Rhombencephalosynapsis (RES)

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

Rhombencephalosynapsis (RES)

The cerebellum is also called the “small brain” and is a very important structure of the nervous system responsible for the coordination of movement and balance, among other important functions. The cerebellum has two sides (“hemispheres”) which are connected centrally by a structure called ‘vermis’. RES is a congenital malformation of the cerebellum, in which the vermis is completely or partially absent and both hemispheres are fused in the midline.

How does RES happen?

The fetal brain develops from folding and differentiation of the neural tube into its different structures. Current research suggests that RES is attributed to a defective developmental process that causes abnormal differentiation of the posterior part of the brain into the cerebellum and vermis in the early days of the embryo development.

Should I have more tests done?

RES often appears with other malformations either in the brain or other parts of the body. It is recommended that women get an advanced assessment of the fetal brain structures (dedicated ultrasound and/or MRI) and other organs by a professional with expertise in fetal brain imaging. This is important to confirm the diagnosis of RES and the presence of additional anomalies. We recommend also to performing an amniocentesis, to rule out other genetic causes of cerebellar anomaly.

What should I watch out for during the pregnancy?

RES does not affect the course of pregnancy by itself. In cases when the fetus develops hydrocephalus (severe dilatation of the lateral ventricles) and the pregnancy continues, sometimes there is a need for caesarean section, depending on the fetal head size.

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

RES can vary in severity. Babies with mild RES and no associated anomalies may have a normal life or just have issues with balance and physical coordination. However, more severe RES can include difficulties in swallowing, delayed motor learning, difficulty in speech, abnormal eye movement, mental retardation, and seizures. A significant number of cases of RES with severe symptoms may result in death by childhood.

The treatment of babies with RES is supportive, meaning the symptoms are treated as needed, and specific for the associated anomalies. When RES is associated with hydrocephalus or neural tube defect, there may be a need to perform a neurosurgical procedure.

Prognosis

The prognosis of postnatal diagnosed RES depends on the presence of other malformations and the severity of vermian absence. However, if RES is diagnosed in a fetus prenatally, the prognosis is typically worse.

Will it happen again?

The vast majority of cases are sporadic and do not recur in future pregnancies. There are very few cases in the medical literature of familial cases in which there is 25% chance of recurrence in these families.

What other questions should I ask?

- Should I be referred to a specialized center for exams and counseling?
- Is termination of pregnancy an option for my case?
- What type of delivery is more suitable for me, when, and where?

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