UNIQUE
UNDERSTANDING GENES & CHROMOSOMES

QRICH1-related syndrome

(Ververi-Brady syndrome)

rarechromo.org

This guide is designed to help families and healthcare professionals looking after people with QRICH1-related syndrome. It contains information about the cause, the ways in which it can affect people and suggestions about the help and management that can benefit people with this condition.

What is QRICH1-related syndrome?

QRICH1-related syndrome, also referred to as Ververi-Brady syndrome, is a rare neurodevelopmental disorder associated with developmental delay, varying degrees of learning (intellectual) disability and behavioural differences. The condition affects multiple systems in the body, and key features can include low muscle tone (hypotonia), seizures, short stature and distinctive facial features.

As is common with genetic conditions, each person can be affected differently - even among affected members within the same family. Not everyone with QRICH1-related syndrome will have all the possible features and each person with a certain feature won't necessarily be affected by it to the same level as other people with that feature.

QRICH1-related syndrome is caused by a change (variant) in the QRICH1 gene.

How common is QRICH1-related syndrome?

QRICH1-related syndrome is extremely rare. To date (2026) just over forty people have been reported in the medical literature, but more will have been diagnosed. It is expected that more people will be diagnosed with this condition as awareness increases and genetic testing becomes more routine.

What features and symptoms do people with QRICH1-related syndrome have?

As is common with many genetic conditions, children and adults with QRICH1-related syndrome can have a range of features and symptoms. As more people are diagnosed, and information is shared, the range of features, and the likelihood of a child or adult having these features, will become clearer.

Common features

Most people with QRICH1-related syndrome have:

- Intellectual disability or global developmental delay, with severity often reported as mild to moderate
- Distinctive facial features, although there is no single unifying facial appearance
- Language delay
- Motor delay

Other possible features include:

The features below are listed according to how many people have been reported in the medical literature to have that specific feature. The list is in decreasing order, it starts with about half of reported individuals and decreases down to about 10% of reported individuals. Since so few people have been reported in the medical literature, the exact values are likely to change as knowledge increases.

- Autism spectrum disorder (ASD)
- Low muscle tone (hypotonia)
- Structural brain anomalies
- Feeding difficulties or gastroesophageal reflux (GORD)
- Short stature
- Low weight
- Seizures (epilepsy)
- Ear anomalies
- A high arched palate
- Curvature of the spine (scoliosis or hyperlordosis)
- Delayed or accelerated bone age
- Structural heart anomalies
- Kidney (renal) anomalies

Elevated creatine kinase levels have also been reported in a few individuals, but the frequency is not well-established. Other findings reported in a small number of people include a common, usually benign, fluid-filled sac in the brain's pineal gland (pineal cyst), hearing loss, birthmarks (congenital melanocytic nevi, café-au-lait macules), atopic dermatitis, tongue-tie (ankyloglossia) and teeth prone to decay. Atypical cartilage and bone growth (chondrodysplasia) has been reported in a couple of people (two out of eleven) and a few people have a bent finger (campodactyly) or a curved finger (clinodactyly).

Pregnancy and birth

Most pregnancies are unremarkable and proceed without complication but where there have been concerns during pregnancy this has mainly been intrauterine growth retardation (IUGR).

The majority of pregnancies go to term but some babies are born prematurely. In a study of 38 individuals, 7 babies (18%) were born prematurely (Kumble 2022). In most of these families, contributing factors included the mother carrying more than one child (multiple pregnancy/gestation), or having a health condition (maternal comorbidities) or placental problems. Some babies show some signs of difficulty at birth, related to difficulties with feeding, jaundice or a heart condition. Some newborns also have low muscle tone (muscular hypotonia). Birth weights are usually below average (in a study of 38 individuals, eleven (29%) had a low birth weight). Reported birth weights have ranged from about 2.4 kg (5 lbs 5 oz) to 3.8 kg (8 lbs 7 oz).

Photo above:

“*Day of birth,* we were all clear from the cystic hygroma* seen prior to 12 weeks in utero, but weren’t sure how he would turn out; small, 6.3 lbs with some extra skin on the back of his neck where the hygroma was and had disappeared, also low muscle tone was noted.”

*(A hygroma is a sac-like structure from the lymphatic system that forms as a bulge under the skin. This is not a known feature of QRICH1-related syndrome).

Facial Appearance

Certain facial features are found more often in children with QRICH1-related syndrome than in other children. However, it is important to note that these features are variable and a single, unifying facial appearance has not been identified. These features may mean that you see unexpected similarities between your child and others with QRICH1-related syndrome. The most common characteristic features include eyes that are wide-set (hypertelorism) and deep-set, with wide openings (palpebral fissures) and puffiness (periorbital fullness). Ears may be low-set, cup-shaped, or otherwise unusually shaped (dysplastic). Noses may be prominent with a broad bridge and a bulbous tip. A short, long or smooth space between the nose and upper lip (philtrum) may also be apparent as well as a wide mouth with thin lips and a high palate.

Development

Gross and fine motor skills

Developmental delay has been reported in most children with QRICH1-related syndrome reported so far (2026). In one study of 31 individuals, 21 had motor delay (Kumble 2022). The severity of motor impairment is highly variable, ranging from standard early motor development in some individuals to profound motor delay in others. For example, one child achieved independent sitting at 7 months and walked at 15 months, while another sat at 13 months and began walking at 21 months. For most affected individuals, gross motor milestones, are mildly delayed.

Photo opposite:

"Biking without training wheels for the first time!" - 9 years

Low muscle tone (hypotonia), which can be present from birth, is common and may affect mobility. Other features that may be present include decreased reflexes and an involuntary shaking of a limb when intending to move it (intention tremor). Some children may have an unusual or unsteady gait when walking because of balance issues (known as ataxia), clumsiness or a tendency to fall.

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Intellectual development and learning

Most children with QRICH1-related syndrome have learning (intellectual) disability (ID) or learning difficulties. In a study of 38 individuals, 27 people (71%) had either ID or global developmental delay (GDD) (Kumble 2022). ID ranges from mild to severe but is usually in the mild to moderate range. The likelihood and severity of ID may be influenced by the type of genetic change. Other specific challenges can include writing difficulties, dyslexia and difficulties with planning and organisation.

