

Diagnostic Challenges in Non-Immune Hydrops Fetalis: A Case of Trisomy 21 with Progressive Fluid Accumulation & Normal Foetal Echocardiography

Delia Marinceu¹, Elizabeth Pond¹, Uma Rajesh¹ 1. Fetal Medicine, Hull University Teaching Hospitals NHS Trust, Hull, England, United Kingdom.



shutterstock.com · 1587087325

INTRODUCTION

Hydrops fetalis is a severe foetal condition characterized by abnormal fluid accumulation in at least two foetal compartments. Non-immune hydrops fetalis (NIHF) accounts for up to 95% of cases, with a significant decline in immune hydrops due to prophylactic measures against red blood cell alloimmunization. NIHF has multiple aetiologies, including cardiovascular anomalies, chromosomal abnormalities, infections, hematologic disorders, and genetic syndromes. Chromosomal abnormalities contribute significantly, with studies indicating that approximately 19% of NIHF cases are associated with abnormal karyotypes. Despite its rarity—occurring in approximately 2.5 per 10,000 live births—NIHF carries a high perinatal mortality rate, reaching 35.1% in the neonatal period and increasing to 43.2% within the first year of life.

LEARNING POINTS

This case highlights that, in Down syndrome, severe progressive hydrops fetalis can be the sole radiological presentation. The diagnostic complexity is further compounded in the absence of genetic testing, which delays definitive prenatal diagnosis and prognostic counselling.

Additional considerations include whether the infant exhibited hepatosplenomegaly or peripheral blood film changes, as these findings could suggest a transient myeloproliferative disorder (TMD), a leukemoid response commonly reported in fetuses with Trisomy 21. Another plausible explanation could be an underlying congenital lymphatic disorder, which may have contributed to the hydrops and its prognosis. While TMD generally has a favourable long-term outcome with no recurrence or significant complications, approximately 20–30% of survivors develop leukaemia within the first three years of life.

Clinicians should maintain a high index of suspicion and consider chromosomal disorders in unexplained hydrops fetalis presentations, in order to ensure appropriate antenatal, plan for delivery and postnatal care counselling for the expecting parents. Multi-disciplinary care is crucial to deliver the best medical and supportive care in these challenging cases.

REFERENCES:

- Smrek JM, Baschat AA, Germer U, Gloeckner-Hofmann K, Gembruch U. Fetal hydrops and hepatosplenomegaly in the second half of pregnancy: a sign of myeloproliferative disorder in fetuses with trisomy 21. *Ultrasound Obstet Gynecol.* 2001;17:403–409.
- Hendricks S, Sorensen T, Baker E. Trisomy 21, fetal hydrops, and anemia: prenatal diagnosis of transient myeloproliferative disorder. *Obstet Gynecol.* 1993;82:703–705.

CASE PRESENTATION

We report the case of a 35-year-old multigravida at 20 weeks' gestation, where routine ultrasonography detected a small left pleural effusion. This progressed to large bilateral pleural effusions, ascites, scalp oedema, and severe polyhydramnios by the time of delivery at 37 weeks via elective lower-segment caesarean section (ELSCS).



Notably, foetal echocardiography revealed a structurally normal heart with no functional anomalies. Combined first-trimester screening indicated a low risk for Down syndrome, and both amniocentesis and non-invasive prenatal testing (NIPT) were declined. Given the absence of genetic testing by parental choice, a poor prognosis was anticipated, and an interdisciplinary team (MDT) approach was undertaken to prepare both the neonatal team and parents for possible complications.



Postnatally, the infant was diagnosed with Trisomy 21. Subsequent scans showed additional findings of Atrial Septal Defect and an atrophic right kidney. Peripheral blood film showed no signs of any myeloproliferative disorders. He underwent drainage of the pleural effusions immediately after birth. At 1 week of age, he was readmitted and diagnosed with duodenal atresia. He underwent surgical correction in a tertiary centre, which was complicated by sepsis post-operatively. Further diagnostic investigations also revealed pulmonary hypoplasia. Sadly, he passed away at six months of age. .