MVA Society

Mosaic Variegated Aneuploidy (MVA) Syndrome

MVA syndrome is rare and its prevalence is unknown.

Contents

MVA Syndrome

If you are reading this information, then you and those around you are probably at the start of your journey with MVA. You will find information here to help you understand MVA. It might feel overwhelming at first, so take your time, write down any questions and come back to find out more when you're ready.

Being tested for or diagnosed with a rare genetic condition can feel isolating, but please know you are not alone. There are support groups who can help you to navigate this journey and connect you with other people and families living with MVA who will be with you every step of the way.

About Mosaic Variegated Aneuploidy (MVA)

Mosaic variegated aneuploidy (MVA) syndrome is a rare disorder in which some cells in the body have an abnormal number of chromosomes instead of the usual 46 chromosomes, a situation known as aneuploidy. Most commonly, cells have an extra chromosome, which is called trisomy, or are missing a chromosome, which is known as monosomy.

In MVA syndrome, some cells are aneuploid and others have the normal number of chromosomes, which is a phenomenon known as mosaicism. Typically, at least one-quarter of cells in affected individuals have an abnormal number of chromosomes. Because the additional or missing chromosomes vary among the abnormal cells, the aneuploidy is described as variegated.

MVA Society

The MVA Society is trying to build a community for MVA patients, families and healthcare professionals. Through this we hope to build up a database of patients and experiences. This in turn will help drive the research in better detection and treatment options.

The MVA Society aims to provide a range of resources to help patients and families affected by this condition. In addition, it aims to be a focal point – a safe environment to share experiences, challenges and advice. It also of course will be able to act as a focal point for medics, clinicians and researchers in this space.

MVA Symptoms

Every person is unique, and different people with the same condition can be affected differently. People are born with MVA, with most diagnosis made in childhood when the child presents with a rare cancer or growth condition.

MVA Commonly observed complications

Key: Low efficacy | Medium efficacy | High efficacy

	Current Treatment	Treatment Pipeline
 Cancer	Surgery and Chemotherapy	Cancer vaccines and targeted therapies via whole genome sequencing NHS initiative
 Short Stature	Nutritional support and growth hormones	NA
 Smaller Head	Surgery or use of a helmet	NA
 Structural Anomalies	Plastic Surgery	NA
 Heart Problems	Medication, Pacemaker or Surgery	Stem Cell Therapies
 Congenital brain malformation	Drain excess fluid	NA
 Seizures	Drugs or Surgery	Focused Treatment

MVA Treatment

Clinical management depends on the affected individual's specific needs. Confirmed cases of MVA syndrome could be offered on-going (3 monthly) surveillance for the pre-disposition to cancer, renal support and/or any other specific support deemed necessary for that individual. The range of symptoms possible are extremely varied – a patient by patient approach is needed as all cases are likely to differ.

CONTACTS

Email: info@mvasociety.org

Facebook:

LinkedIn:

WhatsApp: Whats App MVA Community Hub

Click on the links above to be directed to the relevant pages

USEFUL CONTACTS

If you're struggling and need to talk, at any time of day or night, there are free listening services available.

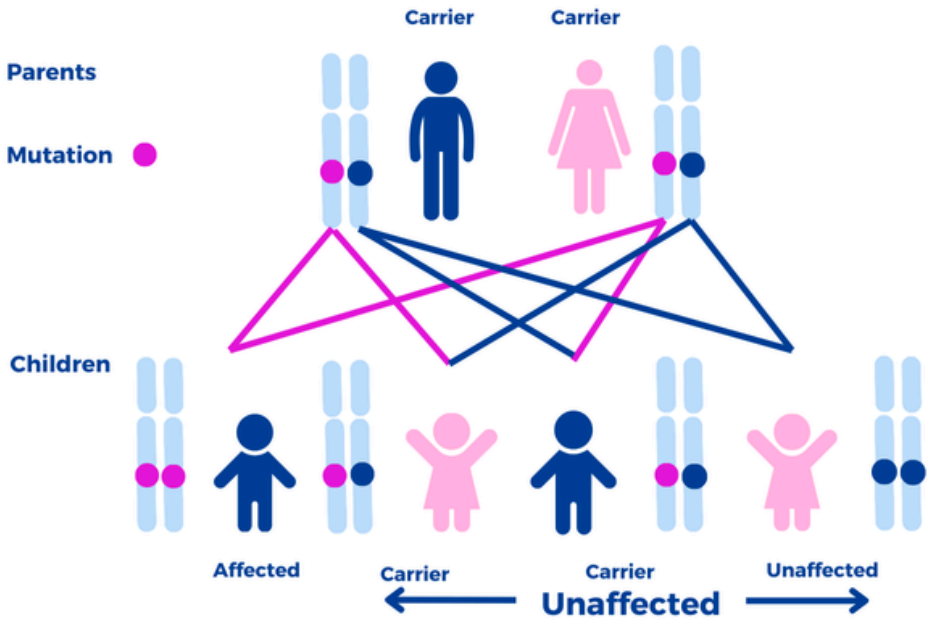
These services offer free and confidential support from trained volunteers and you can talk about anything that's troubling you, no matter how difficult.

