Open Peer Review on Qeios

Mosaic Variegated Aneuploidy Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Mosaic Variegated Aneuploidy Syndrome</u>. NCI Thesaurus. Code C128192.

An autosomal recessive genetic disorder caused by mutation(s) in the BUB1B gene, encoding mitotic checkpoint serine/threonine-protein kinase B. The condition is characterized by a predisposition to mitotic non-disjunction, resulting in a high percentage of aneuploid cells. The phenotype is variable and there is a predisposition to cancer.