UNIQUE

UNDERSTANDING GENES & CHROMOSOMES

SLC2A1 (GLUT1) glucose transporter type 1 deficiency syndrome

rarechromo.org

This guide is designed to help families and healthcare professionals looking after people with SLC2A1 (GLUT1) glucose transporter type 1 deficiency 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 SLC2A1 (GLUT1) glucose transporter type 1 deficiency syndrome?

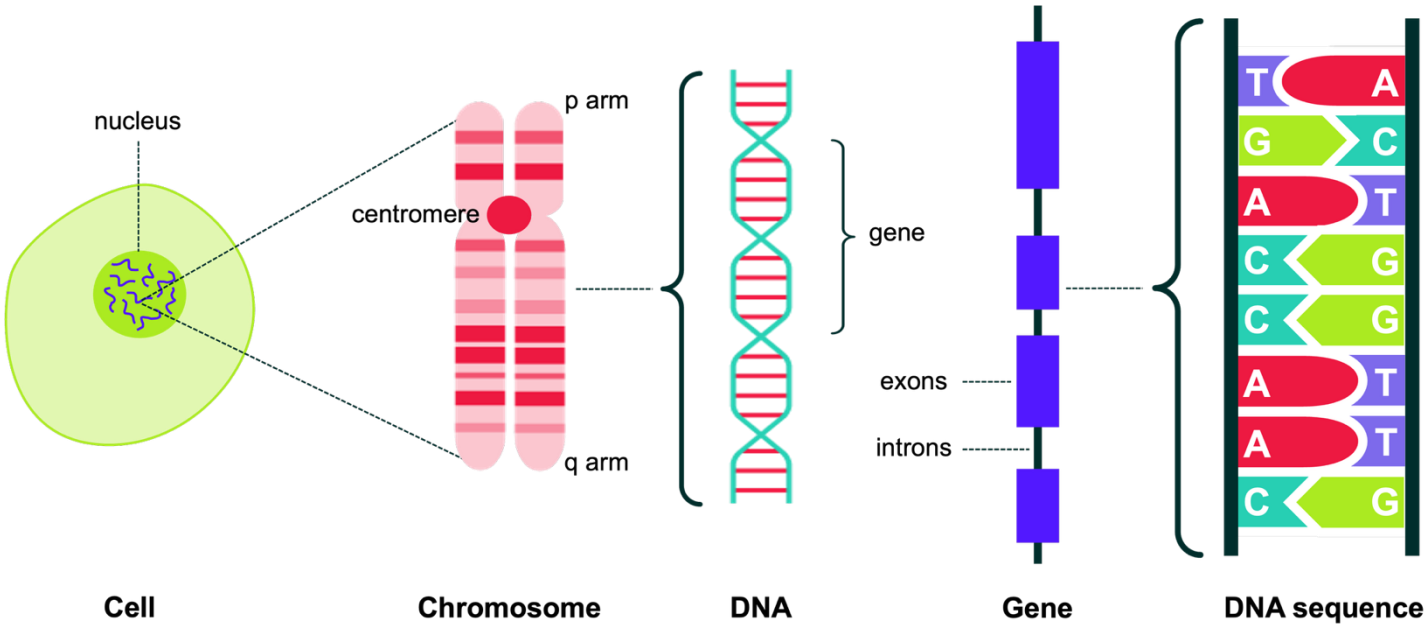
SLC2A1 (GLUT1) glucose transporter type 1 deficiency syndrome, also referred to as GLUT1 deficiency syndrome, GLUT1-DS, and De Vivo disease, is a rare genetic condition associated with movement difficulties, seizures, speech difficulties, and developmental delay. As is common with genetic conditions, each person can be affected differently - even among affected members within the same family. Not everyone with GLUT1 deficiency 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.

GLUT1 deficiency syndrome is caused by a change (variant) in the SLC2A1 gene or the loss (deletion) of one copy of the SLC2A1 gene. The loss of the gene may occur as part of a larger deletion that affects the chromosome on which the gene is located.

What causes GLUT1 Deficiency Syndrome?

