UNDERSTANDING GENES
& CHROMOSOMES

# KMT2B-related dystonia

rarechromo.org

**This guide is designed to help families and healthcare professionals looking after people with KMT2B-related dystonia. 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 KMT2B-related dystonia?

KMT2B-related dystonia, also referred to as DYT-KMT2B, is a rare genetic condition associated with progressive, unexpected and uncontrolled muscle movement (dystonia) and varying degrees of developmental delay and learning disability. As is common with genetic conditions, each person can be affected differently - even among affected members within the same family. Not everyone with KMT2B-related dystonia will have all the possible features of this condition and each person with a certain feature won't necessarily be affected by it to the same level as other people with that feature.

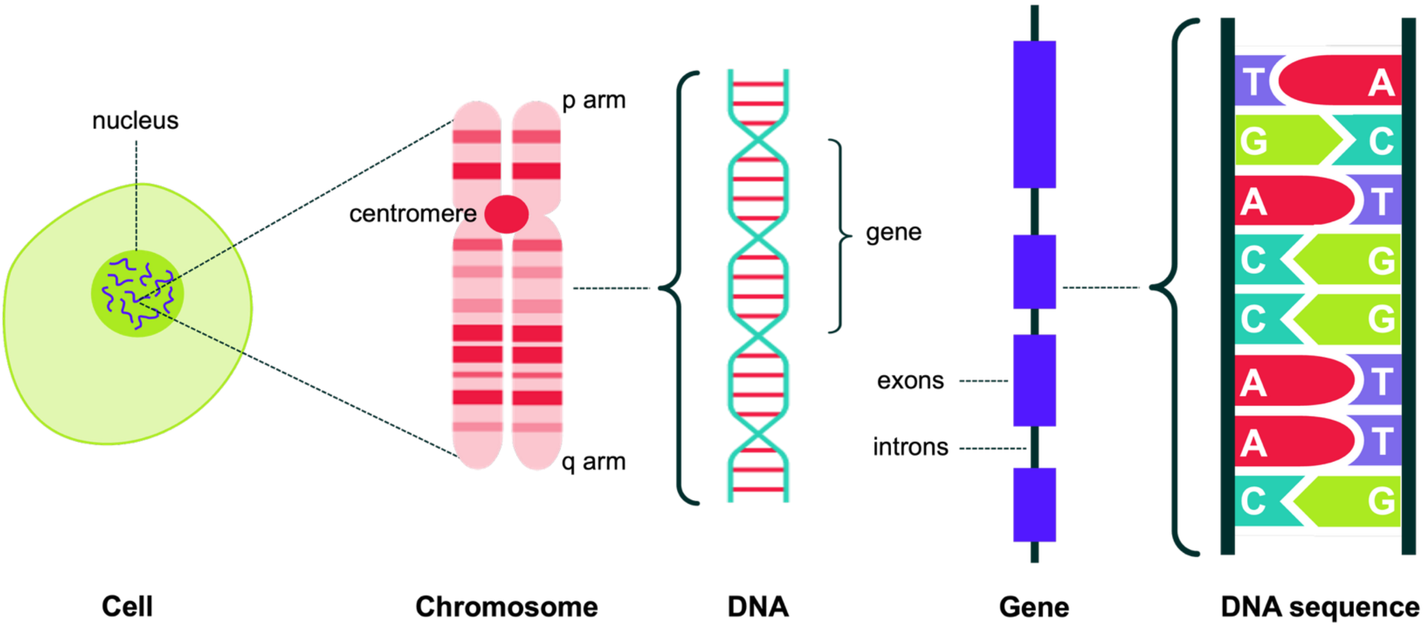
KMT2B-related dystonia is caused by a change (variant) in the KMT2B gene or the loss (deletion) of one copy of the KMT2B gene. The loss of the gene may occur as part of a larger deletion.

It is important to note that not everyone with a change to the KMT2B gene goes on to develop dystonia. A small percentage of people will have KMT2B-related neurodevelopmental disorder (NDD) with no dystonia. Even in families where more than one person has the same change to the KMT2B gene, it is possible for one family member to develop dystonia whereas another does not. People with KMT2B-related NDD can also have other features associated with KMT2B-related dystonia that are discussed in this guide, as well as additional features. This guide is aimed at people and families with KMT2B-related dystonia, but may also be of use to those with KMT2B-related NDD. It is also important to note that dystonia can develop much later in life.

