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Clinical Case Reports

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CLINICAL IMAGE

Vacuolated lymphocytes signifying a metabolic disorder in an infant with developmental delay

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Key Clinical Message

Metabolic disorders sometimes cause accumulation of metabolic byproducts which are manifested as cytoplasmic vacuoles in lymphocytes. We report the case of an infant with final diagnosis of GM1 gangliosidosis who initially presented with developmental delay and peripheral blood vacuolated lymphocytes. Blood film review is recommended in children suspicious for metabolic disorders.

Keywords

Metabolic disorder, peripheral blood smear, vacuolated lymphocyte

Case

Question

An 8-month-old male was presented with developmental delay and worsening respiratory failure. Peripheral blood smear examination revealed abnormal lymphocytes with numerous cytoplasmic vacuoles and eosinophils with unevenly distributed granules (Fig. 1). What do these findings suggest and what to do next?

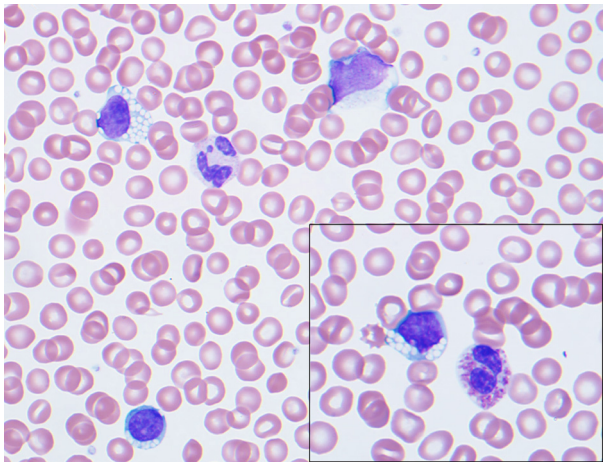

Figure 1. Lymphocytes with distinct cytoplasmic vacuoles and eosinophil with unevenly distributed granules (inset).

Discussion

Accumulation of metabolic byproducts due to metabolic disorder can present as vacuoles in lymphocytes. Therefore, identification of vacuolated lymphocytes in a pediatric patient with developmental delay should trigger more specific testings for metabolic disorders [1]. In addition, eosinophil granule abnormality seen in the image is commonly identified in GM1 gangliosidosis, a disease with lysosomal GM1 ganglioside accumulation due to β-galactosidase deficiency [2]. For the current case, metabolic enzyme test was ordered and showed decreased β-galactosidase activity. Sequencing of beta 1 galactosidase gene indicated mutations in exon 4 and intron 1, respectively. A diagnosis of GM1 gangliosidosis was rendered. Blood film review is recommended in children with signs and symptoms suspicious for metabolic storage disorders.

Conflict of Interest

None declared.

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References

1. Anderson, G., V. Smith, M. Malone, and N. J. Sebire. 2005. Blood film examination for vacuolated lymphocytes in the diagnosis of metabolic disorders; retrospective experience of more than 2500 cases from a single center. J. Clin. Pathol. 58:1305–1310.
2. Chevalier, C., and G. Detry. 2012. Vacuolated lymphocytes and abnormal eosinophils in GM1 gangliosidosis, type 1. Br. J. Haematol. 156:293.

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