Why did this happen?

At fertilisation, a sperm and egg cell join to form a single cell. This cell must continuously make copies of itself and all its genetic material (replicate) to produce the billions of cells that are necessary for human growth and development. Sometimes during the formation of the egg or sperm cells, or during this complicated replication process, changes to the structure of chromosomes may occur e.g. parts of a chromosome are deleted (lost), duplicated (gained) and/or become rearranged. The effect of any chromosomal change varies according to how much genetic material is involved and, more specifically, which genes and/or regions that control genes are included, as well as numerous other factors that we are only just beginning to understand.

What is certain is that, as a father or mother, there is nothing you did to cause the duplication and deletion and nothing you could have done that would have prevented it. Chromosome rearrangements affect children from all parts of the world and all types of background. No environmental, dietary or lifestyle factors are known to cause co-existing 9p duplication and deletion.

Can it happen again?

To answer this question, the parents' and the affected child's chromosomes need to be tested as the possibility of having another pregnancy with a 9p duplication and deletion depends on the parents' chromosomes. The evidence from medical literature and *Unique* members suggest that most cases are *de novo* (dn), meaning that this has happened in the child as a new event. In this case, where both parents have "normal" chromosomes when tested, they are very unlikely to have another affected child. Very rarely (less than 1%), both parents have normal chromosomes by a blood test, but a few of their egg or sperm cells carry a chromosomal change. This is called germline (gonadal) mosaicism and it means that parents whose chromosomes appear "normal" when their blood is tested can have more than one child with the duplication.

Where the test reveals a rearrangement in the parents' chromosomes, the chances of having another affected child are very much higher. However, each family's situation is individual, and all families should be able to discuss the possibilities they face with their geneticist or genetic counsellor.

Families say ...

"At first it was overwhelming because the doctors didn't have answers for us. At times it might feel like you're the only one who has a special child and that no one could ever understand you and your situation, but there are many others with the same issues. You are not alone."

Management options might include:

- Feeding management if necessary e.g. attending a feeding clinic to help with eating, drinking and swallowing difficulties.
- Follow up by a developmental paediatrician.
- Physiotherapy/occupational therapy/speech and language therapy/assessment for behavioural concerns, as needed.
- Eye examination e.g. check if glasses are needed.
- An assessment to identify the most appropriate schooling.
- A high standard of dental care to minimise damage by decay and erosion. Children and adults may also benefit from specialist hospital dental services and may require treatment under general anaesthetic.
- Clinical genetics referral e.g. to help interpret genetic test results, advice about future pregnancy etc..

Inform Network Support

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Trisomy 9p Families - https://m.facebook.com/groups/664602633553853
Trisomy 9 Family - https://m.facebook.com/groups/365958293491370
Chromosome 9 Disorder - https://m.facebook.com/groups/132806160119488
Chromosome 9 - https://m.facebook.com/groups/120832324611655
http://www.9pminus.org/

This 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. Unique does its best to keep abreast of changing information and to review its published guides as needed. This leaflet was compiled by Unique (CA) and Nina Fowler (trainee genomic counsellor (STP), St George's University Hospital, London, UK) in 2020 and reviewed by Prof V.H.W. Dissanayake, Human Genetics Unit, Faculty of Medicine, University of Colombo, Sri Lanka.
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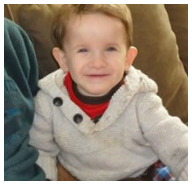
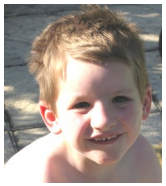
Unique
Understanding Chromosome & Gene Disorders

Co-existing 9p duplication & deletion





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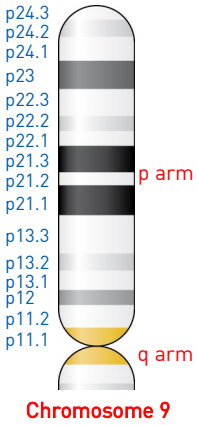
What is co-existing 9p duplication and deletion?

A 9p duplication occurs when there is extra chromosomal material from the short arm of chromosome 9 (9p) in the cells of the body, while a 9p deletion occurs when a piece of chromosome 9p is missing.



Chromosome 9

Sometimes an individual may have co-existing duplication and deletion of chromosome 9p, meaning there is both duplicated and deleted material. The size of both the duplication and deletion can vary. As with other chromosomal disorders, having an extra piece and a missing piece of chromosome 9 may affect the development and intellectual abilities of a child, although there is considerable variability in these and other individual features that are observed.

