CONDITION

Infantile myoclonic encephalopathy

A rare immune-related brain condition in young children that causes chaotic eye movements, jerking, poor balance, and behaviour or sleep changes.

WHAT IT IS
Rare immune-related brain condition affecting movement and development

BODY SYSTEM
Brain and nervous system

MAIN SYMPTOMS
Chaotic eye movements, jerking, poor balance, irritability

WHAT CAUSES IT
Immune reaction, sometimes linked to neuroblastoma or infection

HOW IT IS DIAGNOSED
Characteristic symptoms plus tests for neuroblastoma and mimics

MAIN TREATMENT
Immune therapy, tumour treatment, and developmental rehabilitation

CAN IT BE CURED?
Sometimes - immune treatment achieves lasting remission

IS IT AN EMERGENCY?
Sometimes - loss of consciousness or breathing difficulty

LONG-TERM OUTLOOK
Many improve, but learning or behavioural effects may persist

Overview

Infantile myoclonic encephalopathy is a rare condition in which the immune system mistakenly causes inflammation in the brain. In current medical literature, it is generally discussed as childhood opsoclonus-myoclonus-ataxia syndrome.

"Opsoclonus" means rapid, involuntary eye movements in different directions. "Myoclonus" means brief, shock-like muscle jerks. "Ataxia" means difficulty controlling balance and coordinated movement. "Encephalopathy" describes a change in how the brain is working.

The condition usually begins in a baby or young child. Despite the word "infantile," it can become noticeable during the toddler years. Symptoms may develop suddenly or build over several days or weeks.

In some children, the immune reaction is linked to neuroblastoma, a tumour that develops from immature nerve cells. In others, symptoms follow an infection or no clear trigger is found.

The abnormal movements can improve with treatment. However, the condition may also affect sleep, behaviour, speech, attention, learning, and emotional regulation. Early specialist assessment matters because treatment aims to control brain inflammation and identify any underlying tumour.

Symptoms

The pattern of symptoms is more important than any one symptom by itself. Not every child has all features at the start.

Common symptoms include:

The eye movements may be intermittent at first. They can be mistaken for unusual blinking, eye rolling, or a vision problem. Movement difficulties may initially look like ordinary clumsiness.

Myoclonus is not always an epileptic seizure. Children with this condition can also have seizures, but the brief jerks often come from a different type of abnormal brain activity. Testing may be needed to tell the difference.

Symptoms can fluctuate. They may become more noticeable when a child is tired, unwell, excited, or trying to make a controlled movement.

- Rapid, chaotic eye movements that continue when the child looks in different directions
- Brief jerks involving the body, arms, legs, face, or head
- Shaking or trembling during movement
- Poor balance, frequent falls, or loss of the ability to sit or walk steadily
- New irritability, distress, or sudden changes in behaviour
- Severe sleep disruption
- Loss of words or other previously learned skills
- Reduced attention or difficulty engaging in normal play

Causes and risk factors

Infantile myoclonic encephalopathy is thought to be immune-mediated. This means the

immune system, which normally protects the body from infection, mistakenly reacts against parts of the nervous system.

In some children, the reaction is associated with neuroblastoma. The immune system may recognise proteins in the tumour and then react with similar proteins in the brain. These tumours can be small and may not cause a visible lump or other obvious cancer symptoms.

Other children develop the condition after an infection. This does not mean that the infection directly entered and damaged the brain. It may have triggered an abnormal immune response.

Sometimes no infection, tumour, or other trigger is identified. The absence of a known trigger does not rule out the condition.

There are no established lifestyle-related risk factors. It is not caused by parenting, food, vaccination choices, or something a child did. It is not contagious and is not usually considered an inherited condition.

Diagnosis

There is no single blood test or scan that confirms infantile myoclonic encephalopathy. Diagnosis is based on the combination of characteristic symptoms and tests that rule out other causes.

A specialist will observe the child's eye movements, coordination, muscle jerks, behaviour, and development. Videos recorded at home can be useful because symptoms may come and go.

Testing may include:

A normal brain MRI does not exclude the condition. Urine tumour-marker tests can also be normal when a neuroblastoma is present, so imaging remains important.

If the first tumour search is negative, the specialist team may recommend further or repeated testing. This is planned according to the child's symptoms and initial results.

Other possible causes of unusual eye movements, jerking, and loss of coordination include epilepsy, infection of the nervous system, toxic exposure, metabolic conditions, inherited movement conditions, and structural problems in the brain.

- MRI scans of the brain to look for other causes of abnormal movement
- MRI or other imaging of the neck, chest, abdomen, and pelvis to search for neuroblastoma
- A specialised nuclear medicine scan, such as an MIBG scan, to detect certain neuroblastoma cells
- Urine tests for chemicals made by some neuroblastomas

- Blood tests for infection, inflammation, and metabolic conditions
- A lumbar puncture to examine the fluid surrounding the brain and spinal cord
- An EEG, which records electrical brain activity, when seizures are suspected

Treatment and management

Treatment has two main aims: reducing the harmful immune response and treating an underlying tumour if one is found.

Immune treatment may include corticosteroids or adrenocorticotropic hormone, often called ACTH. Intravenous immunoglobulin, or IVIG, may also be used. IVIG contains purified antibodies collected from donated blood plasma and can modify immune activity.

Some children need additional immune treatment. Rituximab, for example, reduces a type of immune cell called a B cell. Other immune-suppressing medicines may be considered when symptoms continue or return. These treatments require specialist monitoring because they can affect resistance to infection and have other side effects.

If neuroblastoma is found, treatment is coordinated by a paediatric oncology team. Surgery may be used to remove the tumour. Chemotherapy or other cancer treatment may be needed according to the tumour's features. Removing the tumour is important, but neurological symptoms may not disappear immediately because the immune reaction can continue.

