CASE REPORT

Dehydrated hereditary stomatocytosis with transient perinatal ascites

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The case is reported of a mother and baby with dehydrated hereditary stomatocytosis and perinatal ascites, an autosomal dominant condition not previously reported in Britain. Recognition is important for the management of pregnancy and for avoidance of splenectomy which, if performed, can predispose the patient to fatal thromboembolic events.

A 29 year old primigravida was noted to have fetal non-immune hydrops from 19 weeks. Fetal pleural effusions, ascites, and skin oedema were observed, although the kidneys and heart appeared normal (fig 1). Fetal haemoglobin was 104 g/l and the karyotype 46XY. No infective cause was identified. Pleural effusions resolved but ascites persisted. Polyhydramnios developed and an amnioreduction was performed at 28 weeks. Fetal ascites was also drained (390 ml at 28 weeks and 360 ml at 31 weeks) to improve lung growth. Normal vaginal delivery at 32 weeks followed spontaneous onset of preterm labour.

At birth he was electively intubated and ventilated for 12 hours. The abdomen was soft and distended with poorly formed abdominal musculature (fig 2). Genitalia were normal, and ultrasound showed ascites with no other abnormality.

Over five weeks, 1155 ml straw coloured ascitic fluid containing variable lymphocyte numbers was removed (protein 20.8 g/l, glucose and electrolyte concentrations as plasma), after which there was no reaccumulation. Haemoglobin was 159 g/l at birth, mean corpuscular volume 95–97 fl, with anisocytosis, occasional fragments, and target cells. White cells and platelets were normal. He received three blood transfusions in the first two months of life; lowest haemoglobin concentration was 75 g/l.

The maternal grandmother mentioned that the baby’s mother had had ascites at birth. The mother’s microfilmed neonatal notes showed that she was born with unexplained ascites which resolved after drainage in the first 24 hours. She still has very poor abdominal muscle development and cannot sit up from the supine position. She had a normal karyotype and had no evidence of cytomegalovirus or toxoplasmosis. At 13 years of age, she presented with anaemia, jaundice, splenomegaly, gallstones, and persistent macrocytosis. A blood film showed reticulocytosis (14%), anisopoikilocytosis, schistocytes, and target cells (fig 3). Her haemolytic anaemia was extensively investigated, a striking finding being decreased osmotic fragility. Ham’s test was normal, as were screening tests for glucose-6-phosphate dehydrogenase deficiency and pyruvate kinase deficiency. There was no evidence of thalassaemia. Bone marrow investigations showed active erythropoiesis with minor dyserythropoietic changes. Recurrent abdominal pain led to cholecystectomy at 15 years. Studies showed no evidence of preferential splenic sequestration or destruction, so splenectomy was not performed. After extensive investigations, including electron microscopy, her well compensated mild haemolytic anaemia (typical haemoglobin concentration 100 g/l) had been labelled a “congenital dyserythropoietic anaemia” without any real conviction.

It seemed clear that mother and son had the same dominantly inherited and relatively benign problem of intrauterine ascites, possibly associated with the mother’s...
Table 1: Red cell indices, osmotic gradient ektacytometry variables, intracellular [Na⁺] and [K⁺] and isotopic K⁺ influx

| Hb (g/l) MCV (fl) MCHC (g/l) Retic (%) Statmcts O⁻₅₀ (mosmol/kg) Dlmax (arbitrary units) O⁺ (mosmol/kg) Na⁺ (mmol/l cells) K⁺ (mmol/l cells) Na⁺/K⁺ Pump Cotr Leak |
|---|---|---|---|---|---|---|---|---|---|---|---|
| Child 112 87.4 372 5.5 + 125 0.46 312 18.4 88.7 3.539 0.252 0.140 |
| Mother 99 103.6 363 2.7 + 147 0.47 333 11.48 102.2 3.539 0.252 0.140 |
| Father 158 88.9 355 1.7 – 161 0.41 346 8.93 99.1 3.539 0.252 0.140 |
| Normal 120– 85–96 300– 335–375 <2 143–163 0.41–0.53 335–375 5–11 85–105 1–2 0–1 0.05–0.10 |

Hb, Haemoglobin; MCV, mean corpuscular volume; MCHC, mean corpuscular haemoglobin concentration; Retic, reticulocytes; Statmcts, stomatocytes; O⁻₅₀, osmotic fragility; Dlmax, deformability index; O⁺, measure of cellular hydration; Cotr, Na⁺/K⁺ cotransport.

**DISCUSSION**

The “hereditary stomatocytoses” form a group of rare haemolytic conditions in which abnormal sodium and potassium handling by the erythrocyte membrane is a prominent feature. Dehydrated hereditary stomatocytosis is the most common single variant (OMIM 603528). Despite the name, stomatocytes may be very scarce or even absent and the term “hereditary xerocytosis” is often used for this variant. In most pedigrees, it is purely a red cell disease. In some families, as here, it forms part of a pleiotropic syndrome which extends beyond haematology. This syndrome can combine dehydrated hereditary stomatocytosis, perinatal oedema, and pseudohyperkalaemia (artificially high plasma K⁺) levels. Isotopic flux measurements for K⁺ show increased ouabain+bumetanide resistant (“leak”) K⁺ influx and increased ouabain sensitive “Na⁺/K⁺ pump” rates typical of a mild leaky red blood cell condition.

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**REFERENCES**