MECP2 Duplication UK

Our Children

Tel: 07849322368 **www.mecp2duplicationuk.org.uk**
Registered Charity No. SC044781

MECP2 Duplication Committee

The dedicated committee of MECP2 Duplication UK are all volunteers who have a wealth of knowledge and experience as all are parents and grandparents with children affected by the condition.

Rhona Connelly
Chairperson

Tony Kelly
Treasurer

Rachel Lund
Secretary

Becky Breslin
Committee Member

Sharon Gordine
Committee Member

Darren Chapman
Committee Member

Helen Coles Harrington
Committee Member

The medical advisor for our charity is

Professor Jill Clayton-Smith MB ChB MD FRCP

Consultant Clinical Geneticist , Manchester Centre For Genomic Medicine
Honorary Professor in Medical Genetics, Central Manchester University Hospitals Foundation Trust

Who are we?

MECP2 Duplication UK is an independent UK Charity dedicated to improving the lives of those affected by MECP2 Duplication syndrome.

Our Aims:

- To provide support emotionally, practically and financially to families affected by MECP2 Duplication Syndrome, helping to reduce feelings of isolation and exclusion.
- To raise awareness of MECP2 Duplication Syndrome and work with other relevant organisations.
- To promote and support research into MECP2 Duplication Syndrome which could potentially lead to treatments which could enhance the lives of those affected with the syndrome.
- For our Charity to grow and develop for the long term benefits of the MECP2 Duplication Syndrome community.

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What is MECP2 duplication syndrome

The cells, which are the building blocks of our body, contain small structures called chromosomes. There are 46 chromosomes in total made up of 23 pairs. You inherit one of each pair from your mother and the second from your father. Because half of our chromosomes are from our mother and half are from our father we become a mix of both parents. One pair of chromosomes contains information which determines which sex you will be and this pair differs in men (XY) and women (XX). Chromosomes take the form of a long length of a chemical called deoxyribonucleic acid (DNA) This DNA can be split into smaller segments known as genes. Genes act as the blue print for our body, telling cells what proteins to make, and ultimately determining what features we have and how we will grow and develop.

MECP2 duplication syndrome comes about when an individual has an extra copy of the region of DNA on the X chromosome which contains the MECP2 gene. This means that the individual will have one more copy of the MECP2 gene than normal. The region of the X chromosome involved is termed Xq28. So this condition is also often referred to as Xq28 duplication syndrome. MECP2 is just one of the many genes that lie within the Xq28 segment of the X chromosome. Boys are far more likely to be affected by having an extra copy of the MECP2 gene due to an Xq28 duplication. This is because boys have only a single X chromosome, whereas girls have a second, normal X chromosome which can usually cancel out the effects of having a duplication on the other chromosome.There are however some rare occasions when girls may also be affected.

The area of DNA that is duplicated on the X chromosome can vary. Sometimes, other genes can also be duplicated along with the MECP2 gene. The number of genes which are duplicated can vary between one individual and the next. Overall, it is duplication of the MECP2 gene which appears to lead to most symptoms, though as a general rule, the more genes that are duplicated, the more severe the symptoms will be. A lot of research is currently focussed at looking into the other genes that can be duplicated along with MECP2 and trying to find out what effect these other genes have on the symptoms of the syndrome.

Common charateristics of MECP2 are:

Hypotonia: At birth or during the first weeks or months of life babies typically have low muscle tone. In most cases, low muscle tone persists in the face and trunk into adult life.

Increasing Limb Spasticity: Over time, the low muscle tone in the limbs is replaced by increase muscle tone, particularly in the legs. The progression from hypotonia (low muscle tone to hypertonia (increased muscle tone) may be seen in childhood but may not occur until adolescence or even adulthood.

Gastroesophageal Reflux: Reflux has been noted in about 80% of cases.

Significantly Delayed Developmental Milestones: Rolling over, sitting, and other motor milestones are significantly delayed. Some individuals never walk independently, others begin walking at 3-5 years of age or older. Those who do walk typically have a wide unsteady gait.

Severe or Profound Intellectual Disabilities: Severe or profound intellectual disabilities have been reported in almost all cases.

Limited Language: Language skills are impaired. However, most individuals are described as having better understanding and receptive language skills than expressive language. Thus, they understand more than they are able to communicate.

Developmental Regression: The loss of motor and language skills over time has been reported in many but not all individuals. Regression frequently occurs around the same time as the onset of seizures, but the relationship between these symptoms remains unclear at this time.

Autistic Behaviours: Some autistic behaviours, such as hand-flapping and spinning objects, and those who developed speech echolalia, have been observed among many individuals with MECP2 Duplication Syndrome.

Severe and Recurrent Infections: Infectious disease and pneumonia are major health challenges and the most frequently reported cause of death for individuals with MECP2 Duplication Syndrome. The underlying cause for this is not entirely clear, and may be multifactorial. Functional immune system weakness does appear to be present in many. Reflux, swallowing problems, axial hypotonia, and other factors may contribute to the frequency and severity of lower respiratory infections.

Seizures: Seizures are very common in MECP2 Duplication Syndrome. They are less frequent among younger children but increase in frequency by the second decade of life. Many different types of seizure have been reported. And individuals with MECP2 Duplication Syndrome may have seizures of several different types with the nature of the seizures changing over time.

Low Bone Density and Fractures: Fractures and low bone density have been reported frequently. These effects may be associated with diet and anticonvulsant therapy, combined with frequent seizure related falls.

Other common characteristics include drooling, the inability to regulate body temperature adequately, sleep disorders, and bruxism or teeth-grinding.

We don't yet know the whole spectrum of features of the MECP2 Duplication Syndrome. It is important to monitor developmental progress, neurological features (spasticity, seizures), the frequency and type of infections, gastrointestinal symptoms and "unusual" behaviours so that early interventions can be offered and appropriate therapies accessed.

Therapeutic Interventions

Speech and Language Therapy

The main goal is to improve communication. Could also include assessment of swallowing function and safety.

Music Therapy

Can be extremely beneficial in improving hand function. Can be used to promote calm and relaxation. Also of help in aiding communication skills, improving gross and fine motor skills and general motivation and emotional well-being.

Hydrotherapy

One of the most essential of the physical therapies for our children. It can reduce muscle soreness and relieves tension in the limbs, easing the movement as opposed to when on firm ground.

Occupational Therapy

Provides support to people whose health prevents them from doing the activities that matter to them. Children/young people should be fully assessed for each piece of equipment that they require to improve overall quality of life.

Physiotherapy

Physiotherapy is very important in ensuring good postural management. Physiotherapy input ensures and promotes mobility, function and quality of life .

Rebound Therapy

This is an intensive one to one technique that involves the therapeutic use of trampolines to provide opportunities for movement,exercise and recreation.

Support for Siblings

MECP2 Duplication UK have a support group for the siblings of children/young people who have MECP2 Duplication Syndrome who face challenges due to the complex needs of their brother or sister. Family life often has to revolve around the routine of feeds, medicines and hospital visits/stays. This is unavoidable but leaves less time and attention for the siblings, who will also have their own fears and emotions about the situation. The siblings support group gives brothers and sisters a chance to share feelings, experiences, photographs and stories.

Find us on

Days out and Short Breaks

Our families like to meet up regularly for outings and short breaks. By meeting up and providing face to face support, we can avoid exclusion and isolation and all the children and families get to know one another and have a lot of fun.

We hold Family Days for all our children/young people and their families.