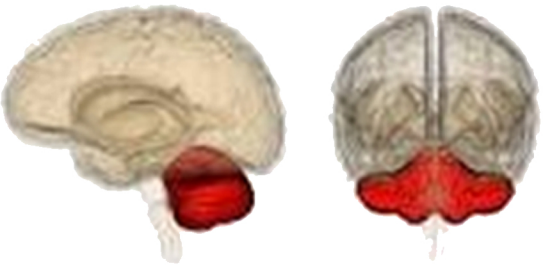
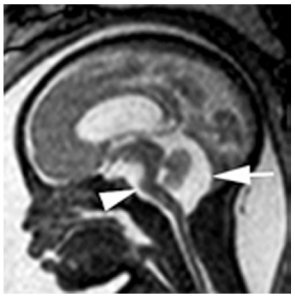
Patient Information Series- 5

Asian Institute of Fetal Medicine & Reproductive Genetics,

Cerebellar hypoplasia (CH) in Fetus



What is Cerebellar Hypoplasia?



The cerebellum is a small but very important part of the brain. It sits at the back of the head and helps the body stay balanced, move smoothly, and coordinate actions like walking, sitting, and picking things up. It also helps with some thinking and learning skills.

CH means this part of the brain is smaller than expected for the baby's age in pregnancy. This does not always mean that the cerebellum is missing; rather, it has not grown as much as usual.

CH can affect one side, the vermis, or the whole cerebellum. It may occur on its own (isolated CH) or along with other abnormalities, in which case it may be part of a more severe medical condition.

Why does CH happen?

There are two main reasons why a baby may have CH: genetic causes and acquired causes.

1. Genetic causes

Genetic causes mean that something in the baby's chromosomes or genes affects how the cerebellum develops. Examples include:

- A whole extra chromosome
- A missing or extra piece of a chromosome
- A change in a specific gene

These changes may happen by chance or may come from one or both parents. Not all genetic changes cause problems,but some can affect brain growth.

2. Acquired causes

These are events during pregnancy that interfere with normal brain development. They may include:

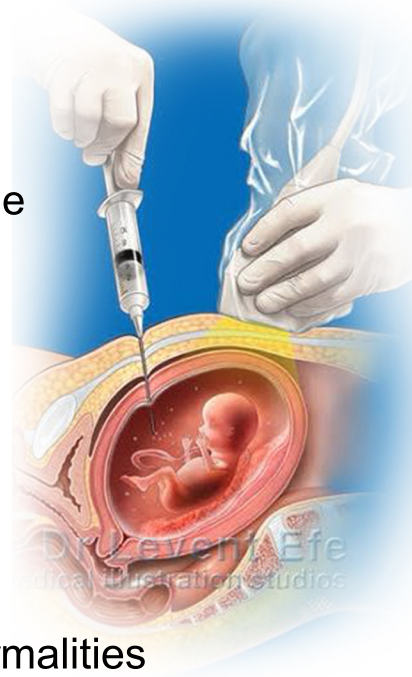
- Bleeding in the cerebellum
- Certain viral infections
- Severe fetal anaemia

These events can damage the tissue that forms the cerebellum or stop it from developing normally. In many cases, the exact cause becomes clear only after further tests.

What tests may be needed?

If the cerebellum appears smaller on a routine pregnancy scan, further tests are recommended to understand how much of the brain is affected and to identify possible causes.

- Detailed Brain & Heart USG scan: Looks closely at the baby's brain &heart
- Amniocentesis: Checks chromosomes and specific gene changes
- Blood tests: Screen for infections or clotting problems
- Fetal MRI: Provides very clear brain images, especially later in pregnancy



Types of Genetic Testing

- Karyotype analysis: Detects large chromosomal abnormalities
- FISH: A limited test but provides quick results
- Chromosomal microarray (CMA): Detects smaller chromosomal changes not seen on karyotype

- Whole exome sequencing (WES): Helps identify a genetic cause when karyotype and CMA are normal

The choice of test is made after considering all risk factors and clinical findings.

What does CH mean for my baby after birth?

The outlook varies depending on how much of the cerebellum is affected and whether other abnormalities are present.

If CH is isolated Children may have:

- Movement and balance problems (ataxia) • Low muscle strength
- Developmental delays • Possible speech or eye-movement difficulties

If the condition is non-progressive, many children improve with therapy, and early support can make a significant difference.

If other abnormalities are present

The outcome may be more serious and depends on which organs or brain areas are affected. Your healthcare team will explain this based on your baby's results.

Will this happen again in future pregnancies?

If CH was caused by infection, bleeding, or anaemia, the chance of it happening again is usually very low.

Because many cases may have a genetic cause, it is helpful to speak with a genetic counsellor before planning another pregnancy.

If a genetic cause is identified:

- The risk may be low or high depending on the condition
- Extra whole chromosomes usually do not repeat
- Missing or extra pieces of chromosomes may be inherited
- Single-gene mutations may carry a 25–50% recurrence risk, depending on inheritance

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