Most children will require significant additional support with their learning throughout their education; even children with an IQ within the typical range may have a history of developmental delay or need extra help at school. Early intervention can prove particularly beneficial and formal testing to assess specific, individual needs is recommended to help each child reach their full potential.

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Speech and language

Children with QRICH1-related syndrome typically experience some degree of speech and language delay, which is often the first concern raised by parents. A few may find it difficult to co-ordinate movement of their lips, jaw and tongue to make the right sounds (apraxia of speech). Other speech production challenges can include slurred speech and articulation difficulties. The eventual range of achievement is broad, but a few people may remain non-verbal, this has been reported in two out of 38 individuals (Kumble 2022). In a few cases, a loss of previously acquired language skills (language regression) has been reported. Those who do develop speech may achieve single words, short phrases or basic sentences, often using a simple vocabulary

with grammatical errors. Difficulties with social communication have also been observed. A common observation is that a child's understanding of language (receptive language) is significantly better than their ability to speak (expressive language).

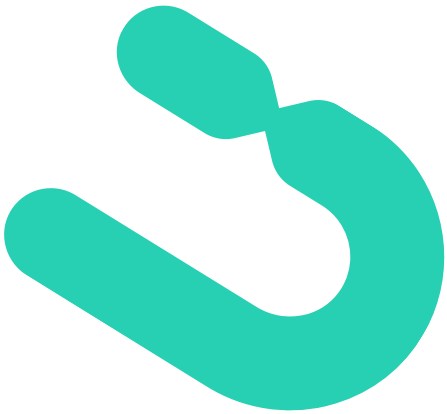
An assessment by a speech therapist should be able to identify your child's specific difficulties, allowing regular therapy sessions tailored to your child's specific areas of need.

Unique publishes a separate guide to Communication

Feeding

Feeding issues in the new-born period are common. These issues are often associated with poor weight gain (sometimes described as failure to thrive). Eleven individuals in one study were reported to have weights significantly below the expected range (Kumble 2022). Many babies also suffer from gastro-oesophageal reflux (GERD/GORD), which may require treatment. Two children reported so far have required the insertion of a PEG/G-tube (percutaneous endoscopic gastrostomy tube) (Kumble 2022), where a tube is passed directly into the stomach through the abdomen and used to administer food. The specific causes for these feeding difficulties are not yet well understood.

Unique publishes a separate guide to Feeding



Growth and stature

Most children with QRICH1-related syndrome described in the medical literature so far (2026) are noted as having growth delay and short stature, with consistently low weight and height for age described as a key feature (affecting 28 of 39 individuals (72%) in one study (Wang 2023)). Growth differences can begin before birth, with some babies having restricted growth in the womb (intrauterine growth restriction). Beyond infancy, height and weight is variable and often remains below average and poor weight gain is common.

Behaviour

Children with QRICH1-related syndrome typically tend to have behaviour in keeping with their overall degree of developmental delay. Some have a happy disposition and have friendly but shy personalities or are cheerful and communicative. Difficulties with social communication and interaction are common and may include a low tolerance for frustration, distress at minor changes and a tendency towards social withdrawal. Some children have an autism diagnosis or traits (reported in 11 of 38 individuals (29%) (Kumble 2022)). Other behaviours include attention deficit hyperactivity disorder (ADHD) (reported in 16 of 38 individuals (42%)) and anxiety (reported in 16 of 38 individuals (42%)). Aggressive behaviours (including biting) and sensory processing differences (such as tactile sensitivity) have also been reported. One person with QRICH1-related syndrome is also known to have Tourette syndrome.

Understanding each child's complex profile is key to providing effective support. Management strategies should focus on addressing the root causes of challenging behaviours - such as providing sensory supports, using strategies to manage anxiety, and creating structured environments to help with attention - while nurturing the child's inherently friendly and sociable personality. Efforts to take into account and introduce strategies to tackle communication and other difficulties can also be beneficial.

Unique publishes a separate guide to Challenging Behaviour

Puberty

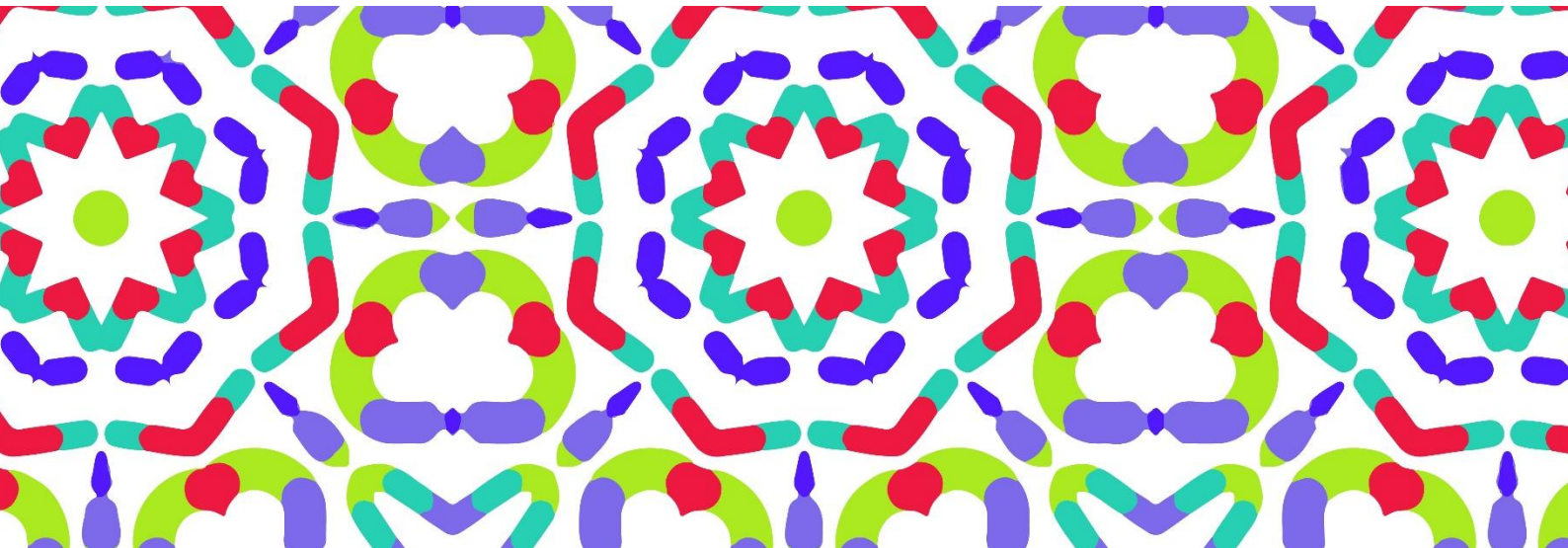
There is limited information available about puberty in children with QRICH1-related syndrome. There is one report of delayed puberty. Some families of children with genetic disorders and behavioural difficulties or learning disability can be particularly concerned at their daughter's ability to cope with menstruation, and for some discussing menstrual regulation options with a paediatrician may be beneficial.

