AGENESIS OF VENOUS DUCTUS

BIBLIOGRAPHICAL REVIEW OF TWO CASES

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INTRODUCTION

The **venous ductus (DV)** is a physiological vascular shunt in the fetus that allows the passage of 20-30% of the oxygenated blood from the umbilical vein (VU) to the coronary and fetal cerebral circulation. **Venous ductus agenesis (ADV)** results from failure of the connection between VU and DV so there is a direct shunt between the VU and an aberrant vessel draining into the inferior vena cava (VCI), right atrium (AD), coronary sinus, iliac vein (VI) or portal system (SP). Its **incidence is 1/500 - 1/2500**. 20% of cases are presented in isolation but 80% are associated with portal system anomalies.

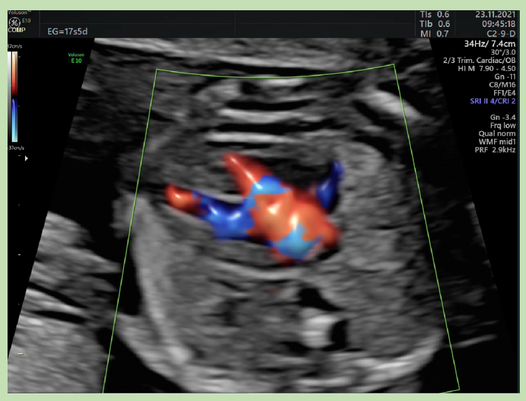
ADV is associated with chromosomal defects, genetic syndromes, structural defects and prenatal complications such as CIR, progressive heart failure, pulmonary edema, hepatic hyperplasia, liver tumors, systemic porto encephalopathy and even fetal death.

CLINICAL CASE 1

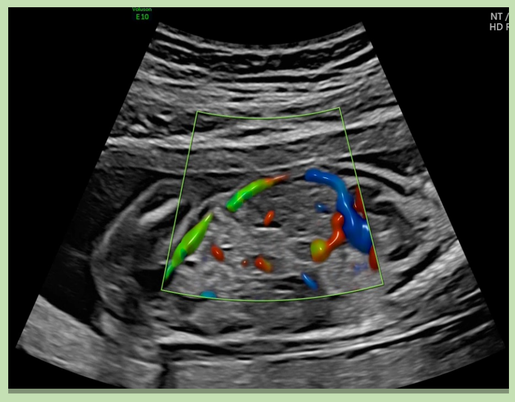
Primiparous with type I DM without other background

Ultrasound I trimester: TN p>99 → Amniocentesis: karyotype, arrays and normal exome.

Early morphological ultrasound (16sg): transitional AV channel and ADV with aberrant vessel that drains extra hepatic form to the AD. Permeable portal system and normal morphology.



Cut of 4 heart chambers with presence of AV channel



Sagital cut at chest level with aberrant vessel draining extrahepatically from VU to AD

Evolution

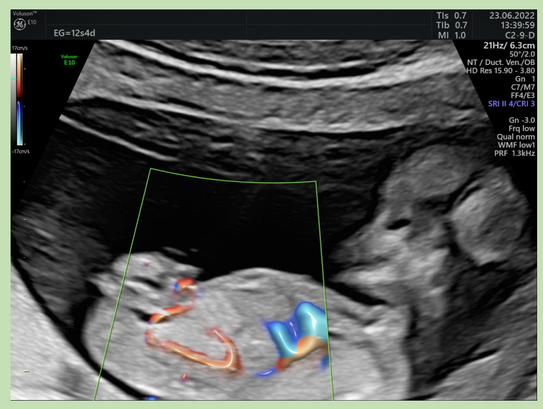
Spontaneous childbirth at 39+4 weeks.

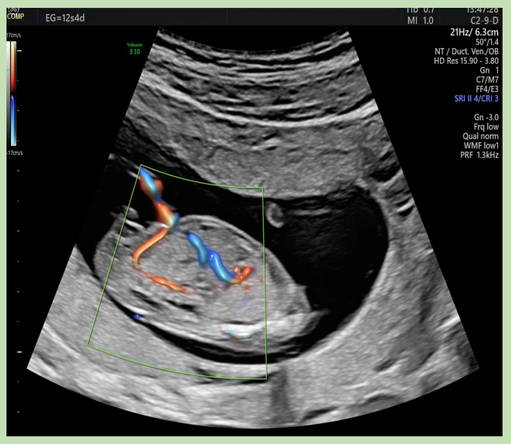
Echocardiography and postnatal MRI confirm transitional atrioventricular canal, agenesis of the left portal vein, moderate failure of the left AV component. Complex dorsal vertebral malformation and right medial diaphragmatic eventration-herniation with IHL ascent is evidenced.

Favorable evolution, high with follow-up by pediatric service.

CLINICAL CASE 2

Primiparous for IVF with ovodonation with type I DM and endometriosis with no other history. Ultrasound I trimester: isolated ADV with intrahepatic drainage.





Intrahepatic agenesis of venous ductus

Evolution

Induced delivery at 39+6sg due to pregestational diabetes ending in C-section because of a stationary delivery.

Right now, 5-mont-old baby without apparent pathology and without any postnatal complementary tests.

DISCUSSION

The diagnosis of ADV is usually performed in the first trimester ultrasound (11+0 to 13+6sg).

There are **two types of VU drainage**:

- Intrahepatic: VU drains into the portal venous system. It is associated with a **better prognosis** and accounts for **70%** of cases.
- Extrahepatic: VU drains into a systemic vein such as the IVC, AD, coronary sinus, or LV. It is associated with congestive heart failure and fetal hyperdrops in 30-50% of cases and is associated with a **worse prognosis**. It accounts for **30%** of cases.

ADV is highly associated with chromosomal (20-25%), cardiac (20%) and extracardiac abnormalities, also with portal venous system agenesis (50%) and persistent portosystemic short circuits.

Genetic study: FISH and karyotype and individualized use of array-CGH +/- exome.

Prenatal control: Umbilical and venous venous return should be evaluated in all fetuses with cardiomegaly, polyhydramnios, ascites or hydrops.

Postnatal control: In cases with ADV depends mainly on the presence of associated anomalies. In isolated cases, the prognosis is generally good (survival of 96%), but if abnormalities are associated, they may present 26% of adverse outcomes such as neonatal death, death in childhood or fetal pre-partum.

CONCLUSIONS

- ✧ ADV is a rare pathology but can be **associated with other malformations** in almost **80%** of cases.
- ✧ If ADV is **isolated** (20%), the **prognosis is excellent**.
- ✧ It can be diagnosed by ultrasound prenatal and is divided into two types depending on drainage**: Intrahepatic** and **extrahepatic** (worse prognosis). It is relevant to study whether there is cardiomegaly, polyhydramnios, ascites or hydrops.
- ✧ It is important to **offer genetic study** with karyotype, expanding with Array-CGH and exome if available in the center. Early diagnosis allows families to be offered different alternatives and treatment on an individual basis by a multidisciplinary team.

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