



# Joubert's syndrome

**Joubert's syndrome is a rare disorder affecting the brain causing varying degrees of physical, mental and, sometimes, visual impairments.**

## What is the cause?

In Joubert's syndrome two parts of the brain (the cerebellar vermis and the brainstem) do not develop completely during pregnancy due to a faulty gene. The cerebellar vermis is responsible for control of posture and muscles and of head and eye movements. The brainstem controls functions such as heartbeat, breathing and temperature control. Underdevelopment or absence of these structures inevitably affects their functions. Some, but not yet all, causative genes have been identified.

## How is it diagnosed?

Diagnosis is confirmed by MRI brain scan appearances and can sometimes be confirmed by genetic testing on a blood sample.

## Does it have an alternative name?

The condition was first described by Dr Joubert in 1969; prior to this, the various features seen in Joubert's syndrome were not recognised as being a specific syndrome (group of symptoms with a single cause occurring in different people).

## Is it inherited?

In some cases Joubert's syndrome appears to be sporadic (just occurring in that particular individual) but it is more often inherited as an autosomal recessive disorder, which means that both parents are carriers of the disease. Human beings have about 30,000 to 40,000 different genes, each of which has a function in making an individual person. The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes. Inevitably some of these genes are faulty. A normal gene can overcome a faulty one, but if both genes in the pair are faulty, the genetic instructions cannot work. Most people carry different faulty genes but in Joubert's syndrome (and other recessive conditions) parents, though healthy themselves, carry the same faulty genes, and risk passing them on to their children. Each pregnancy carries

- a **25 per cent** chance of the child being **affected**
- a **75 per cent** chance of the child **not** being **affected**.

## Is prenatal testing available?

The brain underdevelopment may be detected on fetal ultrasound performed at 18 to 20 weeks of pregnancy. If a specific gene has been identified in an affected child in the family then this gene can be tested for in a blood sample.

## How common is it?

There is no data available on incidence.



## How does the condition present and progress?

Children with Joubert's syndrome can have very variable features and problems, from relatively mild to very severe. All babies are likely to have elements of the following:

- Some children may develop epilepsy.
- Many infants have a somewhat characteristic appearance, with a relatively large head, prominent forehead, broad nose and a tendency to keep their mouths open and their tongues out.
- Floppiness (hypotonia), unsteadiness (ataxia) and developmental or learning delay are present to some extent in all the children; the combination of these factors make sitting and standing delayed or very difficult to achieve.
- Jerky eye movements are likely to be obvious from early infancy and some babies will have very poor vision, or difficulty moving the eyes to look at an object (oculomotor apraxia).
- Some children may develop epilepsy.
- Learning difficulties are common especially surrounding speech and some children may be severely disabled and/or have difficult behaviour problems.
- Some children have kidney cysts, which may affect kidney function.
- An abnormal breathing pattern may be noticed soon after birth. This may improve with age but, sadly, very severely affected infants may die within the first few years of life from breathing problems; this is rare.

All these problems are the result of the brain malformation rather than part of an ongoing disease process and therefore, unless they are at risk from the breathing problem, the children are likely to have the potential to make positive progress in all areas as they get older.

## Is there any treatment?

There is no treatment that can cure the underlying brain malformation and the resulting effects on the rest of the body. Medication may be given to help with the epilepsy, if it occurs, and from the age of a few months various therapies can be introduced and taught to parents to help the children achieve their maximum potential both physically and mentally. When they reach school age learning support and therapies can be integrated into their school day and there are increasingly sophisticated methods of enhancing communication if speech is difficult to achieve.

## Is any research being done?

Research is progressing rapidly in the genetic field, finding out what all the different genes are responsible for so that 'faulty' genes can be recognised. In some cases, this is already enabling earlier and more precise prenatal testing and this may, in the longer term future, lead to replacement of the faulty gene(s). Unfortunately the brain underdevelopment could not be altered so for children already affected research ideas that concentrate on increasing the child's developmental potential and independence are most important. While medical treatment of any neurological disorder is very difficult because of the complexity of the brain, enormous strides are being made in the development of educational and therapy strategies and equipment.

## Is there a support group?

There is no formal support group for families of children with Joubert Syndrome but Contact a Family may be able to provide telephone support and contact with other families. Telephone their helpline on 0808 808 3555 or visit their website at [www.cafamily.org.uk](http://www.cafamily.org.uk).