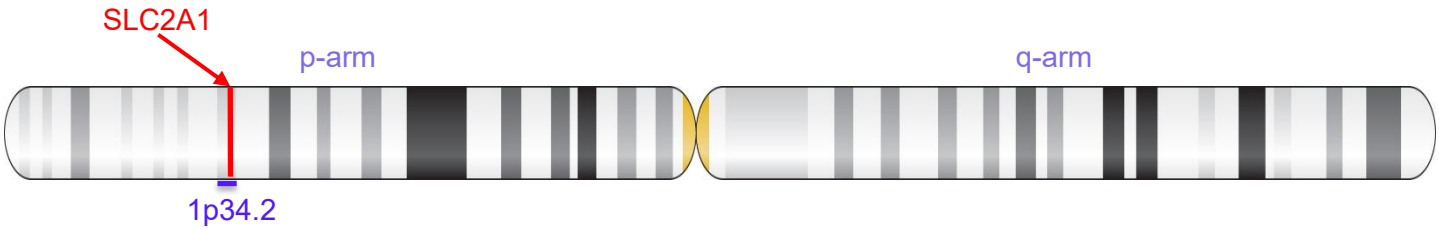
Genes are instructions that have important roles in our growth and development. They are made of DNA and are incorporated into organised structures called chromosomes. Chromosomes therefore contain our genetic information. Chromosomes are located inside our cells, the building blocks of our bodies. In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



GLUT1 deficiency syndrome is caused by specific changes (known as pathogenic variants) to the DNA sequence of a gene called SLC2A1 (SLC2A1 is an abbreviation of the gene's full name, solute carrier family 2 member 1). GLUT1 deficiency syndrome can also be caused by the loss of the SLC2A1 gene due to a chromosome deletion. The SLC2A1 gene is located in the short 'p' arm of chromosome 1 in a region called 34.2 as shown in the image below.

Chromosome 1



We have two copies of chromosome 1 in our cells, so we also have two copies of the SLC2A1 gene. GLUT1 deficiency syndrome occurs when only one copy of the SLC2A1 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

Unique publishes a separate guide to deletions and microdeletions

The SLC2A1 gene sequence is used to make a protein called GLUT1. This protein is important because it is responsible for transporting glucose, which is the body's main source of energy. More specifically, GLUT1 moves glucose from the bloodstream into the brain across the blood-brain barrier (BBB).

Genetic Tests

GLUT1 Deficiency Syndrome, caused by gene sequence variants, can be identified by a type of genetic test called sequencing (e.g. 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, e.g. 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 given to you by your geneticist, a genetic counsellor or the clinician who ordered the test. Depending on the test that was carried out, someone with GLUT1 deficiency syndrome might have results that look like one of these examples.

An example result of a DNA sequencing test (e.g. whole exome sequencing (WES) or whole genome sequencing (WGS)), that can identify gene variants, is shown here for the SLC2A1 gene:

p.Arg333Trp (R333W) (CGG>TGG): c.997 C>T in exon 8 of the SLC2A1 gene (NM_006516.4)

p.Arg333Trp (R333W)	signifies the change to the protein: the amino acid arginine (Arg or R) has been replaced by the amino acid tryptophan (Trp or W) at position 333 in the sequence of amino acids that make up the protein
CGG>TGG	signifies the gene sequence change; the C nucleotide has been replaced by a T nucleotide
c.997	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)
exon 8	signifies which part of the gene has been altered, in this case exon 8
SLC2A1 gene	signifies the gene that is affected
NM	denotes the reference sequence used

