Vacuolated Lymphocytes as a Clue for Diagnosis of Lysosomal Storage Disease like GM1 Gangliosidosis

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The patient was a 19-month-old female child with a prior diagnosis of GM1 gangliosidosis. She had developmental delay, visceromegaly and recurrent episodes of infection. During her follow up, she had anemia, thrombocytopenia and episodes of neutropenia. Her last complete blood counts were as follows: white blood cells = 15,970/ μ L, hemoglobin = 7.1 g/dL, and platelets = 66,000/ μ L. The peripheral blood smear showed prominent vacuolization of lymphocytes (Fig. 1).

GM1 gangliosidosis is a lysosomal storage disease with a prevalence of 1:100,000 to 1:200,000 live births. It is

caused by mutation of galactosidase beta 1 (GLB1) gene encoding the enzyme beta-galactosidase. The disease has a wide range of clinical presentation like coarse facies, hypertrophic gums, cherry red spot in macula, psychomotor delay and visceromegaly [1, 2].

The peripheral blood smear can be a good screening test. Vacuolated lymphocyte identification in an appropriate clinical setting is a key finding for suspecting an undiagnosed lysosomal storage disease [2]. Thus, it is a rapid and cost-effective method for screening lysosomal storage

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Fig. 1 Peripheral blood smear shows many vacuolated lymphocytes (Giemsa stain, magnification $\times 1000$)

disease and familiarity with this finding could be helpful in low source hematology labs.

Compliance with Ethical Standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethical Approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed Consent Informed consent was obtained from individual participant included in the study.

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