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Images in Clinical Hematology

Chanarin-Dorfman Syndrome

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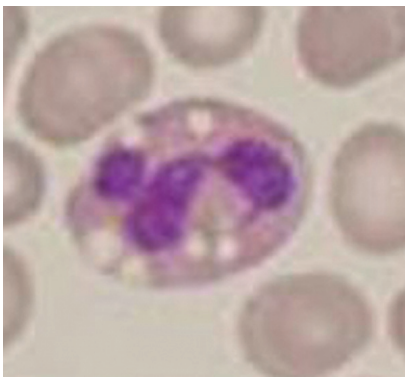

Figure 1. Large cytoplasmic lipid vacuoles in neutrophils suggesting Chanarin-Dorfman syndrome. Peripheral blood, stained with Leishman's stain: magnified 1000 x

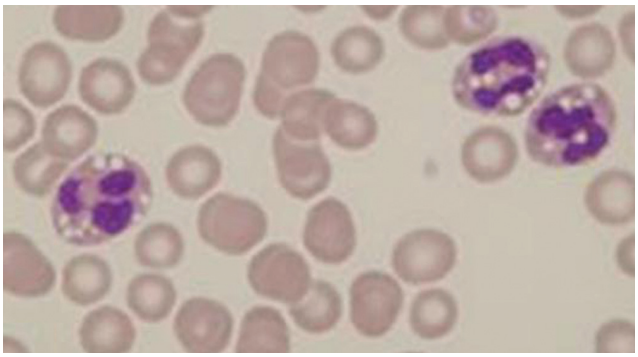

Figure 2. Prominent cytoplasmic vacuoles in neutrophils. Peripheral blood, stained with Leishman's stain: magnified 1000 x

Chanarin-Dorfman syndrome is a rare autosomal recessive disease characterized by the presence of intracellular lipid droplets in the cells of many different tissues, in particular in keratinocytes and granulocytes that may be associated with ichthyosis. Mutations in the gene that encodes the ABHD5 protein of the esterase/lipase/thioesterase subfamily have been identified as the main cause of the disorder. Extracutaneous manifestations are heterogeneous both in intensity and characteristics. Systemic involvement may include hepatosplenomegaly, double-sided cataracts, growth retardation, myopathy, ataxia and bilateral sensorineural hearing loss.(1) Since the first reports of the syndrome(1) only 30 patients have been described in the literature, mostly from Middle Eastern countries.(2) The diagnosis is confirmed by the presence of lipid droplets in granulocytes of peripheral blood.(3)

***Keywords**: Ichthyosis; Lipid metabolism, inborn errors; Syndrome; Granulocytes*

References

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Submitted: 6/22/2010
Accepted: 7/16/2010

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