The result of a chromosome microarray (CMA) test (e.g. arrayCGH or a SNParray), that can identify deletions, is shown here for a microdeletion within band 1p34.2:

arr[hg38] 1p34.2 (42,936,704-42,957,669)x1 dn

arr	The analysis was by array (arr) comparative genomic hybridisation (cgh)
hg38	Human Genome build 19. This is the reference DNA sequence that the base pair numbers refer to. As more information about the human genome is found, new 'builds' of the genome are made, and the base pair numbers may be adjusted. This means base pair positions change depending on the assembly used
1p34.2	the chromosome involved is chromosome 1 and the position of the deletion is in band p34.2
42,936,704-42,957,669	the base pairs between 42,936,704 and 42,957,669 have been shown to be deleted. Take the first long number from the second and you get 20,965 (0.021Mb or 21kb). This is the number of base pairs that are deleted
x1	means there is one copy of these base pairs, not two – one on each chromosome 1 – as you would normally expect, so this a deletion
dn	means *de novo*. The biological parents' chromosomes have been checked and no deletion or other chromosome change has been found at position 1p34.2. The deletion is very unlikely to be inherited and has almost certainly occurred for the first time in this family with this child mat here would mean that the deletion has been inherited from the mother pat here would mean that it has been inherited from the father

Unique publishes a separate guide to Interpreting Genetic Test Results

What features and symptoms do people with GLUT1 deficiency syndrome have?

As is common with many genetic conditions, children and adults with GLUT1 deficiency 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 children with GLUT1 deficiency syndrome have:

- Involuntary (paroxysmal) movements, particularly of the eyes and head
- Seizures, which often reduce in late childhood
- Small head size (microcephaly)
- Some degree of developmental delay ranging from mild to severe
- Some degree of intellectual disability (ID) or learning difficulties (LD) ranging from mild to severe
- Speech and language delay/non-verbal
- Low muscle tone (hypotonia)
- Motor weakness

Other possible features include:

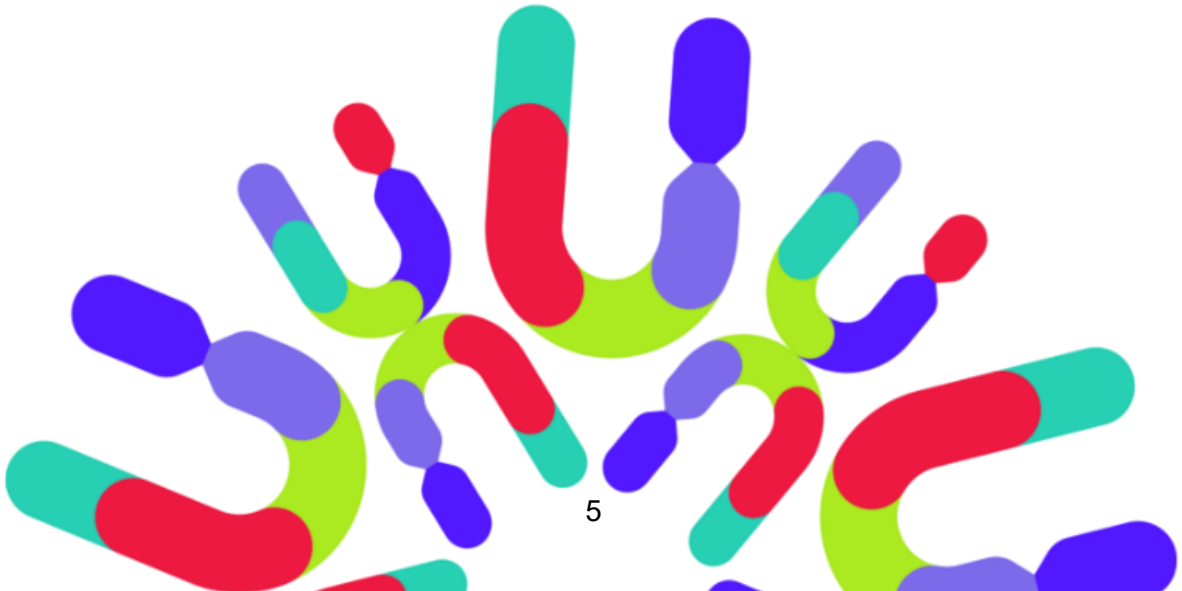
- Muscle spasms
- Small stature
- Migraine
- Cyclical vomiting
- Feeding difficulties
- Sleep issues

Pregnancy and birth

Pregnancy complications are not commonly reported for this condition.

Appearance

Children with GLUT1 deficiency syndrome will often have a smaller head size (microcephaly) than those without the syndrome. A few children have also been reported to be small in height. Other than this, no other changes to appearance have been noted.



