

Hereditary Xerocytosis in Pregnancy: Case Report

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Case Presentation

A 33-year-old G2P1001 presented to MFM clinic at 12 weeks' gestation with known hereditary xerocytosis (HX).

History included:

- Mild macrocytic anemia
- Splenomegaly
- Strong family history (mother, two sisters)
- Diagnosed in utero after fetal ascites was identified

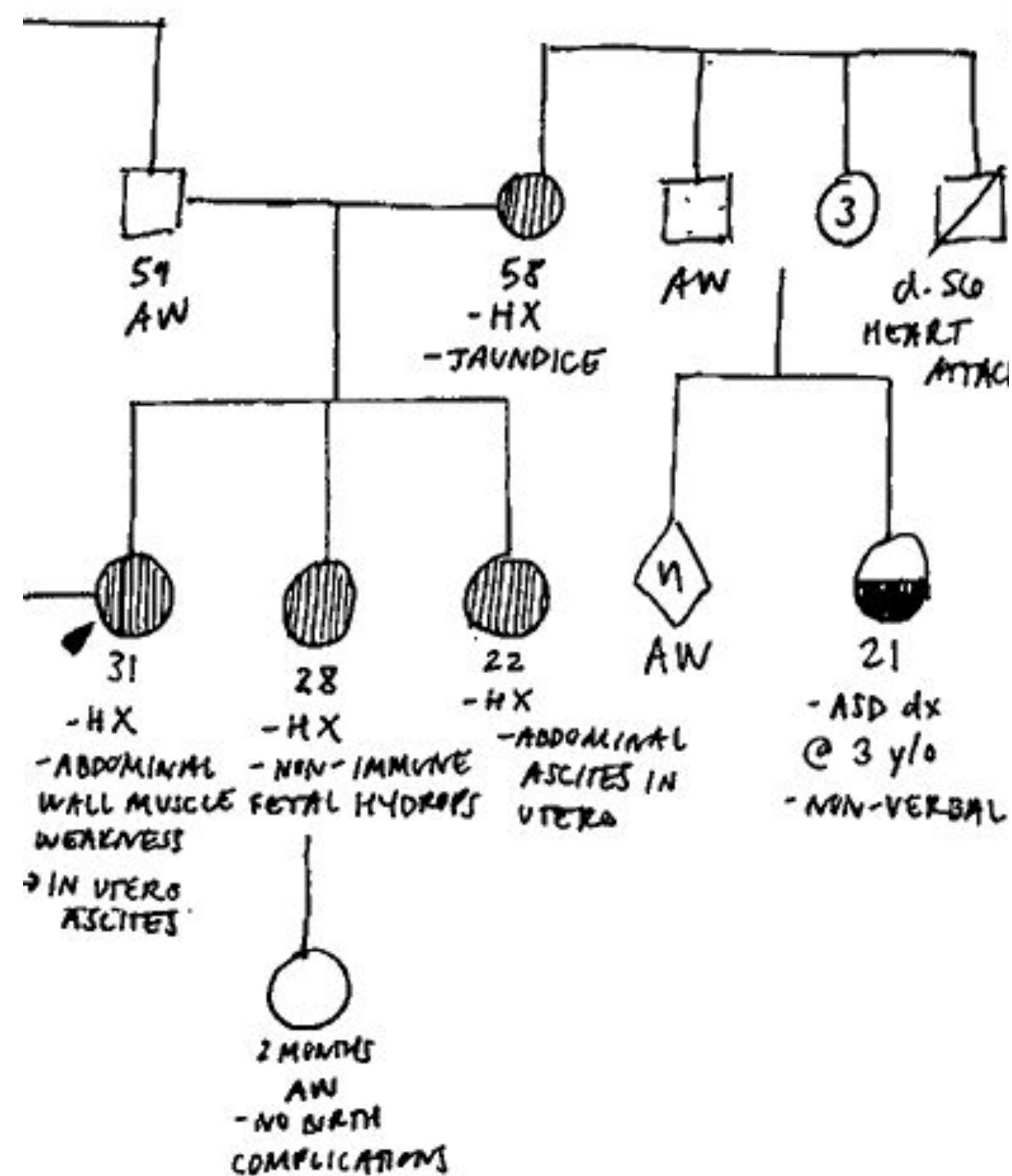


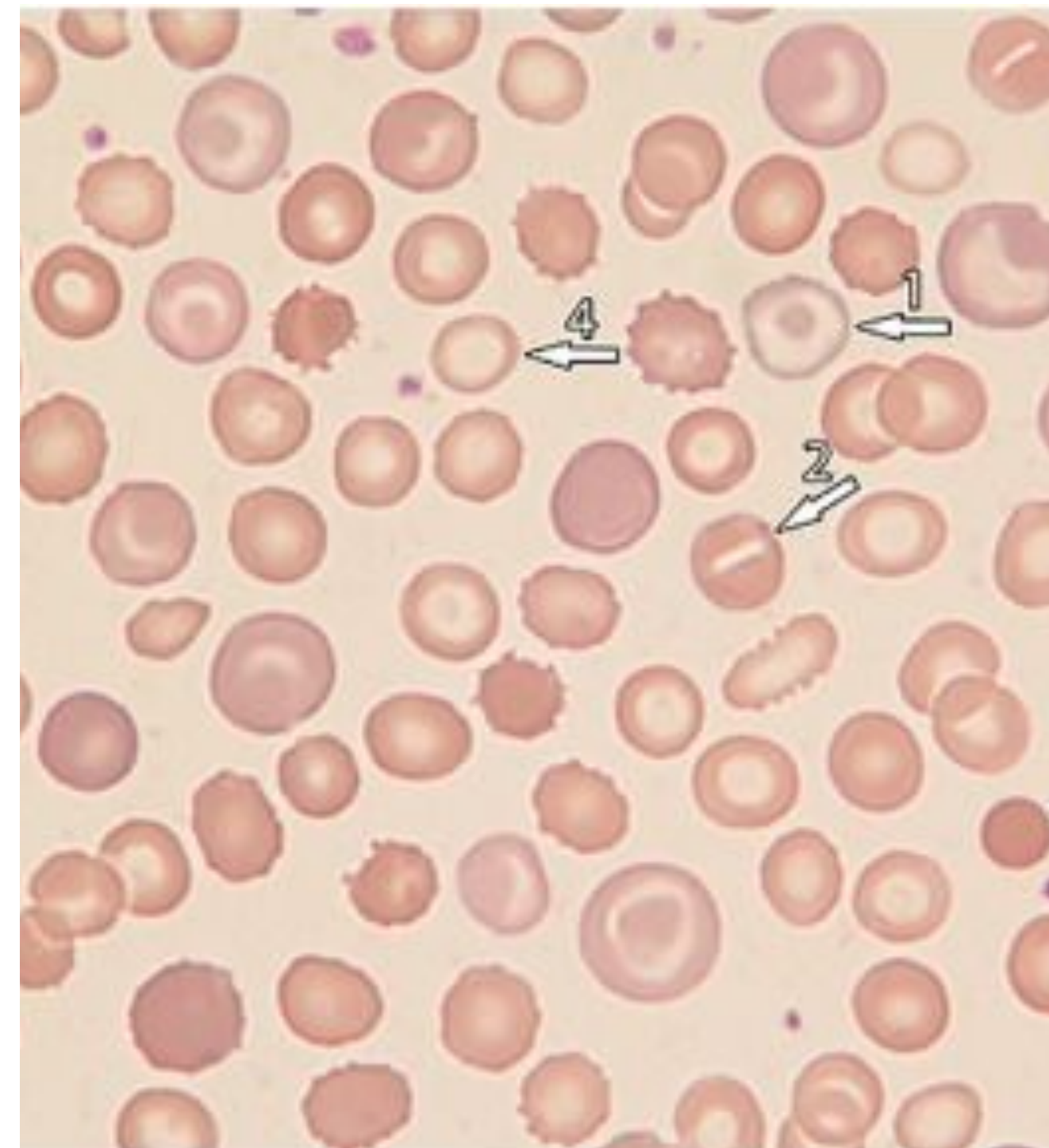
Figure showing ascites and hepatomegaly

Pregnancy Course

- **16 weeks:** Initiated serial MCA Doppler surveillance
- **20 weeks:** Anatomy scan showed fetal ascites & hepatosplenomegaly; MCA Doppler normal
- **Amniocentesis:** Identified PIEZO1 variant (c.7483_7488dup, p.Leu2495_Glu2496dup)
- **Fetal intervention:** 3 paracenteses to mitigate risk of lung hypoplasia
- **36 weeks:** Repeat cesarean delivery; neonate had mild anemia; ascites resolved postnatally

Discussion

Hereditary xerocytosis (HX) is a rare, autosomal dominant congenital hemolytic anemia characterized by abnormal loss of potassium and water, leading to dehydrated, rigid red cells (xerocytes) and a chronic, mild-to-moderate hemolytic anemia. It is caused by PIEZO1 mutations. Fetal ascites may be the first presenting sign, often in the setting of a strong family history. HX in pregnancy poses diagnostic and management challenges due to its rarity and the potential for severe fetal complications.



Cause

Most commonly caused by **gain-of-function mutations in the *PIEZO1* gene**, which encodes a mechanosensitive ion channel.

Key Clinical Features

- **Mild to moderate chronic hemolytic anemia**
- **Reticulocytosis**
- **Elevated MCHC** — a hallmark lab finding
- **Jaundice**
- **Splenomegaly**

