

## Why did this happen? Can it happen again?

To answer these questions, the parents' and affected child's chromosomes need to be tested. What is certain is that, as a father or mother, there is nothing you did to cause the inv dup del 8p and nothing you could have done which would have prevented it. It is no one's fault.

Inv dup del 8p can occur when there are no rearrangements in the parents' chromosomes and tests have shown that the parents' chromosomes are unaffected. This is referred to as "de novo" (dn) by geneticists, meaning the duplication and deletion have occurred as a new event in the child. In these cases, the chances of having another affected child are usually no higher than for anyone else in the population.

Most of the DNA that makes up chromosome 8p is present as a unique sequence, but the presence of two repeated sections of DNA on chromosome 8p are believed to be responsible for an extremely common inversion of 8p in the general population, which is found in 39% of the typical Japanese population and 26% of Europeans. This does not affect a person's health or development and there is no reason why they should know about it unless they have a baby with a chromosome disorder.

This inversion has been found in many of the small number of mothers of inv dup del 8p children tested for it, including several *Unique* members. The presence of this common inversion can be shown by chromosome tests on the parents and means that in theory if you have had a child with inv dup del 8p you could be at risk of having another. However, the sequence of events that leads to the inv dup del 8p chromosome is still extremely rare, even if the harmless inversion on chromosome 8 is common.

Any concerns should be discussed with your Clinical Genetics Service. It is important to remember that no environmental, dietary or lifestyle factors are known to cause these chromosome changes and neither parent is responsible.

## Management recommendations include:

- Feeding management, if necessary.
- Consider a neurology/cardiology/orthopaedic/ophthalmology/audiology/gastroenterology/palate review.
- Follow up by a developmental paediatrician.
- Physiotherapy/occupational therapy/speech and language therapy/assessment for behavioural concerns, if needed.
- An assessment to identify the most appropriate schooling.
- A high standard of dental care.

## Families say ...

"For us the important thing has been to make sure we don't limit our son's world by assuming that he can't or won't do things, but instead to give him all the opportunities for development we can and accept help with that too."

## Inform Network Support



Understanding Chromosome & Gene Disorders

### Rare Chromosome Disorder Support Group,

The Stables, Station Road West,  
Oxted, Surrey. RH8 9EE. UK.  
Tel +44(0)1883 723356

## Websites, Facebook groups and other links:

Chromosome 8p Disorder Parent Support Community  
Chromosome 8 Disorders  
8p23.1 Deletion/Duplication Syndrome Family Group  
Project 8p: Join  
Project 8p: Patient Navigator

This leaflet was made possible by a contribution from  
The Boshier-Hinton Foundation



## Join Unique for family links, information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at [www.rarechromo.org/donate](http://www.rarechromo.org/donate) Please help us to help you!

This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. This information guide was reviewed by Dr Fe García Santiago, Institute of Medical and Molecular Genetics, Hospital Universitario La Paz, Madrid, Spain and by Dr Wendy Chung & Dr Haluk Kavus, Division of Molecular Genetics, Columbia University, New York, USA,

2021 Version 1 [CA]  
2026 Version 1.1 [NM]

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Registered in England and Wales

Charity Number 1110661  
Company Number 5460413



Understanding Chromosome & Gene Disorders

# 8p inverted duplication & deletion



[rarechromo.org](http://rarechromo.org)

## What is 8p inverted duplication & deletion?

Inverted duplication and deletion of 8p, known as *inv dup del 8p*, is a rare genetic condition that is estimated to occur once in every 10,000-30,000 births. Individuals with *inv dup del 8p* have one unaffected chromosome 8, but the other chromosome 8 has undergone a complex rearrangement.

In this rearrangement, a small bit of the tip of 8p is missing (deleted), and a relatively large bit is duplicated. The extra duplicated part runs in the opposite direction to normal and is therefore termed inverted (*inv*). The size of the duplication is not the same in all people and depends on where the chromosome breaks to form the *inv dup del 8* chromosome.

For some people with *inv dup del 8p*, only the extra duplicated material is reported and not the small deletion. This may be due to an incomplete diagnosis, perhaps by karyotyping alone, where the chromosomes are examined under a microscope; the duplication is large enough to be easily detected but the technique used was not sensitive enough to pick up the tiny deletion. However, it is believed that the great majority of people with *inv dup 8p* also have the small deletion.