Development

Gross and fine motor skills

Developmental delay has been reported in all children with GLUT1 deficiency syndrome so far (2025). The degree of delay ranges from mild to severe. Developmental “milestones”, including rolling, sitting, walking, playing with toys, using cutlery, using zips and buttons, and toilet training, are often delayed, although there is a wide range of eventual ability, with some children acquiring mobility and other skills around the same age as a typically developing child and others showing more obvious delay. Low muscle tone (hypotonia) is common and may affect mobility. Some children may have an unusual gait when walking because of stiffness, or balance issues (known as ataxia). For some, independent walking may not be achieved. Many children benefit from early intervention with treatments or therapies such as orthotics e.g. insoles, braces, splints and callipers; occupational therapy (OT); and physiotherapy (PT).

Unique publishes separate guides to Therapies and Toilet training and continence

Intellectual development and learning

All children with GLUT1 deficiency syndrome have intellectual disability (ID) or learning difficulties. ID ranges from mild to severe but is usually in the moderate range and most children have needed additional support with their learning. Early intervention can prove particularly beneficial and formal testing to assess specific, individual needs is recommended.

Unique publishes separate guides to Education and Further education, training and work

Speech and language

Children with GLUT1 deficiency syndrome typically experience some degree of speech and language delay and a few may find it difficult to co-ordinate movement of their lips, jaw and tongue to make the right sounds (apraxia of speech). The eventual range of achievement is broad, but a few may remain non-verbal. Those who do develop speech may achieve single words, short phrases or basic sentences and some go on to develop conversational skills and a broad vocabulary. Many parents say that their child can understand more than they can express.

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. Where individuals have no speech or very few words, Augmentative and Alternative Communication (AAC) methods, including pointing, pictograms, gestures, facial expression and simplified sign language and high-tech communication systems (aided communication) have enabled some to communicate their thoughts and needs well.

Unique publishes a separate guide to Communication

Feeding

Feeding issues in the newborn period are common in babies with GLUT1 deficiency syndrome. Low muscle tone may contribute to difficulties with swallowing. Some babies will suck weakly and may need high energy milks to encourage weight gain.

Unique publishes a separate guide to Feeding

“Although the ketogenic diet feels daunting. It's very hard work for everyone involved, and has significant social implications. Yet, it's worth it. The change in our child was enormous. We finally got to know our child, as a person, as he had enough energy to play, chat, make friends, take up hobbies, and be naughty occasionally.”

Growth and stature

A few children with GLUT1 deficiency syndrome described in the medical literature so far (2025) are noted as having short stature. Most of these children also have a smaller than expected head size (microcephaly). Beyond infancy, height and weight is variable.

Behaviour

Children with GLUT1 deficiency syndrome typically tend to have behaviour in keeping with their overall degree of developmental delay. However, most have a happy disposition, engage happily with others, and are very understanding of others. Some children may have an autism spectrum disorder (ASD) diagnosis or traits. Other behaviours including attention deficit hyperactivity disorder (ADHD) have also been reported. Some children also have sleep problems. Children usually benefit from consistent routines, boundaries, rewards and other behaviour management techniques. Efforts to take into account and introduce strategies to tackle communication and other difficulties can also be beneficial.

Unique publishes separate guides to Challenging Behaviour and Sleep

Puberty

There is limited information available about puberty in children with GLUT1 deficiency syndrome. We do know that most boys and girls appeared to go through puberty as expected. Some families of children with chromosome disorders and behavioural or learning difficulties 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

Experiences of adulthood are likely to vary considerably and will depend on many factors. These include the level of any learning or intellectual disability, possible on-going medical concerns and improvements in early intervention and therapies/treatments.

Adults with GLUT1 deficiency syndrome have varying levels of independence. Some have gone on to have families of their own and manage to run their own households and

work. Some continue to live with their parents or in supported settings such as a group/residential care home, with caregivers who can provide support. A few may live independently but require some support from family or friends with certain tasks. Levels of employment and the nature of employment varies but some do undertake some form of paid or unpaid employment.

