

Absent Pulmonary Valve Syndrome

Patient Information Series – What you should know, what you should ask.

What is Absent Pulmonary Valve Syndrome?

Absent Pulmonary Valve Syndrome (APVS) is a rare congenital heart defect. In a typical heart, the pulmonary valve, located between the right ventricle (the lower, pumping chamber of the heart) and the pulmonary artery (the large blood vessel that carries blood toward the lungs), opens to allow blood to flow from the heart to the lungs. In APVS, the pulmonary valve is either missing or underdeveloped. The absence of a functional pulmonary valve often causes the pulmonary arteries to become enlarged (dilated). The right ventricle may become enlarged due to the increased workload of pumping blood into the pulmonary arteries without a functioning valve. Absent pulmonary valve syndrome may represent a very severe form of Tetralogy of Fallot (TOF).

What causes Absent Pulmonary Valve Syndrome?

The exact cause of this malformation is not fully understood, but it is thought to involve a combination of genetic and environmental factors. In many cases, APVS occurs sporadically, with no identifiable cause or risk factor. It might be a random event during the development of the heart. APVS often occurs in conjunction with other congenital heart defects, such as those seen in Tetralogy of Fallot. This suggests that the factors causing those defects might also contribute to the development of APVS.

Should I have more tests done?

You might be referred for a fetal echocardiogram, a specialized fetal ultrasound that focuses on the fetal heart. This test provides a more detailed view of the baby's heart structure, function, and blood flow. Genetic counseling and testing could be recommended. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing.

What are the things to watch for during the pregnancy?

You should have regular ultrasounds and echocardiograms to assess the baby's well-being and development, and the progression of the heart condition, which can evolve during the course of the pregnancy. You might meet with a multidisciplinary team, including an obstetrician specializing in high-risk pregnancies, a neonatologist (a specialist in caring for newborns), and pediatric heart surgeon, to discuss your baby's condition and their care going forward.

What does it mean for my baby after it is born?

With appropriate medical and surgical management, many children with APVS grow up to lead active and healthy lives. Babies with APVS may require immediate medical attention and possibly surgery

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after birth, so delivering at a facility equipped to provide appropriate neonatal care to meet your baby's needs, is important. Each baby with APVS is unique, in terms of whether other cardiac defects are present, their overall condition and prognosis. You should discuss with your healthcare provider and the multidisciplinary team the best place and mode of delivery for your baby, the potential surgeries they might require, long-term cardiac care they might need, and support for any additional needs. The team caring for you and your baby during pregnancy can advise you how best to plan for delivery and afterward.

Will it happen again?

Each pregnancy is different, and the likelihood of a recurrence of APVS can vary greatly for different families. Genetic counseling can provide more personalized risk assessment and information, if a genetic factor is identified in your case. Most isolated congenital heart defects (those not associated with a genetic syndrome), the recurrence risk in future pregnancies is generally considered to be higher than in the general population but still relatively low.

What other questions should I ask?

- Where should I deliver my baby?
- Are there any additional tests or evaluations that need to be performed during the pregnancy to assess my baby's condition further?
- Can you explain the expected course of treatment and surgeries that my baby will need after birth?
- What are the potential complications and risks associated with this heart defect and its treatment?
- What are the long-term implications of this heart defect for my child's health, development, and quality of life?

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