

Prenatal Sonographic Clues to Congenital Autosomal Recessive Sideroblastic Anemia: A Rare Diagnosis

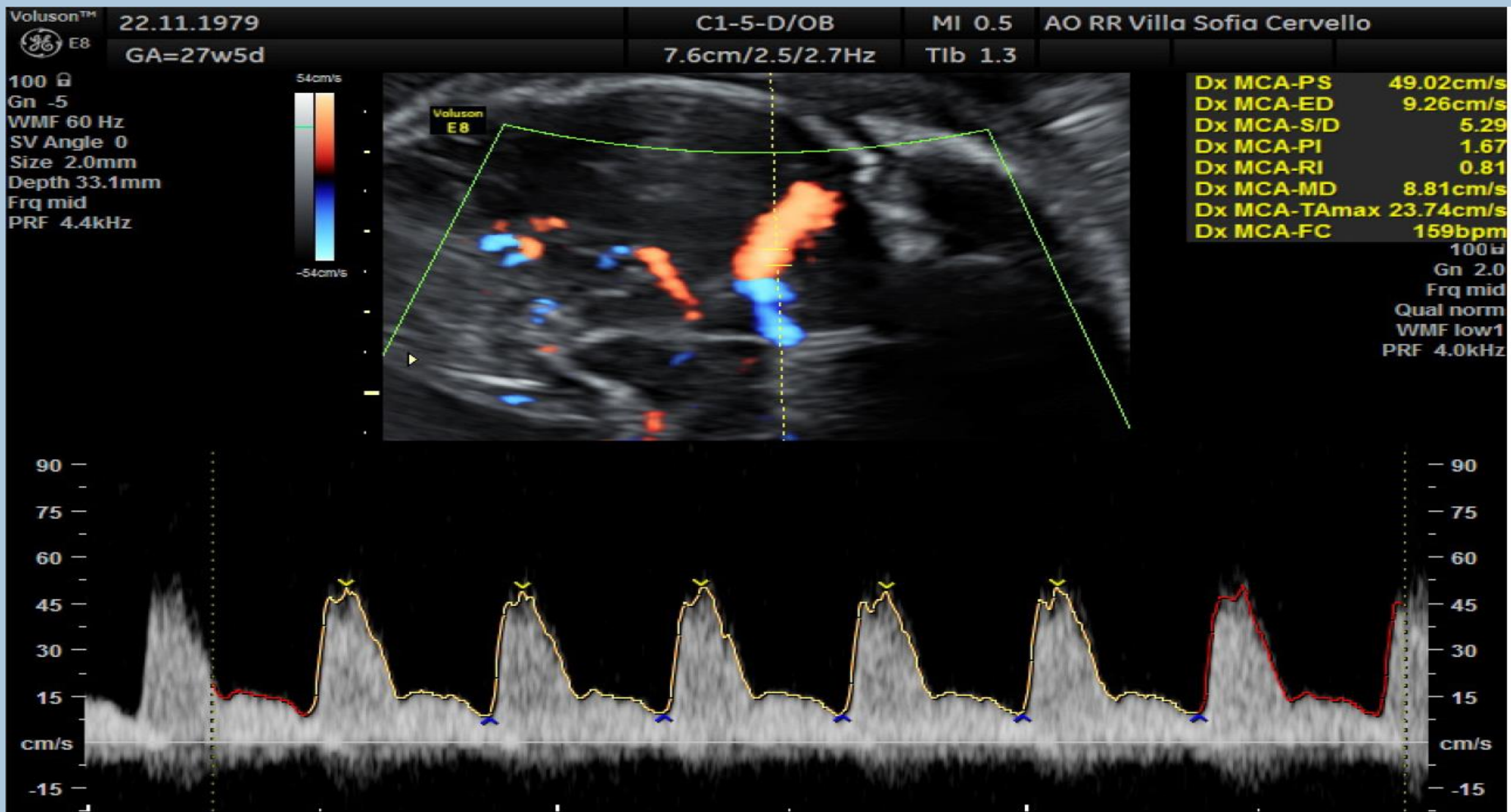


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Background

- ❑ A primigravida presents in third trimester with an ultrasound diagnosis of fetal ascites
- ❑ Further evaluation, including genetic testing, revealed autosomal recessive pyridoxine-resistant congenital sideroblastic anemia due to *SLC25A38* mutation
- ❑ Fetal ascites without alloimmunization on prenatal ultrasound should prompt evaluation for congenital anemia, including sideroblastic anemia.

❑ Less than 10% among congenital sideroblastic anemia.



Conclusion

- ❑ Congenital Autosomal recessive sideroblastic anemia may present as fetal ascites in the third trimester. Such cases require close follow-up, antenatal invasive testing if parents wish to know the genetic cause, and liaison with a geneticist and pediatric hematologist

Case- Report

- ❑ A 26-year-old healthy primigravida was referred at 29 weeks for **moderate fetal ascites**. Detailed ultrasound revealed **fetal hydrops, cardiomegaly, shortened long bones, and placental thickening**. Fetal echocardiography showed **dilated right heart chambers, enlarged pulmonary arteries, and pericardial effusion**.
- ❑ Whole exome sequencing (CVS at 30 weeks) confirmed **autosomal recessive, pyridoxine-refractory congenital sideroblastic anemia** due to **SLC25A38 mutation**.
- ❑ A **preterm male infant** (2.4 kg) was delivered at 34+4 weeks with **hydropic features, hypospadias, and anemia** (Hb 8 mg/dL). Despite transfusions, anemia persisted; peripheral smear showed **Pappenheimer bodies**.
- ❑ A detailed counselling of parents regarding disease, prognosis and treatment options was done by genetist, neonatologist and hematologist

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