Several adults with GLUT1 deficiency syndrome only discovered that they had an SLC2A1 gene variant when their child was diagnosed.

Some features may change in adulthood, for example, seizures (as described in the medical concerns section below) may decrease and movement difficulties may become more severe.

Unique publishes a separate guide to Transition

Medical concerns

The following medical concerns have been found in children with GLUT1 deficiency syndrome. They are not found in all children so not all children with GLUT1 deficiency syndrome will be affected.

Brain

Most children GLUT1 deficiency syndrome have a brain anomaly, which can be detected by a CT (computerised tomography) scan of their brain. The changes seen vary but mainly show a reduction in brain volume (cerebellar atrophy).

Seizures

Many children with GLUT1 deficiency syndrome experience some form of seizure (a sudden and unexpected change in the electrical activity in the brain). Depending on the part(s) of the brain affected, symptoms vary, but include temporary confusion, uncontrollable jerking movements and loss of consciousness or awareness. Age of onset can vary considerably, while seizures may be isolated to a single incident or occur more regularly and many children have epilepsy.

Although seizures from GLUT1 deficiency syndrome tend to be resistant to drug treatment, levetiracetam, sodium valproate, acetazolamide, ethosuximide and lamotrigine have been used for seizure control. A ketogenic diet may also improve seizure control.

More than one type of seizure may be present in the same individual.

Electroencephalograph (EEG) and video telemetry (video EEG) are medical tests that can be used to measure and record the electrical activity of the brain and are tools that, when used alongside other tests, can help diagnose the type of seizure experienced.

Seizures can cause a lot of worry for families and can be frightening to observe, but in some cases they self-resolve or resolve with medical treatment. If your child has a seizure for the first time, it is important to remove nearby hazards so they can't hurt themselves and contact a medical professional.

Seizure types in children with GLUT1 deficiency syndrome include:

Absence seizure: 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.

Atypical absence seizure: a child may appear confused and unresponsive for minutes (very different from a typical absence seizure).

Generalised tonic clonic seizure: at the onset of a seizure, the abnormal electrical activity involves both sides of the brain. The seizure involves a phase of stiffening of the body followed by jerking.

Myoclonic generalised seizure: involving jerky or shock-like contraction of different muscles anywhere in the body but usually the arms or legs. Each myoclonic seizure lasts for a fraction of a second or a second at most.

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.

Movement

Problems with movement are common in children with GLUT1 deficiency syndrome. The main concern is that children can end up falling or hurting themselves.

Eyes

Eye movement anomalies are common in children with GLUT1 deficiency syndrome. The main anomaly is rapid involuntary eye movements, which is often accompanied by rapid and involuntary head movements. Again, the main concern is that children can fall or hurt themselves.

Migraine

Some individuals with GLUT1 deficiency syndrome have intense headaches (migraines).

Sleep

Sleep disturbances can occur in some children with GLUT1 deficiency syndrome but it is not a key feature of this syndrome.

