

Mosaic Variegated Aneuploidy (MVA) Syndrome

Fact Sheet

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What is 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.

MVA Means

Mosaic variegated aneuploidy (MVA) is a rare condition where ... when cells reproduce they don't always have the correct number of chromosomes - this is called, **aneuploidy** sometimes there are more chromosomes, sometimes less and sometimes the correct number - this is called, **variegated** it affects all cells in the body and there are normal cells mixed randomly with affected cells around the body - this is called, **mosaic**

Genes

There are **three** recognised genes that lead to **MVA**:

BUB1B – MVA Type 1

CEP57 – MVA Type 2

TRIP13 – MVA Type 3



There are, however, further genes that are suspected of causing MVA, including **CENATAC (Type 4)** and **MAD1L1 (Type 7)**

Conditions associated with MVA



Cancer



Structural Anomalies



Short Stature



Heart Problems



Smaller Head



Congenital brain malformation



Seizures