Thrombocytopenia and absent radius (TAR) syndrome in pregnancy

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DESCRIPTION

A healthy 36-year-old Caucasian woman, P2002, a non-smoker who did not consume alcohol, with no history of exposure to teratogenic agents, presented, at a gestational age of 15 weeks, for prenatal care at our hospital. Ultrasound findings showed a live fetus with bilateral absence of the radius, with club hands, unilateral renal agenesis, a small stomach and cardiac anomalies. In this situation, with a high suspicion of fetal polymalformative syndrome, an amniocentesis was performed, with consent of the patient, at 16 weeks. The cytogenetic analysis demonstrated a normal male karyotype and the study for Fanconi anaemia was negative. It was not possible to carry out thrombocytopenia and absent radius (TAR) syndrome studies. A fetal echocardiogram was performed at 19 weeks and the diagnosis of type I truncus arteriosus was made, with non-restrictive ventricular septal defect. The patient and her husband, after

counselling, opted for a medical interruption of pregnancy. This was performed at 24 weeks of gestation. A male abortus weighing 589 g was

Figure 2 Male fetus after medical interruption of pregnancy.

Figure 1 Radiograph of the fetus showing bilateral absence of the radius.

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Figure 3 Macroscopic findings: club hands.

delivered. Postnatal findings confirmed a syndrome with malformations of the upper limbs, as seen on the subsequent radiograph (figure 1) and macroscopic findings (figures 2 and 3). Umbilical cord puncture was made for the evaluation of fetal platelet counts. Thrombocytopenia (23 000/ μ L) was found in this case. The autopsy showed bilateral absence of the radius, with normal thumbs, oesophageal atresia, tracheo-oesophageal fistula, unilateral renal agenesis, one single umbilical artery and a congenital cardiovascular anomaly (truncus arteriosus),

without congenital vertebral anomalies. Thrombocytopenia and autopsy findings confirmed the diagnosis of TAR syndrome.

Learning points

- ▶ Radial aplasia is associated with chromosomal, teratogenic and genetic causes.¹ Thrombocytopenia is mandatory to differentiate thrombocytopenia and absent radius (TAR) syndrome from other malformations of the upper limbs.
- ▶ TAR syndrome is a rare disorder characterised by the absence of the radius; it is usually bilateral and presents a reduced platelet count.²

Competing interests None declared.

Patient consent Not obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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