Why did this happen?

To answer this question, the parents' and affected child's chromosomes need to be tested. These tests may indicate that the duplication has been inherited or, where the parents' chromosomes are normal, the cause of the duplication is then likely to have occurred when the sperm or egg cells were being made (this is called '*de novo*' (dn), meaning the duplication occurred as a new event in the child).

The medical literature suggests that the majority of 9p duplications arise because one parent, either the mother (mat) or father (pat) has a rearrangement of their own chromosomes. In most cases this rearrangement is a balanced translocation between chromosome 9 and another chromosome, and the parent would not be expected to show any symptoms.

Can it happen again?

The chances of having another child with a 9p duplication depend on the results of chromosome tests on the parents. Where the tests show that the parents' chromosomes are normal, their chances of having another affected child are usually no higher than for anyone else in the population (although very rare, it can still happen due to a phenomenon known as germline mosaicism). Where the test reveals a rearrangement in the parents' chromosomes, the chances are very much higher. Each family's situation is individual, and all families should be able to discuss the possibilities they face with their geneticist or genetic counsellor.

Management recommendations

- Feeding management if necessary e.g. attending a feeding clinic to help with eating, drinking and swallowing difficulties.
- Follow up by a developmental pediatrician.
- Physiotherapy/occupational therapy/speech and language therapy/assessment for behavioural concerns if needed.
- Eye examination e.g. check if patching, exercises and/or glasses are needed.
- An assessment to identify the most appropriate schooling.
- A high standard of dental care. Children and adults may also benefit from specialist hospital dental services and may require treatment under general anaesthetic.

Families say ...

"I've learned she doesn't need fixing, this is who she is. I've learned patience, acceptance, and happiness from her. I don't know if our lives would be as rich without her. One day at a time. Never give up. Encourage, love, and accept as they are."

Inform Network Support

Rare Chromosome Disorder Support Group,
The Stables, Station Road West,
Oxted, Surrey. RH8 9EE. UK.
Tel +44(0)1883 723356
info@rarechromo.org

Websites, Facebook groups and other links:

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

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 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. *Unique* does its best to keep abreast of changing information and to review its published guides as needed. This information guide was compiled by Amy Clarkson, The Newcastle Upon Tyne Hospitals NHS foundation trust. NHS using information from the comprehensive *Unique* information guide for 9p duplication syndrome (2018).

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Rare Chromosome Disorder Support Group
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Understanding Chromosome & Gene Disorders

Duplication 9p syndrome

rarechromo.org

What is a duplication of 9p?

A 9p duplication is a rare chromosome disorder (RCD) in which there is extra chromosome material from the short arm of chromosome 9 (9p) in the cells of the body. The size of the duplication can vary, and the extra material may consist of the entire short arm, part of the short arm or include some of the long arm (9q).

As with other chromosome disorders, having an extra piece of chromosome 9 may affect the development and intellectual abilities of a child, although there is considerable variability in the 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 a 9p duplication do occur in this way, so the disorder is sometimes known as dup(lication) 9p syndrome. It is also sometimes called trisomy 9p or trisomy 9p syndrome.

Most people have:

- A recognisable "look" to the head and face
- Some degree of developmental delay
- Some degree of learning disabilities, ranging from mild to profound
- Speech and language delay
- Growth delay, which is usually mild
- Abnormalities of the hands and feet, often mild
- Dental concerns
- Low muscle tone (hypotonia)
- Minor anomalies of the genitals or undescended testicles are common in boys
- Constipation
- Feeding difculties, including reflux

There are also other possible features that are described in *Unique*'s longer information guide for 9p duplication syndrome. Features may depend on exactly which piece of chromosome 9p has been duplicated as well as each persons own unique genetic makeup.

How common is a duplication of 9p?

Compared with other rare chromosome disorders, 9p duplications are thought to be more common. Over 150 people with partial or complete duplication of 9p have been reported in the medical literature and *Unique* currently has over 200 members with a 9p duplication who live worldwide.

Medical concerns

- **General well being.** Although generally in good health, some health conditions, including respiratory infections, constipation, and eczema have been reported by some families.
- **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.
- **Hands and Feet.** Almost all children described with a 9p duplication had at least one unusual feature of their hands and/or fingers. A wide variety of specific abnormalities of toe and foot position are also noted.
- **Siezures.** Seizures, including epilepsy, affect some babies and children with 9p duplications.
- **Heart.** A range of heart conditions have been reported, many of which were minor and resolved naturally without any need for treatment or surgery.
- **Brain anomalies.** A range of anomalies of the brain have been reported, many of which were minor.
- **Kidneys.** Several *Unique* families reported minor anomalies of the kidneys, many of which were minor.
- **Minor anomalies of the genitals.** Typical anomalies affecting many boys were: undescended testis/testes (cryptorchidism), a very small penis (micropenis), or a combination of both.
- **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.
- **Eyes and 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).
- **Hearing.** Some degree of hearing loss, due to a combination of glue ear, unusually narrow external ear canals and excess wax is relatively common.
- **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 palate** Anomalies of the palate (roof of the mouth) frequently affected both *Unique* children and patients in the medical literature.

Development

- **Growth.** While a slight delay in growth before birth is sometimes observed, a pattern of slow growth in babies and children is more common but generally deemed to be "mild". Children and adults in the *Unique* series are usually described as average or below average height and weight.
- **Gross and fine motor skills.** Babies and children are typically delayed in reaching their developmental "milestones", including rolling, sitting, moving and walking. Development of hand use and hand-eye coordination are frequently delayed and don't necessarily develop in line with gross motor skills.
- **Learning.** Evidence from *Unique* and the medical literature demonstrates that most 9p duplications are associated with some degree of learning difficulty ranging from mild to profound, although a few children have no learning difficulties.
- **Behaviour.** Children with the same inherited 9p duplication can have strikingly different personalities, but when describing their children, the words most frequently used by *Unique* parents are: loving, happy, fun, caring, gentle, determined, stubborn and sociable. Alongside these positive traits, some "challenging" behaviours, often related to difficulties with communicating, are common. A few children have been diagnosed with autism spectrum disorder (ASD) or have autistic like features. Others have been given the diagnosis of attention deficit disorder (ADHD), obsessive compulsive disorder (OCD) or sensory processing disorder (SPD).
- **Speech and language.** Speech is usually the most obviously affected area of development. For verbal children, first words often emerge between the ages of one and five years, and often between a child's third and fourth birthday. A significant minority of children remain non-verbal. Understanding appears to progress ahead of speech in all children, and clear, literal instructions and statements reinforced by gesture were universally understood.

This information guide was compiled using information from the comprehensive *Unique* information guide for 9p duplication syndrome (2018). Please refer to the longer guide for further details. *Unique* also publishes a guide for balanced translocations.