Unique publishes a separate guide to Sleep

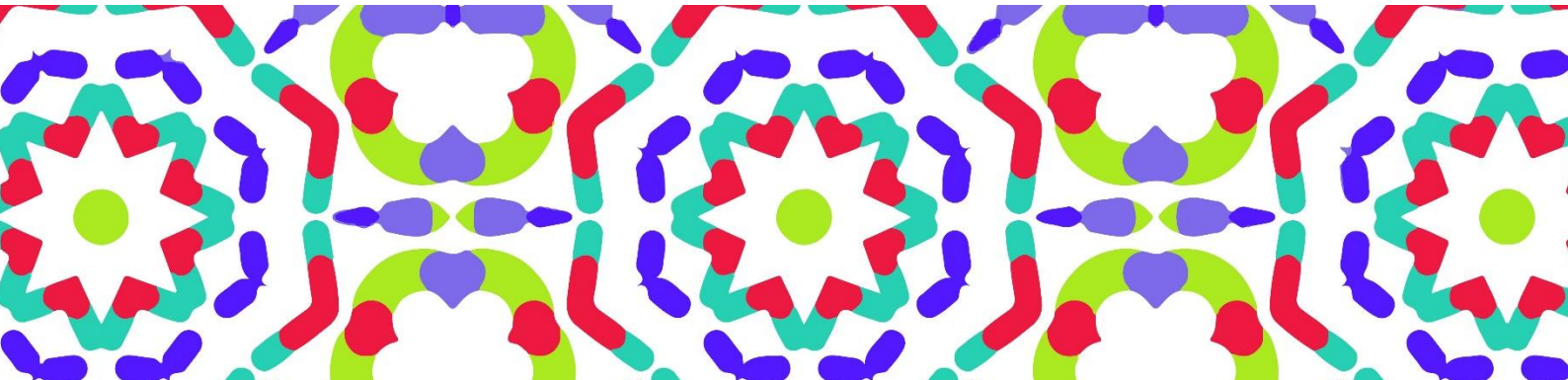
Feeding

Feeding difficulties can occur in some children with GLUT1 deficiency syndrome. Some children might benefit from attending a feeding clinic where an assessment can be made, and advice to help treat any eating and drinking difficulties provided.

Unique publishes a separate guide to Feeding

Breathing

Babies and children with rare chromosome and gene disorders tend to have a higher rate of respiratory concerns, which may become less frequent with age and maturity, although they can persist throughout childhood. Children with GLUT1 deficiency syndrome can experience frequent respiratory illnesses.



How common is GLUT1 deficiency syndrome?

GLUT1 deficiency syndrome is extremely rare. Currently (2025) more than 500 individuals with a SLC2A1 gene variant have been reported in the medical literature. It is expected that more people will be diagnosed with this condition as awareness increases and genetic testing becomes more routine. One estimate suggests that around 105,000 people may have GLUT1 deficiency syndrome without knowing it.

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 SLC2A1 gene then a child will have GLUT1 deficiency syndrome. In almost all people identified so far (2025) with GLUT1 deficiency syndrome, the genetic change was a random (or "de novo") change, meaning the change occurred for the first time in that family in the affected individual. Very rarely, one parent may have a chromosomal rearrangement that led to GLUT1 deficiency syndrome in their child, or 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 GLUT1 deficiency syndrome so far (2025) the genetic alteration has been found to be de novo (dn), which means neither parent was found to have the same SLC2A1 gene change as their child, and neither parent was found to have a chromosomal rearrangement that might have resulted in a SLC2A1 deletion in their child. Therefore, the chance of having another child with GLUT1 deficiency syndrome is 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. Parental genetic testing has been reported many times in the medical literature for individuals with GLUT1 deficiency syndrome and parental mosaicism for a SLC2A1 variant has been reported a few times.

Unique publishes a short general guide to Mosaicism

In families where the SLC2A1 variant has been inherited from a parent, the possibility of having another child - either a girl or a boy - with GLUT1 deficiency syndrome rises to 50% (1 in 2) in each pregnancy. However, the effect on the child's development, health and behaviour cannot be reliably predicted. Your genetics centre should be able to offer counselling before you have another pregnancy.

If your child with a SLC2A1 variant goes on to have children of their own, the chances of passing on the variant to their child are 50% in each pregnancy. Your child's ability to look after their own child is very likely to be closely related to their own learning ability and behaviour.

A clinical geneticist or genetic counsellor can provide specific advice for each family about the chance of having further children with GLUT1 deficiency syndrome.

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 GLUT1 deficiency syndrome be cured?

There is no cure for GLUT1 deficiency syndrome since 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 GLUT1 deficiency syndrome have been published (2025). The following suggestions have been provided by clinicians, who have personal experience of managing/treating individuals with GLUT1 deficiency syndrome, to improve quality of life and reduce complications.

Children and adults with GLUT1 deficiency 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 development and behaviour can be monitored, and the best help given in the form of physiotherapy, occupational therapy, speech therapy and, if needed, behavioural therapy. Individuals may have evaluations with neurology, physiotherapy services, speech and language services, nutrition and dietetics, and gastroenterology.