Unique publishes a separate guide to Puberty

Adulthood

There is not much information available about adults with QRICH1-related syndrome. Experiences of adulthood are likely to vary considerably and will depend on many factors. These include the level of any LD/ID, possible on-going medical concerns and improvements in early intervention and therapies/treatments. Based on the limited information available, learning difficulties are a consistent feature into adulthood. While intellectual disability is an associated feature, thinking and learning skills (cognitive function) can be variable. Behavioural and mental health conditions have also been described in adults, alongside social challenges such as difficulties with social interaction.

Several adults with QRICH1-related syndrome only discovered that they had a QRICH1 gene variant when their child was diagnosed. These adults are known to have mild symptoms.



Medical concerns

The following medical concerns have been found in children with QRICH1-related syndrome. They are not found in all children so not all children with QRICH1-related syndrome will be affected.

Heart

Structural heart anomalies have been noted in five individuals with QRICH1-related syndrome (Kumble 2022). The types of heart conditions seen have ranged from simple to complex. Reported congenital heart conditions (when a baby is born with the condition) include an atrial septal defect (a hole between the top two chambers of the heart). In one individual, a more complex condition where the main arteries leaving the heart are in the wrong positions (called transposition of the great arteries) was seen alongside an atrial septal defect. Information about the long-term outlook for these heart conditions is limited. In one child, an atrial septal defect resolved by the age of five years.

Brain

Some children have a structural brain anomaly. One report noted 10 of 19 individuals (52%), who had had brain imaging, had a structural brain anomaly (Kumble 2022). Anomalies can be detected by MRI (magnetic resonance imaging) or a CT (computerised tomography) scan of the brain. The findings are typically described as minor and not specific to this condition.

The most frequently reported anomalies include:

- Non-specific changes to the white matter of the brain (periventricular white matter anomalies)
- Cysts filled with cerebrospinal fluid that are located between the brain or spinal cord and the arachnoid membrane (arachnoid cysts)
- Cysts on the pineal gland in the brain (pineal cysts)

Seizures

Some children with QRICH1-related syndrome experience some form of seizure (a sudden and unexpected change in the electrical activity in the brain). One report noted 9 out of 38 individuals (24%) experienced seizures (Kumble 2022), the age of onset varied from 6 months to 14 years. Seizure types reported in individuals with QRICH1-related syndrome include:

- Absence seizures: A change in behaviour as if the child ‘switches off’, sometimes with staring, eyelid flickering or lip smacking. Absences are very brief, often lasting less than half a minute
- Infantile spasm: Type of seizure usually occurring in clusters in babies between 3-10 months. Seen most often when a baby wakes and may be obvious or subtle
- Myoclonic-atonic: Seizure involving jerky or shock-like contraction of muscles, followed by a loss of tone so someone standing up falls to the ground (also known as drop attacks)

Photo opposite:

“With EEG leads on head – 6 months old – hospitalized for infantile spasms.”

Photo opposite:

"Being treated with high dose steroids for seizures; lots of weight gain and loss of milestones such as sitting up, rolling over etc." 7 months old

Hormones

Children with QRICH1-related syndrome may have conditions that affect their hormones. These can include growth retardation and short stature, reported in 12 of 38 individuals (32%) in one report (Kumble 2022). The onset of growth issues can be early, and may be linked to feeding difficulties which can lead to poor weight gain. Bone age can also be affected, with reports of both delayed and advanced bone age, while some individuals have a bone age that is consistent with their chronological age.

"Our son was under the endocrine department from when he was 3 and diagnosed as borderline growth hormone deficient (after IUGR and consistently low height and weight). They kept him under review until he was 14 when he had 2 courses of testosterone injections to bring on puberty, which I'm pleased to say has brought on a growth spurt."

Ears and Hearing

A few children have a hearing impairment, but hearing is unaffected in most children. Hearing loss may be conductive, where sound is unable to travel effectively to the inner ear.

While hearing impairment is uncommon, differences in the shape and structure of the ear are frequently observed, affecting around half of individuals (19 of 38 people in one study) (Kumble 2022). The outer ears may be large, prominent, cup-shaped, fleshy, low-set, asymmetrical or rotated towards the back of the head (posteriorly rotated). Inner ear anomalies have also been noted in two individuals, however, these people had large chromosomal deletions involving other genes, so a direct link to QRICH1-related syndrome is uncertain.

Unique publishes a separate guide to Hearing

Eyes and sight

Eye and vision anomalies are common in children with QRICH1-related syndrome, although findings are varied and some individuals may have standard vision. A thorough evaluation by a paediatric ophthalmologist is recommended at diagnosis.