Recovery support is also part of treatment. It may involve:

Treatment plans are adjusted over time. Immune medicines are generally reduced carefully rather than stopped suddenly, because symptoms can return during or after treatment changes.

- Physiotherapy for balance, strength, and movement
- Occupational therapy for play, daily activities, and coordination
- Speech and language therapy
- Help with sleep and behaviour
- Developmental and neuropsychological assessment
- Support at nursery or school

Complications

Possible long-term effects involve more than movement. Some children continue to have difficulties with speech, learning, attention, impulse control, sleep, mood, or social interaction after the eye movements and jerking have improved.

Motor complications can include persistent tremor, poor coordination, an unsteady walk, or difficulty with handwriting and other precise movements.

Symptoms can relapse, meaning they return after improving. Relapses may occur during an infection or while immune treatment is being reduced. A relapse does not necessarily mean that a tumour has returned, but it needs medical review.

Treatment itself can cause complications. Corticosteroids like prednisone or hydrocortisone may affect mood, sleep, blood pressure, growth, bones, or resistance to infection. Other immune treatments have their own risks. The care team will arrange monitoring based on the treatments being used.

A neuroblastoma has separate possible complications and follow-up needs. These are determined by its location, biological features, and whether it has spread.

Prevention

Infantile myoclonic encephalopathy cannot currently be reliably prevented. There is no established screening test for healthy children that predicts who will develop the immune reaction.

Ordinary childhood infections cannot always be avoided, and a child's illness is not evidence that someone failed to prevent the condition. Neuroblastoma is also not known to result from routine family or lifestyle choices.

Prompt assessment does not prevent the condition from starting. It can, however, help the child receive appropriate testing and treatment before symptoms continue for longer.

For a child already diagnosed, following the agreed treatment and follow-up plan may help identify relapses, developmental needs, or treatment side effects early. This is ongoing management rather than prevention.

Living with it and what to expect

The course is different for each child. Symptoms may be most intense near the beginning, followed by gradual improvement with immune treatment and rehabilitation. Treatment and follow-up often continue for months or years.

Eye movements, jerking, and balance may improve before sleep, behaviour, language, or attention. A child who appears physically steadier may therefore still need developmental or educational support.

Some children recover with few lasting effects. Others have ongoing learning, behavioural, emotional, or movement difficulties. These may become clearer when school tasks become more complex, even if early development appeared to recover well.

Relapses are possible. Families are often given guidance about which returning symptoms to record and report. A short video may help the specialist compare new movements with earlier symptoms.

Follow-up can involve neurology, oncology when a tumour was found, rehabilitation services, developmental specialists, and the child's school. Reviews may assess movement, sleep, speech, learning, attention, emotional health, and treatment effects.

Many children take part in family life, play, and education, although some need long-term therapy or classroom adjustments. Regular reassessment helps support needs change as the child grows.

When to get medical help

Seek urgent medical assessment for a child who develops new chaotic eye movements, repeated unexplained jerks, sudden severe unsteadiness, loss of previously learned skills, or a marked change in behaviour and sleep. These symptoms have several possible causes and need proper evaluation.

For a child with an established diagnosis, contact the specialist team promptly if symptoms return, worsen during an infection, or increase while treatment is being reduced. New fever, unusual bruising, persistent vomiting, or other possible medicine side effects should also be reported.

Call emergency services if the child:

Routine follow-up is still important when symptoms are stable. Planned visits monitor development, learning, movement, possible tumour concerns, and the effects of immune treatment.

- Has trouble breathing or turns blue or grey
- Cannot be woken or is much less responsive than usual
- Has a seizure that does not stop promptly

- Has repeated seizures without recovering between them
- Develops sudden severe weakness or suffers a serious injury

Common questions

Is infantile myoclonic encephalopathy a type of epilepsy?

Not usually. Myoclonus means brief muscle jerks, but not every myoclonic jerk is a seizure. An EEG may be used to look for epileptic brain activity if the movements are unclear or seizures are suspected.

Does this condition always mean my child has neuroblastoma?

No. Neuroblastoma is found in some children, while others have a possible infection-related trigger or no identified cause. Because a tumour can be small and otherwise silent, a careful tumour search is an important part of diagnosis.

Will my child recover completely?

Some children make a strong recovery with few lasting problems. Others continue to have difficulties with learning, attention, behaviour, sleep, speech, or coordination. It is not possible to predict an individual child's outcome from the first symptoms alone.

Can the symptoms come back after treatment?

Yes. Symptoms may return during an infection or when immune treatment is reduced. Contact the child's specialist team if eye movements, jerking, unsteadiness, sleep problems, or major behaviour changes reappear.

Is it genetic or contagious?

It is not contagious and is not usually inherited. The condition is generally linked to an abnormal immune reaction rather than a gene passed directly from parent to child.

Why are cancer tests needed when the symptoms affect the brain?

Neuroblastoma can trigger an immune response that also affects the brain. The tumour itself does not usually need to be inside the brain to cause these symptoms. Finding and treating it is an important part of the child's overall care.

Sources

- Rossor T, et al. Diagnosis and Management of Opsoclonus-Myoclonus-Ataxia Syndrome in Children: An International Perspective. Neurology: Neuroimmunology & Neuroinflammation . 2022;9(3):e1153.
- National Institute of Neurological Disorders and Stroke. Opsoclonus Myoclonus.
- National Cancer Institute. Neuroblastoma Treatment (PDQ®)-Health Professional Version.
- Great Ormond Street Hospital for Children. Opsoclonus myoclonus syndrome.