

Images in haematology

An infant with hepatosplenomegaly and a cherry-red spot

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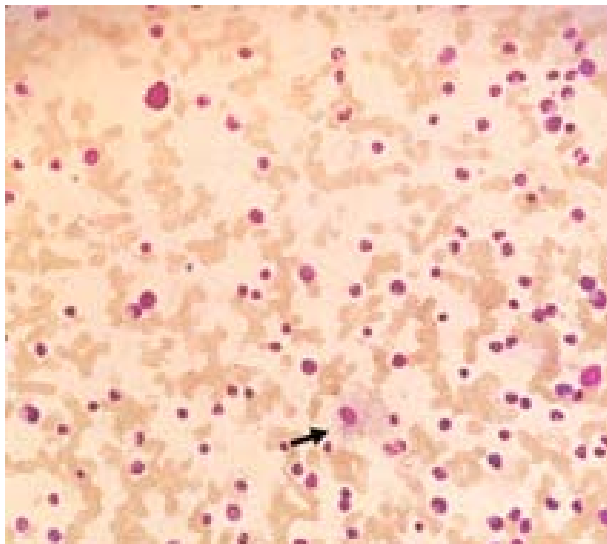


Figure 1. Bone marrow aspirate showing foamy macrophages Leishman stain (10x).

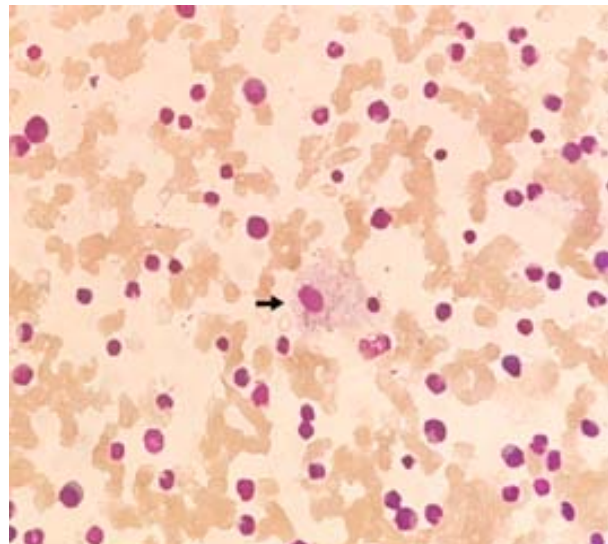


Figure 2. Bone marrow aspirate showing foamy macrophages Leishman stain (40x).

A 9-month-old baby presented with abdominal distension for 7 months duration. Examination revealed extra sacral Mongolian blue spot, hepatosplenomegaly (spleen>liver) with slight gross motor delay. Cherry red spot in the fundus was identified by ophthalmic evaluation. He had severe anaemia and bone marrow evidence of increased histiocytes with occasional foamy macrophages. The high possibility of a storage disorder was concerned. Molecular studies were not performed due to unaffordability. A specific enzyme assay was planned to be sent after 3 months of blood transfusion. The child was managed with supportive care, including blood transfusions and vitamin B12 and folate therapy. Miglustat (an oral inhibitor of glucosylceramide synthase) was planned to start as specific management.

Foamy macrophages are large cells with central nucleus, vacuolated and pale blue stained cytoplasm with Romanowsky stains. Presence of foamy macrophages in bone marrow is a characteristic finding in storage disorders such as Niemann-Pick disease, Fabry's disease, Wolman's disease, Tangier disease and

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metabolic disorders including hypercholesterolemia and hyperchylomicronaemia. Apart from that, they can also be increased in certain conditions resulting in damage to adipose tissue such as trauma, fat necrosis, bone marrow infarction, infection¹.

Therefore, significance of this bone marrow finding needs to be determined by clinical correlation.

References

1. Bain BJ, Clark DM, Lampert IA, Wilkins BS. Bone Marrow Pathology, 3rd edition, Blackwell science, 2006, Miscellaneous disorders.