Although information is limited, the following features have been reported in some individuals:

- Drooping of the upper eyelids (bilateral ptosis)
- Widely-spaced eyes (hypertelorism)
- Deep-set eyes
- Unusually wide or upward-slanting eyelid openings (palpebral fissures)
- Puffiness around the eyes (periorbital fullness)

Hands and feet

Children with QRICH1-related syndrome occasionally have anomalies of the hands and feet. Most common among these are:

- Small hands and feet
- Tapering fingers
- Weak grip strength
- A ‘sandal gap’ between the big toe and second toe

Spine

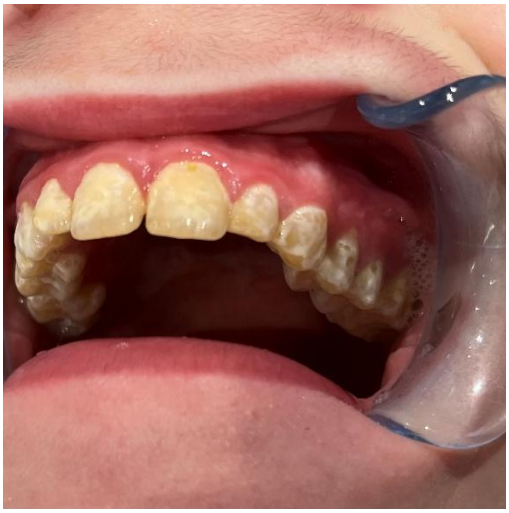
Some babies are born with or develop a spinal curvature, including a sideways curve of the spine (scoliosis) or an increased inward curve of the lower back (hyperlordosis). In a group of 38 individuals, scoliosis was documented in seven (18%) (Kumble 2022). In a separate study of nine individuals, five were found to have either scoliosis or hyperlordosis (Föhrenbach 2021).

Photo opposite:

Post-surgery for traditional growth rods, in a TLSO brace again, finally getting taller with growth hormone.”

Skin

Skin characteristics that have been reported in individuals with QRICH1-related syndrome include moles that are present at birth (congenital nevi) and flat patches of light brown skin (café-au-lait macules spots).



Teeth

Information regarding the dental and oral health characteristics of people with QRICH1-related syndrome is currently limited (2026). A few anomalies have been described including conical teeth (noted in one individual) and anomalies affecting the jaw.

Our son is under the care of a Paediatric dental specialist because he has Amelogenesis Imperfecta, which is a condition where the tooth enamel does not form or mineralise as expected. It is complicated to manage in view of his behavioural issues, but so far we employ various techniques for preventing decay.”

Palate

Anomalies of the palate in particular a high-arched palate is the most commonly reported type of palate anomaly, present in 7 (18%) of a group of 38 individuals (Kumble 2022).

Kidney and urinary tract

A few babies are born with minor anomalies of the kidneys and/or urinary tract, this has been documented in four of 38 (11%) individuals (Kumble 2022). The specific types of structural kidney anomalies have not yet been thoroughly researched.

Photo opposite:

"Post-surgery for unrelated tumor removal in hospital." – 2.5 years

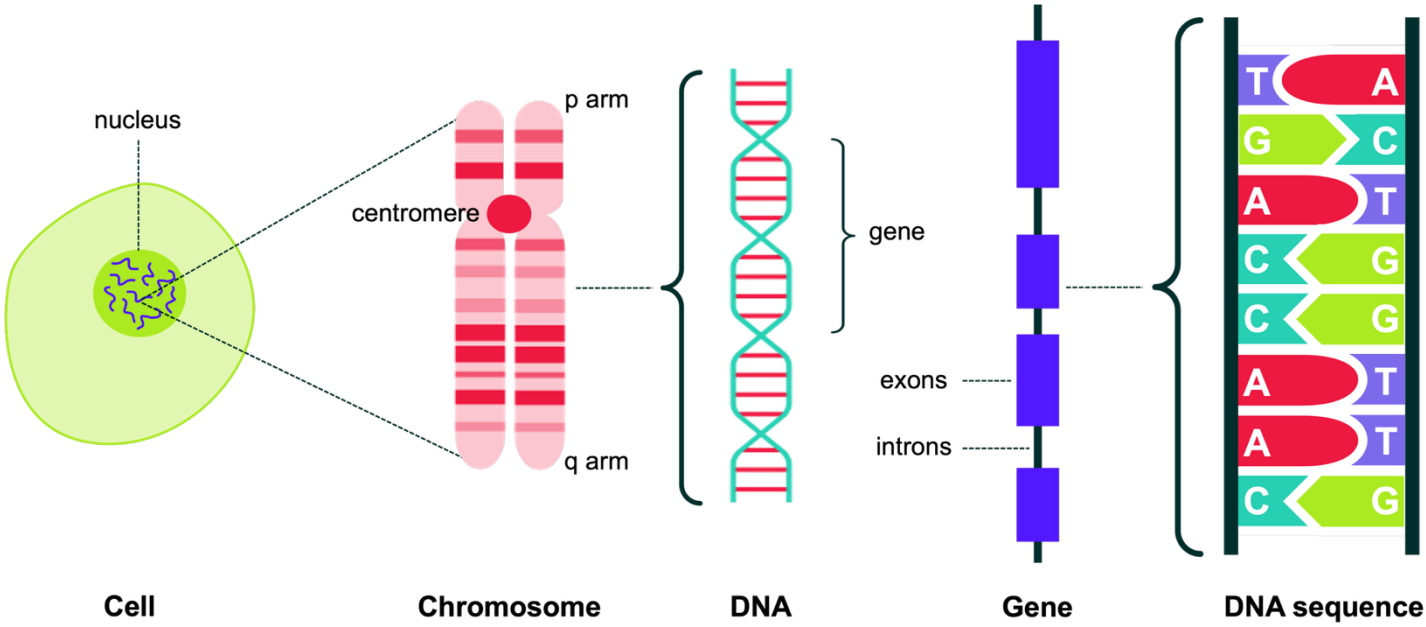
unique

This section includes some more complicated scientific terms and concepts - don't worry if it's a lot to take in, you can always come back to this section later if you want to.

Genes and chromosomes

Genes are instructions which have important roles in our growth and development. They are made of DNA and are incorporated into organised structures called chromosomes. Chromosomes therefore contain our genetic information. Chromosomes are located inside our cells, the building blocks of our bodies. In people with genetic conditions, one or more of their genes don't instruct the body as expected, which can lead to changes in how their body works.

DNA is made up of building blocks called 'bases' or 'nucleotides'. There are four DNA bases which can be abbreviated to the letters A, C, G, and T. These DNA bases are paired up in the DNA structure into 'base-pairs'. The full sequence of our DNA is over three billion base-pairs long. There are changes in the DNA sequence (variants) present in everyone's genes. It is variants in our genes that make each one of us unique individuals.



