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Congenital macrothrombocytopenia: A case report

Seneviratne TSP[1], Kumanan T[1], Sooriyakumar T[1]
[1]*Teaching Hospital Jaffna*

Case report: A 23 year old previously healthy sailor was evaluated for isolated thrombocytopenia. He did not have any bleeding manifestations. His physical examination was unremarkable. On direct questioning he admitted that his mother's sister living abroad having a haematological condition. On request the medical team received a document stating the diagnosis of his aunt as May- Hegglin Anomaly. On investigation his platelet count was ranging between 34 000- 70 000 during the past six months of follow up. The mean platelet volume was high.15.4fl (6.8-12fl).There was no other abnormality noted in the Full Blood Count(FBC).The other laboratory work up including hepatitis screen, renal, liver profile and coagulation profile were normal. The ultrasonography of abdomen was unremarkable. Blood picture revealed marked thrombocytopenia with Giant platelets. The bone marrow examination also supported the above findings. Giant platelets with isolated thrombocytopenia and a normal coagulation, platelet aggregation studies strongly suggested the diagnosis of a congenital large platelet disorder. FBC of the mother showed thrombocytopenia (platelet count 50 000- 100,000) and the blood picture showed giant platelets. FBC of the only sibling was normal.

Discussion: Considering the autosomal dominant nature of the disease; clinical presentation of the patient; pattern of inheritance; normal coagulation studies and the blood picture confirming congenital macrothrombocytopenia, May Hegglin Anomaly a MYH9 gene related disorder is the most probable diagnosis and immunohistochemistry is needed to confirm the presence of cytoplasmic NMMHC-IIA peptide.