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Research Letter

Dehydrated hereditary stomatocytosis: Prenatal management of ascites and pleural effusions

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Dear Editor,

A 32-year-old woman gravida 4 para 3 was referred at 20 weeks of gestation (WG) to prenatal unit for voluminous fetal ascites. No fetal anemia was suspected. Antenatal tests (storage diseases, digestive enzymes, infections) and fetal karyotype (46, XY) were normal. A single puncture (172 mL) resolved ascites (lymphocytes 83%) two weeks later. A 3850 g baby was born at 39 WG without edema. Sometimes, the boy had abdominal pain, yellow complexion, no blood test was realized.

The patient was referred 3 years later at 22 WG for bilateral pleural fetal effusion and marked ascites (Fig. 1A). The same tests were normal (46, XY). An intrauterine peritoneo-amniotic shunt (majority lymphocytes) at 22.6 WG and 28 WG, a thoracic-amniotic shunt (23.6 WG) and amniotic drainage (1000 mL) were performed. Complete resorption of effusions was observed after 30 WG with drains in good position (Fig. 1B). Maternal investigations revealed a compensated hemolysis with significant reticulocytes (212 10^9/l), normal hemoglobin level (14.6 g/dl), increased mean corpuscular hemoglobin volume (MCHV) (99.2 flg/dl) and mean corpuscular hemoglobin concentrations (MCHC) (37 g/dl). The ektacytometry

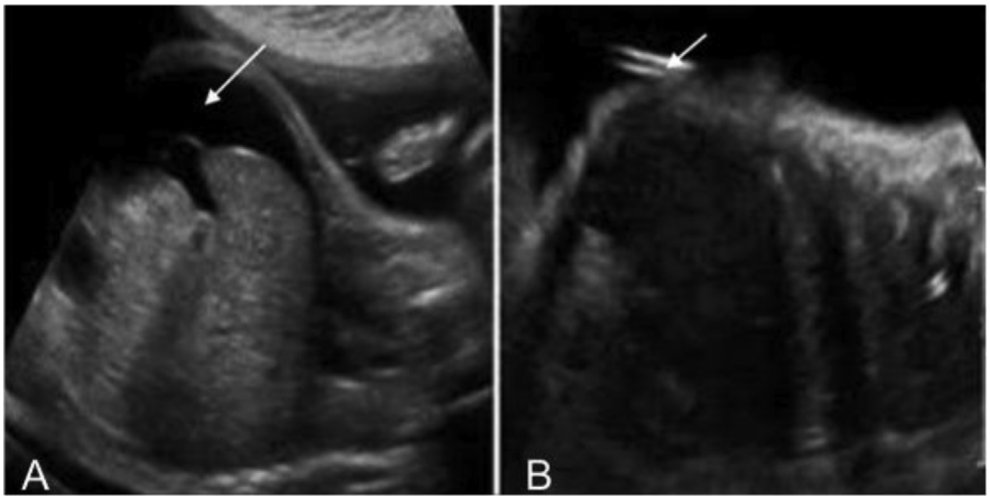

Fig. 1. Ultrasound scans during pregnancy. A) Longitudinal view shows ascites (white arrow). B) Longitudinal view shows peritoneo-amniotic shunt and ascites (white arrow).

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Table 1
Genetic heterogeneity of red cell stomatocyte disorders and other aetiologies of prenatal oedema.

Diseases	Red cell stomatocyte disorders					Other red cell diseases			Infections	Malformations
	Dehydrated hereditary stomatocytosis or xerocytosis	Lymphoedema hereditary type III (Fotiou)	Overhydrated hereditary stomatocytosis	Familial pseudohyperkalemia	Cryohydrocytosis	Hereditary spherocytosis	Elliptocytosis	Red cell immunization	Parvovirus B19 infection	Cardiac malformation
OMIM	#194380, #602754	#616843	#609153	#609153	#185020, #608885	#182900, #270970, #612653, #612690, #616649	#130600, #235370, #611804	–	–	–
Inheritance	AD	AR	AD	AD	AD	AD or AR	AD or AR	–	–	–
Genes	*PIEZO1, KCNN4*	*PIEZO1*	*RHAG*	*ABCB6*	*SLC4A1, SLC2A1*	*SPTA1, SPTB, ANK1, SLC4A1, EPB42*	*SPTA1, SPTB, EPB41, GYPC*	–	–	–
Phenotype	+	– or mild or asymptomatic	+	+	+	–	+	+	+	–
Hemolytic anemia										
Perinatal oedema	+/–	++	–	–	–	+	+	+	+	+/–
Fetal anemia	–	–	–	–	–	+	+	+	+	–
Splenomegaly	+	–	+	+	+	+	+	+	+	+
Hepatomegaly	+	–	+	+	+	+	–	+	+	+

+: sign present; – absent; AD: autosomal dominant inheritance; AR autosomal recessive inheritance.

showed leftward shift of the osmotic curve and for one of her sons. Genetic analyses in the family by direct sequencing after obtained informed consent led the identification of a heterozygous mutation (c.7367G>A; p.Arg2456His) in exon 51 of *PIEZO1* in the patient, his mother and two sibs. They presented all of them hemolytic anemia with splenomegaly, hematomegaly and cholelithiasis but no history of oedema.

A 2580 g boy born at 32 WG after a premature rupture of membranes showed moderate respiratory distress without intensive ventilation. The drains were removed the first day of life, without any paracentesis later. Normal hemoglobin (17.3 g/d), increased MCHV (95.5fl), MCHC (38.7 g/dl) and reticulocytes, and few stomatocytes on blood smears were noted. The boy was discharged after six weeks with minimal ascites resolved spontaneously few weeks later. The ektacytometry showed the same features. The child remains in good health after four years of pediatric follow-up with moderate defect of abdominal wall at clinical examination and surgery of bilateral ectopic testis at 1 year.

Xerocytosis, rare autosomal dominant-inherited disorder of the red cell permeability to monovalent cations, is genetically heterogenous. Mutations in *PIEZO1* gene and, recently in *KCNN4* gene have been identified in families including perinatal oedema and pseudohyperkalemia forms [1,2].

Perinatal oedema, with unclear pathophysiology, is a rare complication with variable severity sometimes lethal, in the same families. Five cases of fetal hydrops and seven with ascites and/or pleural effusions were reported between 14 and 32 WG [3–5]. Effusions often disappear spontaneously within weeks or months following the birth, sometimes before birth [5]. Mild fluid collections require no invasive treatment, only medical attention. In few cases, amniotic fluid and ascites were drained during pregnancy or after birth to avoid severe complications and fatal outcome. Xerocytosis is a not well-known stomatocyte disorder with can cause perinatal oedema. There are other several etiologies of perinatal oedema (other red cell stomatocyte disorders, red cell diseases, parvovirus B19 infection and congenital heart malformation). Heterogeneity of stomatocyte disorders and other etiologies of perinatal oedema are reported (Table 1).

Medical teams should be encouraged by favorable prognosis. It is difficult to predict if effusions disappear in prenatal or postnatal period. To our knowledge, it is the first case reported with thoracoamniotic and peritoneo-amniotic shunt. According to us, this procedure should avoid iterative punctures during pregnancy, improve lung growth and abdominal muscle development, prevent respiratory failure at birth and facilitate initial neonatal care.

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