What causes QRICH1-related syndrome?

QRICH1-related syndrome is caused by specific changes (known as likely pathogenic or pathogenic variants) to the DNA sequence of a gene called QRICH1 (QRICH1 is an abbreviation of the gene's full name, GlutamineRich 1). The QRICH1 gene is located in the short 'p' arm of chromosome 3 in a region called 3p21.31 as shown in the image below.

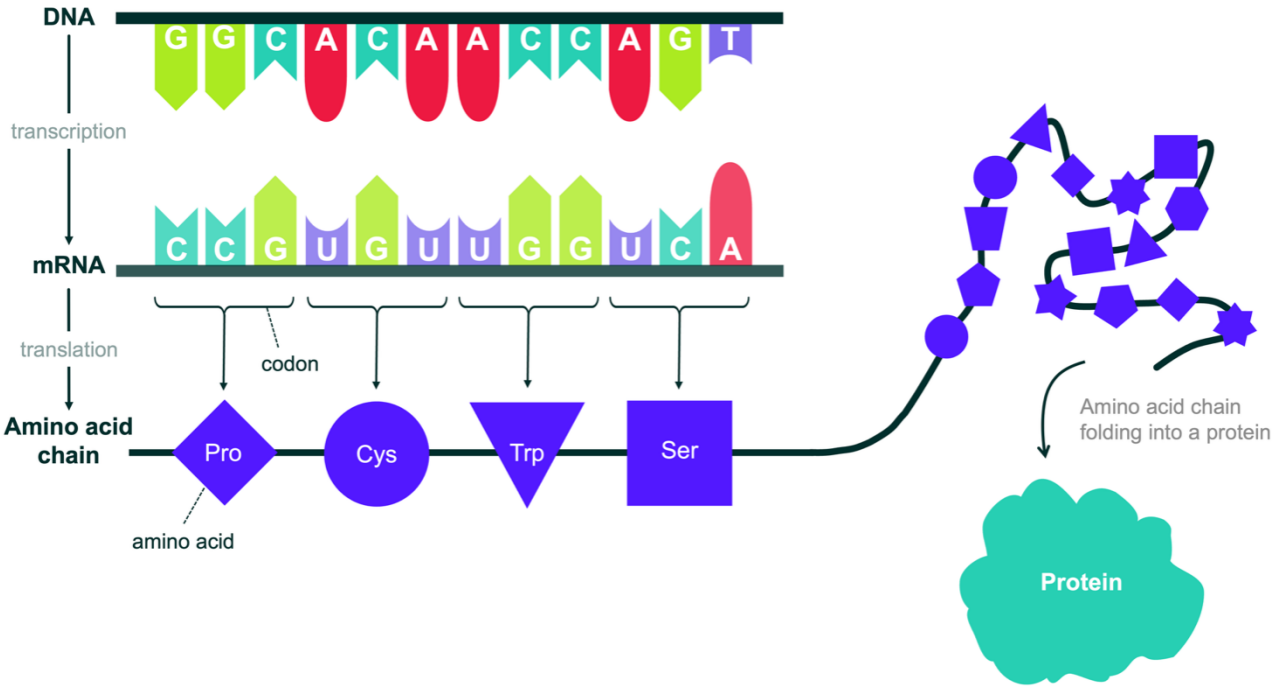
Chromosome 3

We have two copies of chromosome 3 in our cells, so we also have two copies of the QRICH1 gene. QRICH1-related syndrome occurs when only one copy of the QRICH1 gene is affected; the second copy is fully functional. This is known as autosomal dominant since all numbered chromosomes are called autosomes and genetic conditions that occur when only one copy of an autosomal gene is affected are known as dominant.

Unique publishes a separate guide to single gene disorders – autosomal dominant inheritance

Genes and chromosomes

Most genetic conditions are caused by changes to genes that provide instructions for making proteins; these are called protein-coding genes. QRICH1 is one of thousands of these protein coding genes. Proteins are molecules that are made up of long chains of chemicals called amino acids and play critical roles in the body. This is because proteins are essential for lots of processes such as breaking down food, moving our muscles, and growing the body's organs and making sure they function properly. To make these amino acids a copy of the DNA sequence is created, called messenger RNA (mRNA). mRNA uses the same A, C and G bases as DNA, but the 'T' bases are swapped to 'U' bases. Within a gene, each three-letter sequence (codon) of mRNA bases provides the instructions for one amino acid. Changes to the amino acid sequence can affect the function of the protein.



The role of the QRICH1 protein

The QRICH1 gene sequence is used to make the QRICH1 protein. This protein is important because it has a number of different functions. These include regulating the expression of other genes essential for brain development, and other parts of the body.