## What causes KMT2B-related dystonia?

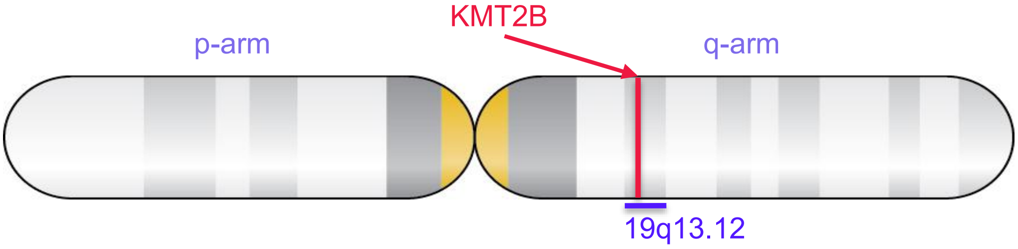
Genes are instructions that have important roles in our growth and development. They are made of DNA and are incorporated into organised structures called chromosomes. Chromosomes therefore contain our genetic information. Chromosomes are located inside our cells, the building blocks of our bodies. In 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.



KMT2B-related dystonia is caused by specific changes (known as pathogenic variants) to the DNA sequence of a gene called *KMT2B*. KMT2B-related dystonia can also be caused by the loss of KMT2B due to a chromosome deletion. The *KMT2B* gene is located in the long 'q' arm of chromosome 19 in a region called 19q13.12 as shown in the image below.

## Chromosome 19



We have two copies of chromosome 19 in our cells, so we also have two copies of the KMT2B gene. KMT2B-related dystonia occurs when only one copy of the KMT2B 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 KMT2B gene sequence is used to make a protein called Histone Methyltransferase 2B. This protein is important because it acts like a switch for other genes, controlling whether they turn on or how strongly they work.

## Genetic Tests

KMT2B-related dystonia 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 they 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 KMT2B-related dystonia might have results that look like one of the following 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 KMT2B gene:

**p.Gln204Ter (Q204*) (CAA>TAA): c.610C>T in exon 3 of the KMT2B gene (NM_014727.3)**

| | |
|---|---|
| p.Gln204Ter (Q204*) | signifies the change to the protein: the amino acid glutamine (Gln or Q) has been changed into a stop codon (*) at position 204 in the sequence of amino acids that makes up the protein |
| CAA>TAA | signifies the gene sequence change; the C nucleotide has been replaced by a T nucleotide |
| c.610 | 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 3 | signifies which part of the gene has been altered, in this case exon 3 |
| KMT2B gene | signifies the gene that is affected |
| NM_014727.3 | 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 19q13.12:

**arr[hg19] 19q13.11-13.12 (35,197,252–38,140,100)x1 dn**

| | |
|---|---|
| arr | The analysis was by array (arr) comparative genomic hybridisation (cgh) |
| hg19 | 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 |
| 19q13.11 -13.12 | the chromosome involved is chromosome 19 and the position of the deletion is in band q13.11 - 13.12 |
| 35,197,252–38,140,100 | the base pairs between 35,197,252 and 38,140,100 have been shown to be deleted. Take the first long number from the second and you get 2,942,848 (2.9 Mb). 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 19 – 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 19q13.11-13.12. The deletion is very unlikely to be inherited and has almost certainly occurred for the first time in this family with this child<br>mat here would mean that the deletion has been inherited from the mother<br>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 KMT2B-related dystonia have?

As is common with many genetic conditions, children and adults with KMT2B-related dystonia 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

### While all people with KMT2B-related dystonia have progressive uncontrolled dystonia, additional features can include:

- 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
- Swallowing difficulties that usually progress as the muscles in the face, mouth, and throat begin to move or clench on their own (oromandibular and laryngeal dystonia)

### Other possible features include:

- Irregular and uncontrolled eye movement (nystagmus)
- Skin changes
- Behavioural/psychiatric differences (e.g. autism spectrum disorder, attention-deficit/hyperactivity disorder (ADHD), anxiety, obsessive-compulsive disorder, and depression)
- Brief, jerking movements (myoclonus)
- Seizures
- Muscle stiffness (spasticity)
- Hearing loss
- Characteristic facial features, including an elongated face, bulbous nose and reduced head size (microcephaly)

## Pregnancy and birth

While almost all pregnancies are unremarkable and proceed without complication, some concerns during pregnancy have been reported. Where a cause for concern was noted, most often parents reported slow growth in the womb (intrauterine growth retardation), though this is very rare.