Each person with *inv dup del 8p* is unique and will have different

## Features of *inv dup del 8p*

developmental and medical concerns. Even individuals with the same or very similar duplication and deletion can show variation in the range and severity of features, but the most likely features and/or those that are the most likely to make a difference to a child's health or development are:

- Some degree of developmental delay
- Some degree of learning disability
- Speech and language delay or absence of speech
- Low muscle tone (hypotonia)
- Structural anomalies of the brain
- A heart condition, which often resolves naturally
- Feeding difficulties
- A recognisable “look” to the head and face

Features that are not usually obvious at birth but may develop during childhood include:

- Spinal curvature
- Contracted joints, making movement difficult

Other features have been noted in medical literature and among *Unique* members.

Please refer to *Unique's* longer guide for further details.

## Medical concerns

- **New-born babies** Babies are likely to show some sign(s) of difficulty at birth, including breathing difficulties, apnoea events, jaundice and feeding difficulties. A significant number of new-borns are described as “unusually inactive and placid”. Some children have on-going health conditions, including asthma, seizures, spinal curvature and gastrointestinal problems.
- **Seizures.** Seizure disorders, including epilepsy, appear to be common.
- **Head & brain** Anomalies of the brain are common, most often absence or underdevelopment of the corpus callosum, although a wide range of other anomalies have been reported. Approximately half of babies have an unusual head shape or head size.
- **Heart** A range of heart conditions have been reported. If needed, treatment will be decided on an individual basis but can include monitoring to see whether they resolve naturally, as was the case in the majority of cases.
- **Joint anomalies** Problems with loose or unstable joints are particularly prevalent and, as children get older, there appears to be a marked tendency for progressive hypertonia (increased muscle tone) to develop in the legs. If left untreated, this can lead to contracted joints, which can limit normal activity and movement.
- **Skeletal anomalies** Skeletal anomalies such as scoliosis, kyphosis and kyphoscoliosis have been reported.
- **Minor anomalies of the genitals** Minor anomalies of the genitals are sometimes seen in boys with *inv dup del 8p*, but girls are much less likely to be affected.
- **Eyes & vision** Problems with vision and structural eye anomalies have been reported, including: long-, short- or partial-sightedness; astigmatism; nystagmus (uncontrollable, repetitive movement of the eye); and strabismus (a squint).
- **Hearing** Some babies and children have a temporary hearing loss caused by glue ear; others, much less commonly, have a moderate to serious sensorineural hearing loss in one or both ears.
- **Anomalies of the palate** A high/arched palate is a common feature and very rarely babies may have a cleft lip or palate.
- **Eosinophilic Oesophagitis (EO)** Almost half of *Unique* parents said that their child had a diagnosis of EO. Symptoms vary depending on age and there is overlap with those associated with acid reflux.

## Development

- **Feeding & growth** Feeding difficulties in the early months can sometimes lead to a slowing of weight gain relative to length. Often these difficulties are mild and temporary, but sometimes they may be more severe or long-lasting and require treatment. Most children find chewing difficult and avoid hard and lumpy foods. Roughly two thirds of *Unique* children are described by parents as having growth delay (usually mild to moderate). Children are often slightly short and underweight for their height; a few children are tall and above average weight for their age.
- **Gross & fine motor skills** A delay in reaching developmental milestones, such as holding the head up and sitting, is to be expected, although there is a wide range of eventual ability. Difficulties with hand use and hand-eye coordination should also be anticipated. These skills may improve with age, but children, teenagers and adults usually need help to carry out personal care tasks.
- **Learning** Individuals will typically need support with learning and in many cases this may be considerable. While there are children who have only mild learning disability, they seem to be the exception and many have severe to profound disability. There is some evidence to suggest that those with a smaller duplication or mosaicism may be less severely affected.
- **Speech and language** Speech and language are specifically delayed or absent in children, but communication is typically good. For some children first words appear between the ages of two and three years, while for others a more significant delay is observed, and many remain non-verbal. Almost universally, parents believe that their child can understand a lot more than they can express.
- **Personality & behaviours** Families consistently describe a child with a happy, loving, charming, contented and upbeat outlook. Parents talk of empathetic and tactile children who are extremely sociable, sometimes preferring the company of older children or adults. Children particularly enjoy sensory activities involving music, dance, lights and noise. Against this usually sunny disposition, difficulties in communicating needs or completing tasks can lead to frustration, outbursts of temper and “challenging behaviours”. An association between *inv dup del 8p* and autism spectrum disorders (ASDs) and attention deficit hyperactivity disorder (ADHD) has been suggested.