Different types of genetic changes

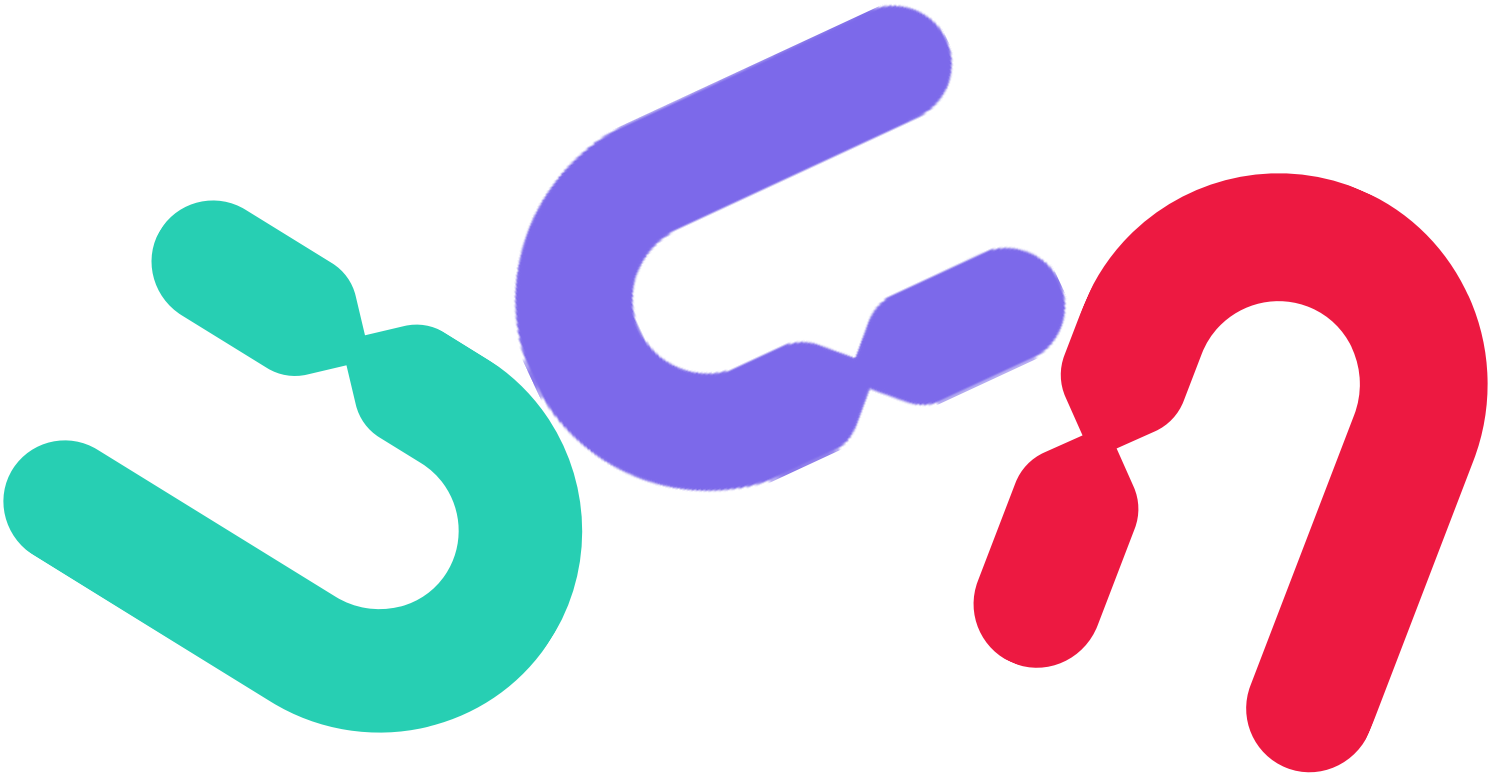
The specific type of genetic change in the QRICH1 gene is an important factor that could influence an individual's symptoms and features. Research suggests that different types of variants can affect the QRICH1 protein in different ways, which might account for the variability in symptoms and features, or their severity, in different people with QRICH1-related syndrome.

Gene sequence variants, loss of function (LOF): People with QRICH1-related syndrome are more commonly found to have a gene sequence variant, as opposed to a deletion of the entire gene. Such variants include:

- Nonsense variants: Where the amino acid change results in a premature ‘stop’ signal, stopping protein production too early, leading to a shorter, unfinished (truncated) protein. In one study of 38 people, 13 had this type of variant (Kumble 2022)
- Frameshift variants: Where the DNA coding sequence is shifted by the addition or deletion of individual DNA bases which means the incorrect amino acid sequence is used to produce the protein or a premature ‘stop’ signal is introduced at some point after the sequence change. This variant type was also found in 13 of the 38 individuals in the same study (Kumble 2022)
- Missense variants: Where one amino acid is replaced by another, producing an altered protein. In QRICH1, these have been found in the glutamine-rich (Gln-rich) domain and the domain of unknown function (DUF3504)
- Splice-site variants: These affect how genetic information is pieced together, which can cause parts of the protein to be missing (skipped)

All these types of variants have been reported as pathogenic or likely pathogenic. A nonsense variant, p.(Arg652*), has been identified in four unrelated individuals.

These variants are often thought to be loss of function variants (LOFs). Variants that shorten the protein (protein-truncating variants), such as nonsense and frameshift variants, are predicted to result in a non-functional protein that is broken down by the cell.



Genetic Tests

QRICH1-related syndrome caused by gene sequence variants, can be identified by a type of genetic test called sequencing, such as whole exome sequencing (WES) or whole genome sequencing (WGS). Gene deletions can also be identified by a sequencing test but are more commonly found using a different type of genetic test called a chromosome microarray (CMA), also known as an arrayCGH or SNParray.

Unique publishes separate guides to DNA sequencing, arrayCGH and SNParrays

Genetic Test Results

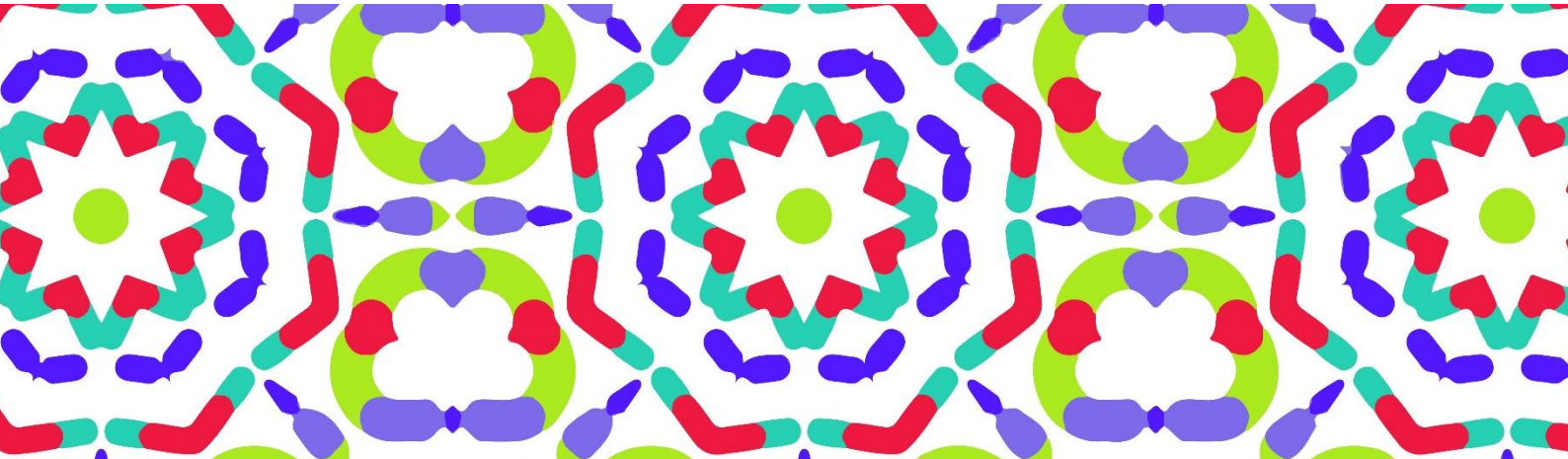
The results of genetic (genomic) testing are likely to be provided by a geneticist, a genetic counsellor or the clinician who ordered the test. Depending on the test that was carried out, someone with a QRICH1-related syndrome might have results that look like one of these examples/the following example:

An example result of a DNA sequencing test (e.g. WES or WGS), that can identify gene variants, is shown here for the QRICH1 gene:

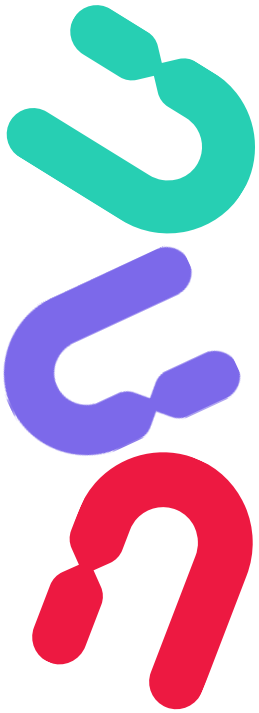
QRICH1 NM_017730.2:c.1954C>T: p.Arg652Ter

p.Arg652Ter	signifies the change to the protein: the amino acid arginine(Arg) has been replaced by a codon that terminates protein production, it stops another amino acid from being added to the chain. The three base codon sequence has been changed to a stop codon.
C>T	signifies the gene sequence change: the C nucleotide has been replaced by a T nucleotide
c.1954	signifies the base pair position of the change within the gene sequence (the position where the C nucleotide has been replaced by the T nucleotide)
QRICH1	signifies the gene that is affected
NM_017730.2	this is the specific ‘reference sequence’ or blueprint of the gene that scientists use to identify the location of the variant

Unique publishes a separate guide to Interpreting Genetic Test Results



Why did this happen?



When children are conceived, their parents' genetic material (DNA) is copied in the egg and sperm that makes a new child. The biological copying method is not perfect, and random changes occur in the genetic code of all children that are not seen in the DNA of their parents. This happens naturally and is not due to the parents' diet, environment or lifestyle. Most of these DNA changes have no obvious effect. But in rare instances these random DNA changes can lead to health issues or affect development. When such a random change disrupts the function of the QRICH1 gene then a child will have QRICH1-related syndrome (also known as Ververi-Brady syndrome).

In most people identified with QRICH1-related syndrome, the genetic change was a random (or 'de novo') change, meaning the change occurred for the first time in the egg or sperm that went to make up the affected individual. Occasionally QRICH1 variants are inherited from an affected parent (typically the parent has mild features). Very rarely one parent may have the same change (or variant) in some of their egg or sperm cells and pass it on to their child (this is known as germline mosaicism). However, it is important to recognize that no one should be blamed for variants in their DNA and no parent is at fault when a new DNA change occurs in their child.

Can it happen again?

The possibility of having another child affected by a rare gene disorder depends on the genetic code of the parents. In almost everyone reported with QRICH1-related syndrome so far (2026) the genetic change has been found to be de novo (dn), which means neither parent was found to have the same QRICH1 gene change as their child. Therefore, the chance of having another child with QRICH1-related syndrome is low, usually less than 1%.

One reason why there is some residual chance of recurrence is due to the rare phenomenon called germline mosaicism that was mentioned above. This is when a parent carries a genetic change, but it is limited to some of their egg or sperm cells. The genetic change would not, therefore, be detected in the parents' blood tests.

Unique publishes a short general guide to Mosaicism

In families where the QRICH1 variant has been inherited from a parent, the condition follows an autosomal dominant pattern of inheritance, and the possibility of having another child - either a girl or a boy - with QRICH1-related syndrome is 50% (1 in 2) in each pregnancy. However, the effect on the child's development, health and behaviour cannot be reliably predicted. The clinical presentation is highly variable, and a parent with mild symptoms can have a more significantly affected child. Your genetics centre should be able to offer counselling before you have another pregnancy.

If your child with a QRICH1 variant were to go on to have children of their own, the chances of passing on the variant to their child would be 50% in each pregnancy.

A clinical geneticist or genetic counsellor can provide specific advice for each family about the chance of having further children with QRICH1-related syndrome, including discussing their specific test results, recurrence risks, and reproductive options.

Unique publishes separate guides to Planning your next child, Prenatal genetic testing and diagnosis, A clinical genetics appointment and Supporting siblings of children with a rare genetic condition

Can QRICH1-related syndrome be cured?

There is no cure for QRICH1-related syndrome since most of the effects of the genetic change took place during a baby's formation and development. However, knowing the diagnosis means that appropriate monitoring and interventions can be put in place.

Management

No clinical practice guidelines for QRICH1 syndrome have been published (2026). The following suggestions have been provided by clinicians, who have personal experience of managing/treating individuals with QRICH1-related syndrome, to improve quality of life and reduce complications.

Children and adults with QRICH1-related syndrome should be under the care of a multidisciplinary team. The team should include a geneticist and paediatrician (for children) who can oversee care so that growth, development and behaviour can be monitored, and the best help given in the form of different therapies. Individuals may also have evaluations with various specialist doctors.

Immediately following diagnosis

When not carried out as part of the diagnostic process, an evaluation of the features of QRICH1-related syndrome that are present in the child or adult who has been diagnosed with this genetic condition is recommended. This can determine which of the features of QRICH1-related syndrome are present and how severe they are.

