Hereditary Elliptocytosis: A Rare Red Cell Membrane Disorder

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Received: 7 February 2018 / Accepted: 6 July 2018 / Published online: 2 August 2018
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Hereditary elliptocytosis (HE) occurs due to a defect in the red cell membrane proteins. It is a heterogeneous group of disorders mainly caused by defect in α -spectrin, β -spectrin, or protein 4.1. Various mutations have been identified in these genes which affect red cell cytoskeleton and membrane protein integrity [1, 2]. Clinical presentation in patients with HE is variable with most patients being asymptomatic. Approximately 10% of cases have moderate to severe anemia with jaundice and splenomegaly. We report two patients with this disorder. The first patient was a 30 years old lady born of a non consanguineous marriage. The second patient was a 4 years old boy who presented with fever and chills. The diagnosis of HE in both patients

was made incidentally when the peripheral blood smears examination showed 70–80% elliptocytes. Hematological investigations in both patients showed mild anemia with unconjugated hyperbilirubinemia. Eosin-5-maleimide test was borderline positive in patient 2. The diagnosis of hereditary elliptocytosis in both patients was confirmed by running red cell membrane proteins ghost on SDS PAGE. Gel electrophoresis was suggestive of spectrin defect in patient 1 and protein 4.2 defect in patient 2 [3] (Fig. 1).

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Fig. 1 **a** SDS gel electrophoresis (SDS-PAGE) of erythrocyte membranes showed a spectrin defect in case-1 (Lane-3) and protein 4.2 defect in cases 2 (Lane-1) and M indicates protein marker, **b** peripheral blood smear showed 70–80% elliptocytes

Compliance with Ethical Standards

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

Ethical Standards The article follows the ethical guideline as per NIIH Institutional ethical committee guidelines as laid under Helsinki's declaration 1976.

Informed Consent Informed consent was obtained from both the cases before blood collection and permission for publication of clinical and laboratory data.

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