

THROMBOCYTOPENIA AND MUCOSAL BLEEDING

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A 9 year old girl was seen on several occasions in different centers because of nose bleeds, multiple small echymosis and thrombocytopenia with the diagnosis of probable idiopathic thrombocytopenia purpura (ITP). When seen at hematology clinic of Ihsan Do ramacı Children's center in 1969 her periferial smear indicated thrombocytopenia and slightly stained platelets were notified (compatible with lack of granulation). Therefore the diagnosis

of 'gray platelet syndrome' (GPS) was made, (Figure 1) without any explanation for its pathogenesis.

The genetic basis of GPS was recently explained by Meral Günay Aygün *et. al.* (1) due to mutation in the neurobeacheri 2 (NBEAL 2), which encodes NBEAL 2 for protein that is inherited by chromosome 3p location.

This syndrome can not be diagnosed without examining preferal smear.

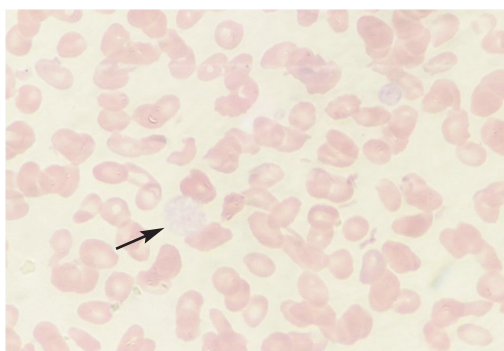


Figure 1: Arrow indicates gray platelet.

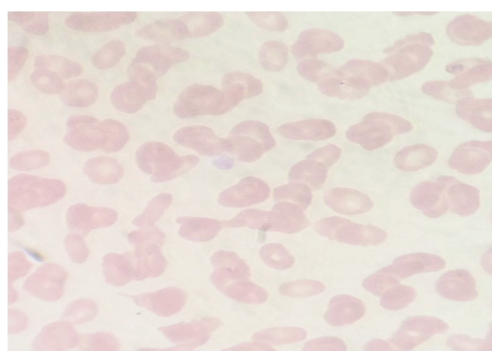


Figure 2: Normal platelets on the peripheral smear.

REFERENCE

1. Günay-Aygün M, Zivony-Elboun Y, Gumruk F, *et. al.* Gray platelet syndrome: natural history of a large patient cohort and locus assignment to chromosome 3p. *Blood* 2010;116:4990-5001.

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