## Appearance

Certain facial features are found more often in children with KMT2B-related dystonia than in other children. These features may mean that you see unexpected similarities between your child and others with KMT2B-related dystonia. The most common characteristic features include an elongated face, a broad nose with a bulbous tip, a smaller head size (microcephaly), skin folds at the inner corner of the eye (epicanthal folds) and a high-arched palate.

> ***As an infant, he had some structural issues with his head and we did have a helmet to correct a flat spot. He also had trouble latching and breast-feeding.”***

# Development

## Gross and fine motor skills

Developmental delay has been reported in about a quarter of children with KMT2B-related dystonia 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 typically developing children and others showing more obvious delay.

> ***He has a sister who is exactly one year older than him, they share a birthday. So when he was younger, it was easy to compare milestones. I wouldn't say that he was super far behind on hitting majority of milestones, such as sitting walking and talking, but he was delayed in comparison to his sister."***

The most common feature of KMT2B-related dystonia is the progression of uncontrolled muscle tone and movement (dystonia). This is usually progressive, beginning with twisting, repetitive movements or unusual posture in the legs (lower limb dystonia). This often manifests as toe walking, and uncontrolled posturing of the foot, such as the foot rolling inwards (ankle inversion) or pointing as if on tip-toes (plantar flexion). This can also lead to differences in the way that children with KMT2B-related dystonia walk, such as in toeing (where feet turn inward while walking or running) or they may have frequent falls. Over time, the dystonia spreads to other parts of the body, including the arms, hands, throat and mouth. As the dystonia progresses, children may become unable to walk independently. Many benefit from early intervention with treatments or therapies such as orthotics, e.g. ankle-foot orthoses, walkers, occupational therapy (OT), and physiotherapy (PT).

While dystonia can start in early childhood, it can also develop later in life in adults of almost any age.

> ***So far, he has not shown any signs of KMT2B related to dystonia, and I'm hoping that it remains that way. He has endless energy is always on the go, and I would hate to see that impeded in anyway."***

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## Intellectual development and learning

Just under half of children reported with KMT2B-related dystonia to date (2026) have intellectual disability (ID) or learning difficulties. ID ranges from mild to severe, but is usually in the moderate range and many children have needed additional support with their learning. Early intervention can prove particularly beneficial and formal testing to assess specific, individual needs is recommended.

> ***His favorite subject is math and recess. He absolutely loves numbers, and entered kindergarten at a 3rd grade math level. He does show some tendencies of OCD, in regards to needing things to always be in even numbered groupings."***

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## Speech and language

Children with KMT2B-related dystonia often experience some speech and language delay which precedes their movement difficulties but can worsen as they progress. Many find it difficult to coordinate the movement of their lips, jaw, and tongue to make the right sounds (dysarthria), often because the muscles controlling these areas may move or clench on their own. Every child is different; some may have only mild speech difficulties, while others may have more noticeable challenges. Some children may remain non-verbal, but many are able to use single words, short phrases, or sentences, and some go on to develop conversational skills and a wide vocabulary. Parents often notice that their child understands much more than they are able to say. Low speech volume (hypophonia) is a common feature (Abela 2021).

A speech and language therapist can help identify your child's unique strengths and challenges, and regular therapy sessions can be tailored to support your child's communication skills. For children who have little or no speech, Augmentative and Alternative Communication (AAC) methods-such as pointing, using pictures or symbols, gestures, facial expressions, simple sign language, or high-tech communication devices-can be very effective. Many children have found these tools helpful for expressing their thoughts, needs, and feelings, allowing them to connect and communicate with those around them.

**Unique publishes a separate guide to Communication**

## Feeding

Feeding issues in the newborn period are common. Low muscle tone may contribute to difficulties with swallowing and some babies will suck weakly and may need high energy milks to encourage weight gain. Aspiration can also occur (where fluid, food, or saliva enters the airway or lungs). This happens because the muscles in the face, mouth, and throat begin to move or clench on their own (oromandibular and laryngeal dystonia). Some children have benefited from attending a feeding clinic where an assessment can be made, and advice to help treat any eating and drinking difficulties provided.