The following assessments are recommended:

- A physical examination to look for low muscle tone (hypotonia), spinal curvature (scoliosis or lumbar hyperlordosis) and unusual facial features
- A cardiac assessment to look for congenital heart anomalies
- A developmental and behavioural assessment to evaluate developmental milestones, motor and language skills, and to screen for Autism Spectrum Disorder (ASD) only if concerns are evident
- A growth and feeding assessment, including regular monitoring of height, weight and head circumference

If seizures or neurological signs are apparent, an EEG or brain MRI may be recommended. A referral to a feeding specialist may be required if feeding difficulties are severe and to an endocrinologist for significant growth concerns.

Supportive care

How a person with QRICH1-related syndrome (Ververi-Brady syndrome) is cared for is likely to require co-ordinated care by a multidisciplinary team of specialists, often overseen by a care coordinator such as a developmental paediatrician or clinical geneticist. The team may include a/an:

Paediatrician – a doctor who specialises in the physical, mental and social health of children from birth to young adulthood

Neurologist – a doctor who specialises in conditions of the brain, spinal cord and nervous system

Cardiologist – a doctor who specialises in heart conditions

Endocrinologist – a doctor who specialises in hormones and their effect on the body, particularly in managing growth as short stature can affect around one-third of individuals

Gastroenterologist – a doctor who specialises in the digestive system and can help manage feeding issues and gastro-oesophageal reflux disease (GORD)

Nephrologist – a doctor who specialises in conditions affecting the kidneys

Orthopaedic specialist/surgeon – a doctor who monitors and treats skeletal issues such as scoliosis

Occupational therapist (OT) – a health care professional who uses activities to aid self-management of a condition and can provide equipment

Physiotherapist/physical therapist (PT) – a health care professional who uses exercise, movement, manual therapy, education and advice to help with the body's strength and mobility

Speech and language therapist (SALT) – a health care professional who helps with speech, language communication and sometimes feeding/swallowing difficulties

Nutritionist/Dietitian and/or **Feeding therapist** – healthcare professionals who create plans to address poor weight gain and improve feeding skills

Psychologist/Psychiatrist – doctors who specialise in mental health and can diagnose and manage behavioural conditions, including Autism Spectrum Disorder (ASD)

Clinical Geneticist and/or **Genetic Counsellor** – healthcare professionals who can help with diagnosis, interpret genetic results and refer families to support services

Social worker – can connect families with financial, emotional, and community resources

Specialist nurses and/or other **healthcare professionals** may need to systematically and comprehensively plan a child or adult's treatment.

Treatments and therapies

Early intervention can be beneficial and formal testing to assess specific, individual needs is recommended. An education, health and care plan (EHCP) in the UK, individualized education plan (IEP) in the US, or equivalent document in other countries, may be issued after a child has undergone an assessment, to help ensure that the educational, health and social provisions deemed necessary to support the child's needs are delivered.

Management is supportive and focuses on addressing an individual's specific symptoms through a multidisciplinary team. There is currently no curative treatment for the underlying genetic cause. Treatment will depend on the specific features and symptoms experienced by the person with QRICH1-related syndrome but may include:

Physiotherapy for low muscle tone (hypotonia), gross motor delays, and an unsteady walk (gait ataxia), which usually includes exercises to build core strength, improve balance, coordination, and postural control to help children achieve motor milestones like sitting, crawling, and walking, and to improve overall mobility

Occupational therapy for fine motor skill delays, visual-motor skills, and feeding challenges, which can include activities to improve hand strength and coordination for daily tasks (e.g. feeding and dressing). It can also help to improve social communication and interaction

Speech therapy for a range of speech and language delays or difficulties, including significant expressive and receptive delays

Behavioural therapy for features of autism, ADHD, anxiety, hyperactivity, low frustration tolerance, and social withdrawal, which can include structured therapies like Applied Behaviour Analysis (ABA) to help manage challenging behaviours, improve social skills, and develop coping strategies

Medications may be prescribed such as standard anti-epileptic medication to manage seizures, e.g. anti-seizure medications (ASMs) to help control or prevent seizures

Hormone treatment for short stature to improve growth (a feature seen in about a third of reported cases). In a small study, two out of three individuals treated with growth hormone showed a positive response, though larger studies are needed to confirm its effectiveness

Surgery, such as the placement of a feeding tube to manage feeding difficulties which can lead to poor weight gain, e.g. the placement of a gastrostomy tube (G-tube) for severe, long-term feeding difficulties to ensure adequate nutrition

Regular monitoring for skeletal anomalies such as a curve of the spine (scoliosis) and an inward curve of the lower back (lumbar hyperlordosis)

Research into new treatments for QRICH1-related syndrome

The genetic change causing QRICH1-related syndrome affects development of the brain and other parts of the body before birth. Therefore, a complete cure is unlikely, even in the future, since the brain has already formed by the time a diagnosis is made. However, research into improved treatments and management for various features of QRICH1-related syndrome, like e.g. autism, is ongoing. As of early 2026, there are no specific curative treatments, gene therapies or targeted drugs in clinical trials for the condition. The clinical approach is supportive, focusing on managing the specific symptoms of each individual through a multidisciplinary team.

In addition, although QRICH1-related syndrome is a relatively rare condition, the QRICH1 gene is the subject of a lot of research, which is currently in a foundational phase. Scientists are working to define the full clinical spectrum of the syndrome and to understand the function of the QRICH1 gene. This includes functional studies to learn how genetic changes affect cellular processes, which may reveal new pathways for therapeutic intervention. The path to developing new treatments involves natural history studies, biomarker discovery and preclinical research in cell and animal models. Participation in research registries is encouraged to help advance understanding and accelerate research.

Details of clinical trials related to a particular condition or gene can be found at **ClinicalTrials.gov** and **EU Clinical Trials Register**.

Sources

The information in this booklet is drawn from the published medical literature and information from Unique members. In 2026, Unique had 10 members with QRICH1-related syndrome. The first-named author and publication date for articles in the medical literature are given to allow you to look for the abstracts or original articles on the internet in PubMed (**pubmed.ncbi.nlm.nih.gov/**).

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Inform Network Support

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

Join Unique for family links, information and support:

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Websites, Facebook groups and other links:

- Ververi-Brady Syndrome (QRICH1) Support Group (Facebook)

This guide was reviewed by Dr Angela F Brady. Consultant in Clinical Genetics, North West Thames Regional Genetics Service | Northwick Park Hospital, UK.

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