental milestones. With other genetic disorders, the first signs may appear anywhere from childhood to middle age.



At CPA we help babies, children, teenagers and adults living with neurological and physical disabilities lead the most comfortable and independent lives possible. Our therapy teams work with individuals and families in many ways. Get in touch to find out how we can support you and improve your child's development.

Diagnosis of genetic conditions

An affected child will generally see a GP, before being referred to a specialist. Some may also see a geneticist or a genetic counsellor. The health professionals will take a family and medical history and examine the person. They will also order tests, which might include:

- genetic tests
- blood tests
- X-rays, CT scans and ultrasounds
- biopsies, where a small sample of tissue is removed and tested in a laboratory.

Some genetic conditions can be picked up fairly easily, but rarer conditions may take many months or even years to diagnose.

Some genetic conditions can be tested for, and it can be good to know what's going on. But for families thinking about having genetic testing, it's very important to talk to someone about it first like a genetic counsellor, who is an expert in genetics.

Living with genetic conditions

Having a genetic condition might or might not have a significant impact on the life of the person affected, and on the lives of people around them.

Having a genetic condition might mean the future looks less certain, and there might be effects on work, relationships, family and independence of carers.

Talking to doctors and a genetic counsellor can be helpful to learn more about the genetic condition and the therapies, services and treatments available. Reliable sites include Healthdirect and Genetic Alliance Australia. American sites such as GARD and the Rare List also have a lot of information on rare genetic conditions.

Talking to a psychologist, a counsellor or a social worker can also be helpful for psychological and emotional support.

Treatment and therapy

Treatment will depend on the particular condition and how it affects a child. They should be cared for by a team of people, led by a GP. Others include:

- specialists
- a physiotherapist who helps with movement and mobility
- an occupational therapist who can help with advice around mobility and independence
- a social worker, psychologist or a counsellor to help manage both the practical and the emotional aspects of the condition
- other allied health professional such as a speech therapist, a physiotherapist or an occupational therapist.

There is no cure for genetic conditions. But there is often a lot that can be done to manage symptoms, keep disability to a minimum, prevent complications and improve quality of life.

Early intervention programs are often recommended for babies and young children with genetic conditions.

Interpreters are available to support you through all of your appointments. Interpreters are free of charge from the government and can be included in your NDIS plan.

Get in touch with us:

Call us on 1300 888 378

Email us at ask@cerebralpalsy.org.au

Visit cerebralpalsy.org.au

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