

Xerocytosis With Concomitant Intrauterine Ascites: First Description and Therapeutic Approach

To the Editor:

Xerocytosis is a rare, autosomal dominant-inherited membrane defect of the erythrocytes with an increased osmotic resistance and a reduced intracellular potassium concentration. It was first described by Glader in 1974.¹ Affected individuals show mild anemia and reticulocytosis. About 20 cases have thus far been described in the world literature. In most cases, there is an autosomal-dominant hereditary transmission.²

CASE REPORT

A healthy primigravida at 25 weeks of gestation was referred to us because of fetal hydrops. Sonographic examination showed severe polyhydramnios and marked fetal ascites.

The husband and his mother and sister had confirmed xerocytosis. His sister had a slightly anemic child with postnatally diagnosed xerocytosis. The child showed intrauterine ascites that spontaneously resolved a few days postpartum. In our case, prenatal diagnosis of the xerocytosis was achieved by cordocentesis at 25 weeks of gestation: the red blood cell potassium concentration was low (78 mmol/L), osmotic resistance was increased (initial hemolysis at less than 0.3% NaCl, complete hemolysis could not be obtained), the fetal hemoglobin level was 9.7 g/dL, the hematocrit level was 0.27 L/L, the mean corpuscular hemoglobin concentration was 35.5 g/dL, the reticulocytes level was 250‰, and the total serum protein level was low (27 g/L). These prenatal findings were consistent with the diagnosis of xerocytosis. A puncture of 400 mL ascites showed only lymphocytes. The ascites reaccumulated within a few days.

Prenatal therapeutic management. We assumed that there may be a correlation between the xerocytosis and the fetal ascites, as suggested by the family history of the cousin with a similar association. We therefore attempted to treat the fetal ascites by a partial intrauterine exchange transfusion by cordocentesis. In two sessions, 10 mL of blood was withdrawn from the fetoplacental unit and replaced by 40 mL of packed red blood cells up to a hemoglobin concentration of 13.7 g/dL. The percentage of adult donor erythrocytes reached 50%. However, the fetal ascites did not disappear but

increased in the following 2 weeks. Subsequently, the fetal ascites was punctured several times in hopes of preventing a development of pulmonary hypoplasia due to the elevation of the diaphragm and to treat the overextension of the fetal abdominal wall, which can cause a prune belly syndrome.³

At 33 weeks of gestation, the mother developed preeclampsia, and the infant was delivered by elective cesarean section. Before the cesarean section 900 (!) mL of fetal ascites was removed by intrauterine puncture to facilitate initial neonatal care. A female neonate weighing 3,040 g (>95 percentile of weight per gestational age) was born with an Apgar score of 8/9 and 9 at 1.5 and 10 minutes postnatally, respectively; umbilical artery pH 7.35; and a hemoglobin concentration of 10.8 g/dL. She required mechanical ventilation for 7 days because of respiratory insufficiency, which improved after the application of surfactant. Postnatally, a partial exchange transfusion of 35 mL was performed, but there was no reduction in the ascites. The reappearing ascites required six punctures of about 300 mL each during the first 6 weeks of life. The abdominal wall was markedly distended (Figs 1 and 2), similar to that seen in prune belly syndrome.

Only minimal ascites was present 8 weeks postpartum, which resolved spontaneously at the age of 10 weeks. The child was discharged after 3 months. At 6 months, she was normal except for mild anemia with reticulocytosis.

DISCUSSION

This is the first report of two cases of a fetus and a neonate with xerocytosis and concomitant ascites. The mechanism for the development of ascites is unclear. The peritoneal epithelium may possibly show a membrane defect similar to the erythrocyte membrane defect, but it remains unclear why, postnatally, the ascites resolved spontaneously. The hypoproteinemia observed in the fetus was probably a result and not the cause of the ascites, because xerocytosis does not lead to hypoproteinemia.

Hydrops fetalis including ascites may result from severe fetal anemia, but usually does not appear unless the hemoglobin concentration is less than 4 to 5 g/dL.⁴ The highest hemoglobin concentra-

tion resulting in ascites so far described was 6.8 g/dL,⁵ which is markedly less than the 9.7 g/dL measured in our case.

In fact, the intrauterine and neonatal exchange transfusions with packed red blood cells did not have any effect on ascites formation. Obviously the mild anemia of our patient was not the cause of the ascites.

In our case, serial punctures of the ascites were performed to prevent lung hypoplasia and to reduce abdominal wall distension. Despite this management, the infant showed marked abdominal wall distension resembling prune belly syndrome. In case of a consequent pregnancy in this patient with marked fetal ascites, we suggest establishing an intrauterine peritoneo-amniotic shunt to prevent the over-extension of the abdominal wall.

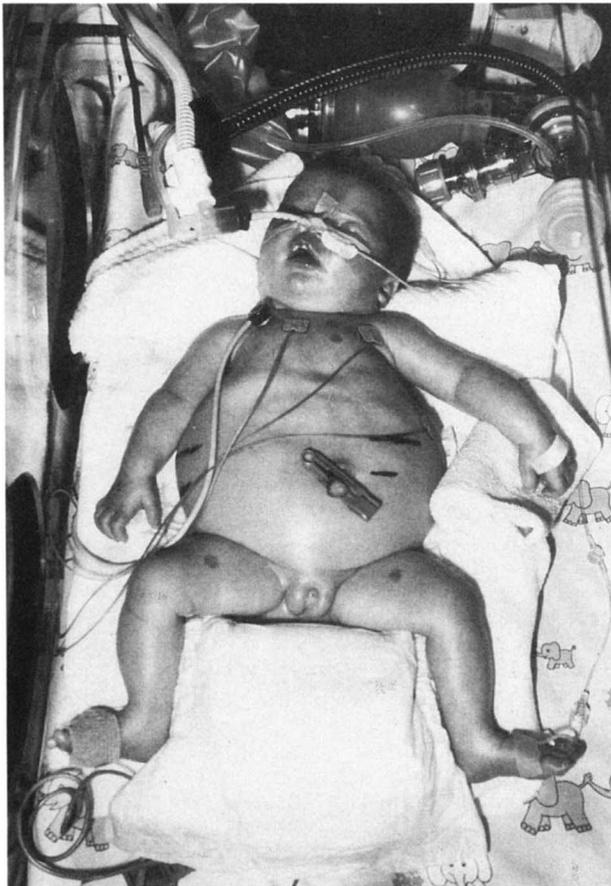


Fig 1. Neonate with xerocytosis 5 days after delivery.



Fig 2. Detail showing marked distension of the abdominal wall (prune belly) due to intrauterine ascites.

The anemia of the newborn due to the xerocytosis was mild and did not require any treatment.

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