When a particular set of developmental features occurs in a recognisable and consistent pattern as a result of a single cause, the condition is called a syndrome. The features of both 9p duplication and 9p deletion occur in this way, so these disorders are sometimes known as dup(lication) 9p syndrome (or trisomy 9p or trisomy 9p syndrome), and del(etion) 9p syndrome (or 9p minus or monosomy 9p syndrome), respectively.

People with concurrent duplication and deletion of chromosome 9p are likely to have features associated with both duplication 9p and deletion 9p syndromes.

Most common features:

- A "recognisable" look to the head and face
- Some degree of developmental delay
- Some degree of learning disabilities
- Speech and language delay
- Growth delay (often mild)
- Low muscle tone (hypotonia)
- Feeding difficulties, including reflux
- Constipation
- Anomalies of the hands and feet
- Dental concerns
- Minor anomalies of the genitals in boys

Medical concerns

Respiratory infections Respiratory infections and/or asthma are common in children but often become less frequent with age and maturity, although they can persist throughout childhood.

Seizures Some individuals experience seizures, which are usually successfully controlled with medication.

Brain anomalies A range of anomalies of the brain have been reported, many of which were minor.

Heart A range of heart conditions have been reported, many of which were minor and resolved naturally without any need for treatment or surgery.

Eyes & vision Problems with eyes and vision have been reported including: long-, short- or partial-sightedness; nystagmus (uncontrollable, repetitive movement of the eye); and strabismus (a squint). Interventions like patching, exercises or glasses may be recommended.

Hearing Some degree of hearing loss, due to a combination of glue ear, unusually narrow external ear canals and excess wax in the ear canal, was relatively common. The evidence suggests that problems with hearing loss are usually temporary, generally affecting only children in the early years.

Anomalies of the palate Anomalies of the palate (roof of the mouth) frequently affected both *Unique* children and patients in the medical literature.

Constipation Constipation is particularly common and can cause considerable discomfort. It is important that parents discuss the possible causes with their health visitor or doctor.

Skeletal & joint anomalies Cases of scoliosis (a sideways S-shaped curve of the spine) and kyphosis (an outward curve resulting in a hump) have been reported, often in teenagers or adults. Extremely loose (hypermobile) joints are relatively common.

Anomalies of the genitals & ambiguous genitalia Typical anomalies affecting many boys were: undescended testis/testes (cryptorchidism), a very small penis (micropenis), or most frequently, a combination of both. When descent does not occur, the testes can be brought down in a surgical operation (orchidopexy) and anchored in the scrotum. Some boys were born with a small penis or buried (hidden) penis, but the penis often had a "normal" appearance after puberty.

Other conditions Occasionally, other medical conditions were reported such as hypersalivation and drooling; narrow or blocked nasal passages; delayed bone maturation; kidney anomalies; and tracheomalacia/laryngomalacia.

Development

Growth

While a slight delay in growth before birth is occasionally observed, a pattern of slow growth in babies and children was more common. In *Unique* families, delays were generally deemed to be "mild" or "moderate". Children and teenagers in the *Unique* series were of below average or average height and often of average weight.

Gross and fine motor skills

A delay in sitting, moving and walking appears to be universal. The majority of children are also likely to experience a delay in the development of hand use and hand-eye coordination. For many these skills remain poor into the teenage years. Most co-existing 9p duplication and deletions, no matter the size and location, are associated with reduced muscle tone (hypotonia), sometimes with increased muscle tone (hypertonia). Hypotonia can make the body floppy and may lead to an obvious head lag in babies, while hypertonia makes the body feel overly rigid. Both of these features can result in difficulties in sitting, moving and walking.

Learning

Evidence from *Unique* and the medical literature indicates that some degree of learning disabilities (LD) is to be expected and is usually moderate to severe. Several children have shown a marked improvement after struggling as toddlers and in early childhood, especially following intervention to improve communication skills.

Behaviour

Alongside many positive personality traits - with parents telling us about sweet, loving, happy and cheerful children - so-called "challenging" behaviours, often related to difficulties with communicating, were common. These ranged from temper tantrums and more aggressive or destructive behaviour to extreme shyness and an inability to adapt to new situations. Other parents say that they have experienced no problems with behaviour. A few children had received a specific diagnosis for a social, emotional or anxiety disorder.

Speech and language

There is typically some delay in speech and language development. For some, first words appeared between the ages of two and three years, while others had more significant delays and a few remained non-verbal. Most parents believed that their child could understand a lot more than they could express and even those children who developed more sophisticated speech still sometimes experienced difficulties expressing themselves.