Immediately following diagnosis

When not carried out as part of the diagnostic process, an evaluation of the features of GLUT1 deficiency syndrome that are present in the child or adult who has been diagnosed with this genetic condition should be carried out. This can determine which of the features of GLUT1 deficiency syndrome are present and how severe they are.

Supportive care

Children with GLUT1 deficiency syndrome are likely to be under the care of a multidisciplinary team. The team should include a community or hospital paediatrician who can oversee care; monitor growth, development and behaviour; and link in with affiliated services.

How a person with GLUT1 deficiency syndrome is supported is likely to require co-ordinated care by a team of specialists, which 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.

Ophthalmologist – a doctor who specialises in conditions affecting the eyes.

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

Physiotherapist (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.

Dietician – a healthcare professional who specialises in diet.

Psychiatrist – a doctor who specialises in mental health.

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 prove particularly 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

Treatment will depend on the specific features and symptoms experienced by the person with GLUT1 deficiency syndrome but may include:

A ketogenic diet which means removing carbohydrates (e.g. pasta, potatoes, rice, sugar etc.) as much as possible, and replacing these with protein-rich foods as well as unsaturated fat-rich foods which act as an alternative fuel for the brain.

Physiotherapy for seizures, low muscle tone, and muscle spasms, which usually includes gait training, stretching, hydrotherapy, and balance exercises. This is to give children more control over their movements and relax their muscles, so that fewer falls are likely to occur.

Occupational therapy for motor weakness to help children with fine motor skills such as dressing, grooming, feeding, and writing.

Speech therapy for speech and language delay, which can include improving articulation, fluency, and language comprehension to help improve communication. Alternative communication devices (AAC) may also be used.

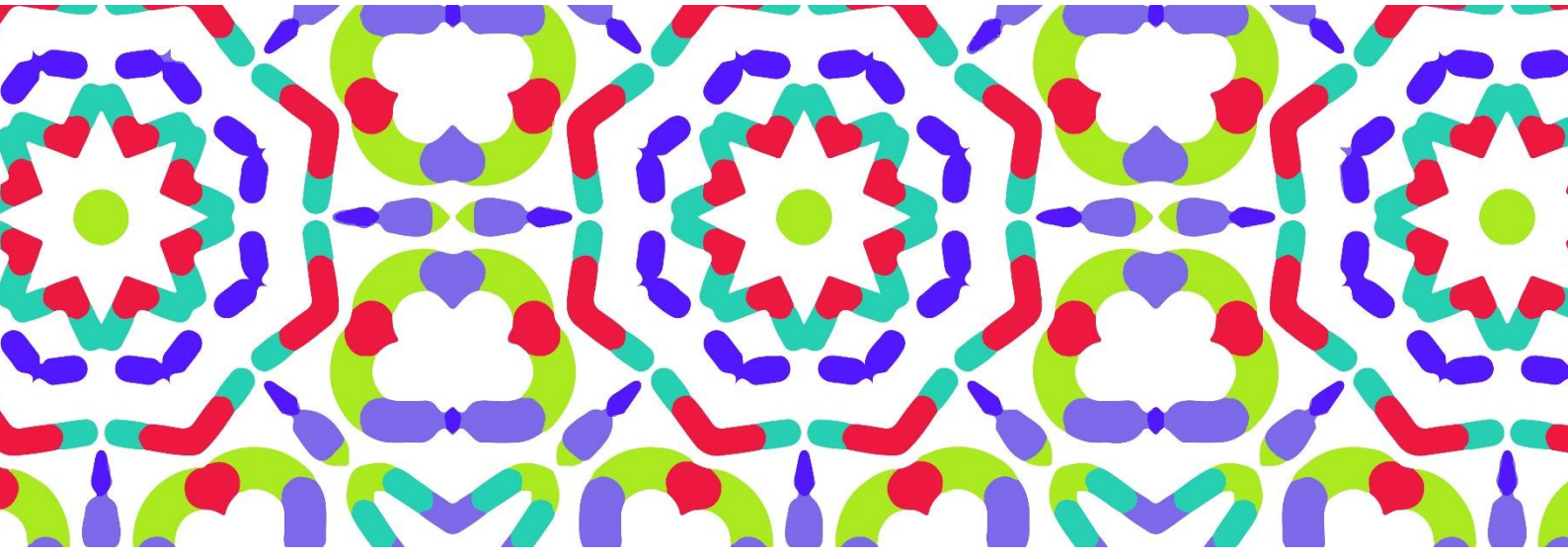
Medications may be prescribed to ease the different symptoms of GLUT1 deficiency syndrome including seizures, ADHD or cyclical vomiting. Some examples include levetiracetam (for seizures), carbidopa/levodopa (for movement disorders), methylphenidate hydrochloride (for ADHD), and antiemetics (for vomiting).