While many children require a PEG (percutaneous endoscopic gastrostomy) tube, which is a tube that is placed through the abdomen directly into the stomach, to deliver food (and sometimes medication), almost all infants progress to oral eating as they age. Children may 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**

## Growth and stature

Some children with KMT2B-related dystonia described in the medical literature so far (2025) are noted as having short stature. Many of these children also have a smaller than expected head size (microcephaly). The bone age of some children may also seem more advanced than their actual age. Beyond infancy, height and weight can remain below average.

## Behaviour

Children with KMT2B-related dystonia typically tend to have behaviour in keeping with their overall degree of developmental delay, and most have a happy disposition and display a great deal of adaptability and resilience. Some children have an autism spectrum disorder (ASD), diagnosis or traits. Other behaviours including attention deficit hyperactivity disorder (ADHD), anxiety, aggressive behaviours and obsessive compulsive disorder (OCD) have also been reported. 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.

> ***He is a happy, bubbly, go lucky six-year-old boy. He was the happiest baby I've ever met, and still maintains that positive attitude to this day."***

> ***He was diagnosed with autism spectrum disorder at the age of three. We did two years of in-home ABA therapy and supports with speech. He was diagnosed with ADHD in kindergarten and is closely looked after by the schools occupational therapist."***

> ***He has done exceptionally well, socializing with other kiddos and making friends in and out of school. He struggles with some behavioral issues at school, but nothing of malicious intent. Mostly related to his ASD, ADHD and endless energy."***

**Unique publishes separate guides to Challenging Behaviour and Sleep**

## Puberty

There is limited information available about puberty in children with KMT2B-related dystonia. We do know that some appeared to go through puberty early. Some families of children with chromosome disorders and behavioural or learning difficulties can be particularly concerned about their daughter's ability to cope with menstruation, and for some, discussing menstrual regulation options with a paediatrician may be beneficial.

## Adulthood

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

Adults with KMT2B-related dystonia 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. Some 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.

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# Medical concerns

The following medical concerns have been found in children with KMT2B-related dystonia. They are not found in all children so not all children with KMT2B-related dystonia will be affected.

## Hands and feet

While KMT2B-related dystonia is not linked to any congenital (apparent at birth) structural differences in hands and feet, anomalies of toe and foot position can occur as a result of the lower limb dystonia, which can lead to further difficulty walking. Over time, chronic dystonia can lead to permanent tightening of muscles or ligaments (fixed contracture), meaning that the foot or ankle can become "stuck" in a position with a limited range of motion. Some children are only mildly affected and may not require treatment. Others may benefit from massage, orthotics and physiotherapy. Treatment is tailored to the individual child, and in some cases surgical correction will best enhance eventual mobility.

## Eyes and sight

Problems with eyes and vision are uncommon in children with KMT2B-related dystonia but sometimes occur due to uncontrolled movement of the muscles controlling the eyes (nystagmus). This can also cause a squint (strabismus).

## Hearing

Some children have a hearing impairment, but hearing is unaffected in most children. This hearing loss is usually progressive and sensorineural, where there are problems with the inner ear, sometimes with the cochlea or auditory nerve (the nerve that sends signals to the brain about sound). Hearing tests at birth are unlikely to detect this, as when it does develop it usually happens later in childhood or adulthood.

Many types of hearing loss can be managed by using hearing aids. As children are at risk of speech delay, parental concerns should be acted on early and home- or school-based therapy provided.

**Unique publishes a separate guide to Hearing**

## Brain

A few children have a structural brain anomaly, which can be detected by MRI (magnetic resonance imaging) or a CT (computerised tomography) scan of their brain. The changes seen vary but include subtle and symmetrical dark streaks or shadows (hypointense lateral streaks) in a region of the brain called the external globus pallidus. Very rarely, an underdeveloped pituitary gland can also be observed.

## Seizures

Very rarely, children with KMT2B-related dystonia 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. More than one type of seizure may be present in the same individual.

The type of seizures that have been seen in individuals with KMT2B-related dystonia are 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).

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 the majority of cases, they self-resolve or resolve with medical treatment. There is no preference for a particular seizure medication for children with KMT2B-related dystonia. 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.

