Schizencephaly

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

What is schizencephaly?

Schizencephaly, which means “split brain,” is a brain malformation at birth characterised by abnormal unilateral and/or bilateral slits or clefts in the cerebral hemispheres. It a rare condition with an estimated incidence of 1:5:100,000 births. The typical features include hemispheric grey-matter filled clefts (which might be unilateral in 50% of the cases) which lead to abnormal communication between the ventricular system and the subarachnoid space, and other multiple intracranial malformations such as absence of the septum pellucidum and corpus callosum.

What causes schizencephaly?

The underlying cause of schizencephaly remains unknown. Some have proposed an abnormal migration of primitive embryological cells (neuroblasts) in the brain to explain a spectrum of disorders amongst which schizencephaly is thought to be part of. Others believe schizencephaly results from vascular infarcts which have been associated with congenital infections, recreational drugs, and even monogenic mutations.

How is schizencephaly diagnosed?

There are two types of schizencephaly: closed- (or fused-) lips (type I) and open-lips (type II). Whereas type I is unlikely to be identified antenatally, type II with the open-lip clefts might be recognised at the mid-trimester scan (around 20 weeks’ pregnant). Additional cerebral malformations are almost always present including an absent septum pellucidum and corpus callosum. One-third of all post-natal cases will also present with extra-cerebral disruptive-in-origin malformations including gastroschisis, bowel atresia and amniotic bands.

Should I have more tests done?

If schizencephaly is suspected, further evaluation should be carried out by an expert in Fetal Medicine. Schizencephaly might present very similarly to other anomalies in the scan such as lobar holoprosencephaly and/or intracranial cysts. The presence of cortical clefts is therefore paramount to arrive at the right diagnosis. Additional evaluation should include a detailed exam of all other organs by ultrasound, as the presence of other anomalies will inform prognosis and further management.

What is the prognosis?

The prognosis is variable and depends on the severity of the brain lesions and extra-cerebral anomalies. It ranges from a mild seizure disorder to extensive motor impairment and mental retardation.

Will it happen again?

Apart from the few familial cases with a germinal mutation, the risk recurrence is thought to be exceptionally low.

What other questions should I ask?

- Are there any other abnormalities on the ultrasound?
- How often should I have ultrasound examinations?
- Where should I deliver?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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