- Call 116 123 (free from any phone) to talk to Samaritans, 24 hours a day, 365 days a year or email: jo@samaritans.org for a reply within 24 hours. You can also call the Samaritans Welsh Language Line on 0808 164 0123 (19:00-23:00 every day).
- Text 'SHOUT' to 85258 to contact the Shout Crisis Text Line, or text 'YM' if you're under 19
- If you're under 19, you can also call 0800 1111 to talk to Childline. The number will not appear on your phone bill.

MVA Causes

Genes are the instruction manual for a persons body. 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. Genetic mutations can be hereditary, when parents pass them down to their children, or they may occur randomly when cells are dividing. MVA is caused by genetic mutations in the following genes: BUB1B, CEP57 and TRIP13.

All types of MVA syndrome are inherited in an autosomal recessive pattern. This means the child will need to inherit the mutated copy of the gene from both the mother and the mutated copy from the father in order to inherit the condition. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene and one copy of a 'good' gene, but they typically do not show signs and symptoms of the condition. This means parents have a 1:4 chance of a child inheriting this condition. MVA is not anyone's fault, a copy of the mutated gene isn't caused by anyone's choice of life style or where they live, so its nothing the parents have done.
If parents are both found to have 1 copy of the faulty gene this doesn't mean all of their children would develop MVA but if one child is affected this doesn't mean future siblings would be.



IN PARTNERSHIP WITH

BOURNE PARTNERS

Tanner Pharma GROUP

APPROVED MEMBERSHIP & PARTNERING CHARITIES

Alice's ARC
CHILDREN'S CANCER CHARITY

MEMBER OF EURORDIS
RARE DISEASES EUROPE

GENE PEOPLE
PARTNERSHIP NETWORK

GENETIC ALLIANCE UK
Member

2024
NCVO MEMBER

PatientGroups

NORD®
2025 MEMBER
PLATINUM

Meet Our Team

Jonathan Bracey - Founder

Jonathan – JB – is the Founder of The MVA Society and George's Dad. All the Trustees supporting the MVA Society have a very personal connection with the Bracey family and of course George.

Professor Anil Dhawan

MBBS, MD, FRCPCH

Prof Dhawan is a Consultant in Paediatric Hepatology. He is helping the MVA Society to investigate treatments and improved healthcare outcomes for MVA patients

Dr Sharon Jheeta

Dr Jheeta, Consultant Paediatrician, was the first doctor to treat George. Now helping support the MVA Society with its ground-breaking work to find a cure for this ultra-rare condition.

Dr Harry Leitch

Dr Leitch is an Associate Professor and Honorary Consultant in Clinical Genetics and Genomic Medicine at Great Ormond Street Hospital. He has a longstanding interest in germline development (the process by which sperm and eggs are made) and will be using his expertise in paediatric rare disease genetics to help MVA understand more about this rare disease.

Dr Eleanor Seaby

Ellie is an NIHR Academic Clinical Fellow in Paediatric, completing a PhD in rare disease genomics. Now helping the MVA Society with their quest to better understand this ultra-rare condition and ultimately find a cure for it.

Scott Healy

Scott has dedicated his career to performance improvement in healthcare and life sciences working both within a multinational consulting firm (where he first met Jonathan as a client), and for the last decade as a co-founder of Akeso.

Kate Diston-Hunter

Kate has been a close friend of Jonathan's for nearly 20 years and has taken a keen personal interest in George and his recent MVA diagnosis.

Rob Keel

Rob is one of the Executive Vice President at Tanner Pharma Group. In addition, Rob has been a great friend of the Bracey family for the last 20 years and is one of George's godparents.