## Skin

Some children with KMT2B-related dystonia have skin (dermatological) conditions such as dry or scaly (ichthyotic) skin, and acne, which may be more pronounced than in the general population. These skin changes are not harmful but can be noticeable and may require supportive care. This could include the use of moisturisers (emollients) and other treatments, such as specific topical treatments to help relieve discomfort and keep the skin healthy. Some children have sparse or absent eyelashes, sparse hair growth (hypotrichosis) or, conversely, increased hair growth (hypertrichosis).

> ***“Since about five years old, he has struggled with eczema.”***

## How common is KMT2B-related dystonia?

KMT2B-related dystonia is extremely rare. Currently (2025) about 258 individuals with a KMT2B gene variant have been reported in the medical literature, but more are known to have been diagnosed. It is expected that more people will be diagnosed with this condition as awareness increases and genetic testing becomes more routine.

## 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 KMT2B gene then a child will have KMT2B-related dystonia. In almost all people identified so far (2025) with KMT2B-related dystonia, 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 KMT2B-related dystonia 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 KMT2B-related dystonia so far (2025) the genetic alteration has been found to be de novo (dn), which means neither parent was found to have the same KMT2B gene change as their child, and neither parent was found to have a chromosomal rearrangement that might have resulted in a KMT2B deletion in their child. Therefore, the chance of having another child with KMT2B-related dystonia 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.

**Unique publishes a short general guide to Mosaicism**

In families where the KMT2B variant has been inherited from a parent, the possibility of having another child - either a girl or a boy - with KMT2B-related dystonia rises to 50% (I 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 KMT2B 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 KMT2B-related dystonia.

**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 KMT2B-related dystonia be cured?

There is no cure for KMT2B-related dystonia 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 KMT2B-related dystonia have been published (2025). The following suggestions have been provided by clinicians, who have personal experience of managing/treating individuals with KMT2B-related dystonia, to improve quality of life and reduce complications.

Children and adults with KMT2B-related dystonia 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, physiology, endocrinology, ophthalmology, and audiology.

## Immediately following diagnosis

When not carried out as part of the diagnostic process, an evaluation of the features of KMT2B-related dystonia 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 KMT2B-related dystonia are present and how severe they are.

## Supportive care

Children with KMT2B-related dystonia 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 KMT2B-related dystonia 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.

**Audiologist** – a health care professional who diagnoses, treats and helps manage a condition that involves hearing or balance.

**Occupational therapist (OT)** – a health care 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.

**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, an individualised 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 KMT2B-related dystonia but may include:

**Physiotherapy** to help maintain mobility and strength, which usually includes tailored stretching and strength exercises to build strength and flexibility, and balance and coordination training to improve posture, walking patterns (gait) and reduce the risk of falls.

**Occupational therapy** for fine motor skills, social participation and developmental support which can include exercises and adaptive tools to help develop self-care skills such as feeding, writing, grooming and dressing.

**Speech therapy** can help combat speech and language delay by improving articulation, breath control and volume. Augmentative and alternative communication (AAC) aids (e.g., communication boards, speech-generating devices) may also be considered to support effective communication and social participation.

**Behavioural therapy** for intellectual disabilities and autism, ADHD, anxiety or other behaviours, which can include Applied Behaviour Analysis (ABA) to teach new skills and reduce challenging behaviours.

**Medications** may be prescribed, such as anticholinergics to alleviate dystonia; however, the effectiveness of these medications is variable. If your child experiences seizures, antiepileptic medication may also be prescribed to treat this. Similarly, ADHD medications and antidepressants may be considered if your child experiences behavioural or psychiatric difficulties.

**Surgery** such as bilateral globus pallidus pars interna deep brain stimulation (GPi-DBS) can be performed to alleviate dystonia and improve mobility. This procedure involves the surgical implantation of electrodes into a region of the brain called the globus pallidus pars interna, connected to a small generator implanted in the chest. This generator sends painless electrical signals to the brain to help control motor symptoms.

## 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:

- Regular Musculoskeletal review
- Neurologic review (potentially including MRI and EEG, if indicated by seizures)
- Regular speech, language and feeding review
- Check-ups for psychological/behavioural difficulties
- Consider an ophthalmologic review
- Consider a dermatological assessment

# Research into new treatments for KMT2B-related dystonia

The genetic change causing KMT2B-related dystonia affects the 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 KMT2B-related dystonia, like autism, is ongoing. In addition, although KMT2B-related dystonia is a relatively rare condition, the KMT2B gene is the subject of a lot of research. One avenue of research is to identify specific regions within the KMT2B gene where variants causing KMT2B-related dystonia are more common. By linking these hotspots to specific symptoms and features, this can help to predict the impact of certain variants prior to development and allow suitable support to be arranged as early as possible.