Behavioural therapy as needed.

Surveillance

It is recommended that the following evaluations are carried out to monitor an individual's existing symptoms, how they respond to care and treatment, and whether any new symptoms emerge over time:

- Consider a neurologic review (including MRI and EEG, if indicated by seizures)
- Consider a physiotherapy review to evaluate any movement difficulties experienced
- Consider a speech and language review
- Consider an occupational review
- Consider a dietary review to evaluate the ketogenic diet treatment
- Consider an ophthalmology review to evaluate involuntary eye movements



Research into new treatments for GLUT1 deficiency syndrome

The genetic change causing GLUT1 deficiency 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 GLUT1 deficiency syndrome, such as seizures and movement difficulties, is ongoing.

In addition, although GLUT1 deficiency syndrome is a relatively rare condition, the SLC2A1 gene is the subject of a lot of research. The current focus of research is finding out if gene therapy could become a treatment option for people with this condition. This is because gene therapy has been shown to improve symptoms in mice. Gene therapy is a technique where a copy of the SLC2A1 gene with a pathogenic variant is replaced with an unaffected version. There is also some weak evidence that other drugs may help in controlling the symptoms of GLUT1 deficiency syndrome, and so these drugs are likely to be studied further.

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 2025, Unique had 8 members with GLUT1 deficiency 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 (**https://pubmed.ncbi.nlm.nih.gov/**). You can obtain most articles from Unique.

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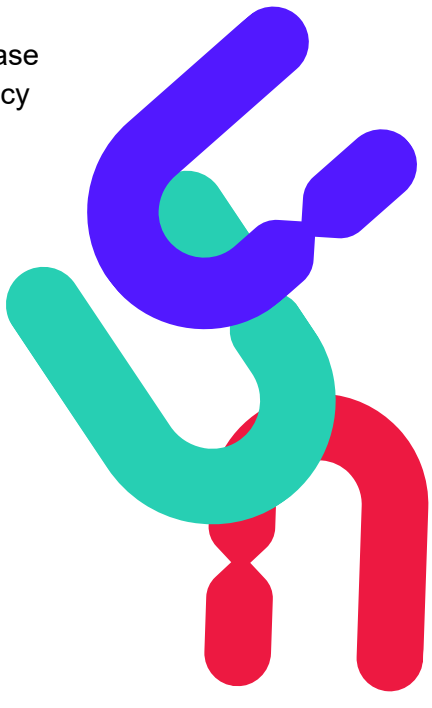
* Note: an asterisk indicates articles which are “open access” and available to everyone at **https://pubmed.ncbi.nlm.nih.gov**



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

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Websites and Facebook groups:

- Glut1 Deficiency UK
- Glut1 Deficiency UK Facebook group
- Glut1 Deficiency Foundation (US)
- Glut1 Deficiency Foundation Facebook group
- Glut1 Deficiency Australia Facebook group
- Glut1 Deficiency Syndrome
- GLUT1 DS Care and support Facebook page
- Glut1 deficiency Facebook group Serbia

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 guide was written by Dr Adam Hodgson, Duncan Baker, Dr Charles F Cranston, Rhiannon Davies, Katie Jowett, Dr Sarah Patterson, Dr Emily Cottrell | Julia Garnham Centre, The University of Sheffield and Sheffield Children's NHS Foundation Trust in Partnership, and Unique (AP) and verified by Dr Sarah Wilson, Clinical Genetics Registrar | Sheffield Children's NHS Foundation Trust, UK.

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