What is Down Syndrome

Human cells normally contain 23 pairs of chromosomes. One chromosome in each pair comes from your father, the other from your mother.

Down syndrome results when abnormal cell division involving chromosome 21 occurs. These cell division abnormalities result in extra genetic material from chromosome 21, which is responsible for the characteristic features and developmental problems of Down syndrome. Any one of three genetic variations can cause Down syndrome:

- **Trisomy 21.** About 95 percent of the time, Down syndrome is caused by trisomy 21 — the child has three copies of chromosome 21 (instead of the usual two copies) in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.
- **Mosaic Down syndrome.** In this rare form of Down syndrome, children have some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.
- **Translocation Down syndrome.** Down syndrome can also occur when part of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional material from chromosome 21 attached to the translocated chromosome.

There are no known behavioral or environmental factors that cause Down syndrome.

Is it inherited?

Most of the time, Down syndrome isn't inherited. It's caused by a mistake in cell division during the development of the egg, sperm or embryo.

Translocation Down syndrome is the only form of the disorder that can be passed from parent to child. However, only about 4 percent of children with Down syndrome have translocation. And only about one-third of these children inherited it from one of their parents.

When translocations are inherited, the mother or father has some rearranged genetic material, but no extra genetic material — this means he or she is a balanced carrier. A balanced carrier has no signs or symptoms of Down syndrome, but he or she can pass the translocation on to children, causing extra genetic material from chromosome 21.

The chance of passing on the translocation depends on the sex of the parent who carries the rearranged chromosome 21:

- If the father is the carrier, the risk is about 3 percent.
- If the mother is the carrier, the risk is between 10 and 15 percent.