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

## Families say ...

**I feel it. It’s very important to have a good support team surrounding any child with KMT2B. We call our team of doctors, specialist and teachers “Team (child’s name)”.**

**“Being the mother of a child with this rare condition is challenging at times, but rewarding all the same. Everyone believes that their child is special and wonderfully unique, and I can say without a doubt that mine is, and he has a diagnosis to prove it.”**

## Sources

The information in this booklet is drawn from the published medical literature and information from Unique members. In 2025, Unique had 11 members with KMT2B-related dystonia. 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/**).

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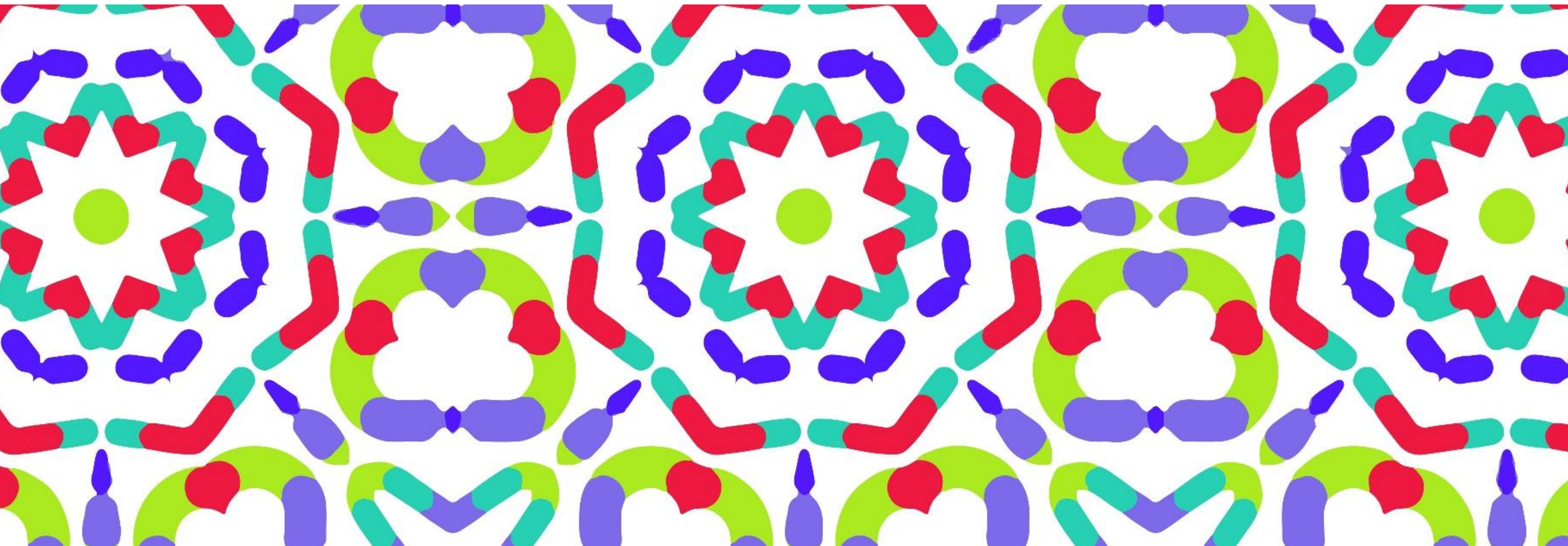
Note: an asterisk indicates articles which are “open access” and available to everyone at **https://pubmed.ncbi.nlm.nih.gov**



# 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

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

Become a member

## Please help us to help you!

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation:

Donate via our website

## KMT2B specific websites and Facebook groups:

- KMT2B Dystonia Foundation
- KMT2B Community Facebook page

This guide was written by Dr Adam Hodgson, Duncan Baker, Hollie Weedon, William Thompson, 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 Prof Meena Balasubramanian, Professor of Medical Genetics & Honorary Consultant Clinical Geneticist | University of Sheffield, Clinical Director of Research | Sheffield Children's NHS Foundation Trust, UK.

Version 1 (AP)

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Registered in England and Wales

Charity Number 1110661
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