Cerebellar hypoplasia (CH)

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

What is CH?

The cerebellum is an important structure in the back of the brain that, among other functions, helps coordinate balance and movement. CH is a term used when the size of the fetal cerebellum is smaller than the expected for the gestational age. This is a generic term. CH may be found alone or as part of several medical conditions, including brain malformations, congenital syndromes or acquired lesions (such as brain bleeding or infections).

How does CH happen?

It all depends on the underlying cause leading to the abnormal development of the cerebellum – either a genetic or an acquired lesion can destroy the normal structure or prevent normal cerebellar development. This finding may affect only one side of the cerebellum, one side and and the central part (called “vermis”) or all parts in different degrees of severity. If only the central part is affected it is called vermian hypoplasia and has different clinical significance. Sometimes the lesion is caused by bleeding inside the cerebellum, a viral infection or severe fetal anemia. In other cases, the anomaly is caused by a genetic disease (chromosomal or specific gene mutation).

Should I have more tests done?

Once the fetal cerebellum is found to measure smaller than expected for gestational age (bellow the 10th percentile) an advanced imaging investigation of the brain structures and other systems of the body is needed. First of all, a dedicated neurosonography (detailed br ain ultrasound) is indicated as well as genetic counseling. If needed and feasible, an amniocentesis will be advised for genetic testing of the fetus. Laboratory tests for infections or coagulation tests are indicated depending on the ultrasound findings. In some cases, fetal magnetic resonance imaging (MRI) is also needed and has the highest contribution when performed late in pregnancy.

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

The postnatal presentation of children with CH varies according to the main cause and the presence of other anomalies. When the finding is restricted to the cerebellum, most children have neurological problems such as difficulty in walking that presents early in infancy (called “ataxia”), muscular

weakness and may present with developmental delay in different degrees (more than 60% of cases). The delay is severe in about 35% of cases. Later, eye movement problems, speech disorders and small brain volume may arise. When the clinical presentation is static (i.e. does not worsen with time), the prognosis is better.

When other malformations are present, the clinical presentation is more severe and will depend on the other organs affected.

Will it happen again?

For developmental CH, the risk of recurrence will depend on the type of genetic disorder present and if the genetic change was or was not inherited from one or both parents. If the problem is an additional whole chromosome, most cases are isolated and do not occur again. Some cases have deletion or duplication of only part of a chromosome and these cases are often inherited. When there is a mutation in one gene, it depends on whether the parents are “carriers” and if so, may “transmit” the variant to every new fetus in different risk proportions according to the type of variant. In cases when the parents also have the mutation (either healthy or also having the anomaly) normally the risk may vary from 25% to 50%. In some cases, the chance of having abnormal outcome is associated to the fetal sex. If the cause is acquired, the recurrence chance is very low. Because many cases of CH have a genetic cause, the genetic counseling is very important.

What other questions should I ask?

- Does this look like a severe case of CH?
- Are other organs involved?
- Does CH have any implications for the route and timing of the delivery?
- How often will I have ultrasound examinations done?
- Should I be referred to a tertiary center for further investigation and delivery?

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