Absence of the Ductus Venosus (ADV)

What is the typical function of the ductus venosus?

The fetal ductus venosus is a funnel-shaped vessel that connects the umbilical vein (which carries oxygenated blood from the placenta to the baby) to the heart. Some of the oxygenated blood flowing through the umbilical vein is directed towards the liver. The ductus venosus acts as a shunt, allowing the remaining blood in the umbilical vein to bypass the liver and flow directly towards the heart. Thus, the ductus venosus plays an important role in the regulation of oxygenated blood in fetal life.

What does it mean if my baby has absence of the ductus venosus?

Absence of the ductus venosus (ADV) is a rare developmental change that can be diagnosed before birth. It has been separated into two different types based on the route of the baby's blood.

1) Intrahepatic: The umbilical vein connects to the liver but does not give rise to the ductus venosus.

Key Facts About ADV

- Occurs in 2 in 5,000 pregnancies to 25 in 5,000 pregnancies
- Two Types: Extrahepatic and Intrahepatic
- Diagnosis is made via fetal ultrasound
- Associated with an increased risk for birth defects and genetic conditions

2) Extrahepatic: The umbilical vein bypasses the liver and connects to regions involved in cardiovascular circulation.

The prognosis for a baby that lacks a ductus venosus differs depending on the route of their blood and whether other birth defects or genetic conditions are present.

How is absence of the ductus venosus typically diagnosed?

Absence of the ductus venosus can be diagnosed before birth via fetal ultrasound examination. Babies with absence of the ductus venosus typically present with an abnormal course of the umbilical and/or portal vein.

What conditions are associated with absence of the ductus venosus?

Absence of the ductus venosus is associated with an increased risk for birth defects and genetic conditions, with major developmental changes present in a significant number of cases. Conditions that are frequently observed in babies with absence of the ductus venosus are listed below.

- Variations in chromosome number
    - E.g., Turner syndrome, Down syndrome
- Partial or complete absence of the portal venous system

- Atrial septal defects
- Ventricular septal defects
- Congestive heart failure
- Malformations of the gastrointestinal and genitourinary systems
- Abnormal accumulation of fluid in several fetal compartments (i.e., fetal hydrops)

What should I expect moving forward? How might this diagnosis impact the management of my pregnancy?

Your baby's prognosis depends on the route of their blood–extrahepatic or intrahepatic–and whether additional developmental changes are present. There is greater concern for a baby's health when there are additional birth defects, an underlying genetic condition, and/or evidence that there is strain on the baby's heart. Among babies with no additional developmental changes, those with intrahepatic blood circulation have significantly better outcomes, compared to those with extrahepatic blood circulation. Some babies can survive with no long-term effects. The prognosis of babies with no additional developmental changes and extrahepatic blood circulation depends on the presence and extent of strain on the baby's heart.

Next Steps for Your Baby

- Detailed ultrasound
- Fetal echocardiogram
- Genetic screening and/or diagnostic testing
- Discuss pregnancy management with your doctor

Babies with absence of the ductus venosus are typically followed with a detailed ultrasound, a fetal echocardiogram, genetic screening, and/or diagnostic testing. Genetic testing can help identify the cause of your baby's developmental change(s). A variety of genetic tests are available, depending on the changes that are observed in your baby.
A genetic counselor can help you decide which, if any, testing option(s) you would like to pursue. Pregnancy management will vary depending on individual circumstances and should be discussed with your doctor.

If I have another child, what is the likelihood that they will have this same condition?

The recurrence risk of absence of the ductus venosus is unknown and depends on the chromosomal and/or structural abnormalities that are present.