Understanding Down Syndrome

Down syndrome is a genetic disorder. It's also called trisomy 21. It causes certain birth defects, learning problems, and facial features. A child with Down syndrome may have heart defects. They may have problems with vision and hearing. How severe or mild these are varies from child to child.

Down syndrome is one of the most common genetic birth defects. It affects about 1 in 700 babies. Adults with Down syndrome may live about 60 years. But this can vary.

What causes Down syndrome?

When a baby is conceived, a sperm cell enters an egg cell. A normal egg cell and normal sperm cell start with 46 chromosomes. The egg and sperm cells then divide in half. They then have 23 chromosomes each. When a sperm with 23 chromosomes fertilizes an egg with 23 chromosomes, the baby will then have a full set of 46 chromosomes. Half are from the father and half are from the mother.

But sometimes an error occurs when the 46 chromosomes are dividing in half. An egg or sperm cell may keep both copies of chromosome number 21, instead of just 1 copy. If this egg or sperm is fertilized, then the baby will have 3 copies of chromosome number 21. This is called trisomy 21.

In some cases, the extra number 21 chromosome or part of it attaches to another chromosome in the egg or sperm. This may cause translocation Down syndrome. This is the only form of Down syndrome that may be inherited from a parent.

A rare form is called mosaic trisomy 21. This is when an error in cell division happens after the egg is fertilized. People with this syndrome have both normal cells and some cells with an extra chromosome number 21.

A mother’s age at her child’s birth is a risk factor. This risk increases with each year of age, especially after age 35.

Symptoms of Down syndrome

Symptoms can vary in each child. Most children with Down syndrome will have some but not all of these features:

- Eyes that slant up
- Small ears that may fold over slightly at the top
- Small mouth that makes the tongue seem large
- Small nose with a flattened bridge
- Short neck
- Small hands with short fingers
- 2 instead of 3 palm creases, including 1 across the palm and 1 around the base of the thumb
- Short height
- Loose joints

Down syndrome can also include:

- Heart defects
- Intestinal problems

- Vision problems
- Hearing problems
- Thyroid problems
- Blood conditions such as leukemia
- Higher risk of infections
- Learning problems

Diagnosing Down syndrome

Chromosome changes can often be found before birth with screening tests. These may combine a blood test with an ultrasound. A screening test can show if the baby has more or less risk. But they don't make a diagnosis. A diagnostic test is more invasive and risky. But it can find the disorder.

Screening tests include:

- Screening ultrasound. This test looks at the amount of amniotic fluid. Extra fluid means there is a problem.
- Blood tests. These look at many substances in the blood to show a possible risk. The blood tests include MS-AFP, Triple Screen, and Quad-screen. These tests look at levels of things such as alpha-fetoprotein, human chorionic gonadotropin, and estriol.
- Fetal ultrasound. This can show the possibility of Down syndrome. But it's not 100% accurate. Some problems from Down syndrome may not be seen with ultrasound.

Diagnostic tests look at cells in the amniotic fluid or from the placenta. They look for changes in the chromosomes. These tests include:

- Chorionic villus sampling. This test looks at cells from the placenta.
- Amniocentesis. This tests the fluid in the sac around the baby (amniotic fluid).
- Percutaneous umbilical blood sampling. This tests blood from the umbilical cord.

After birth, your baby may be diagnosed with a physical exam. The healthcare provider may take a blood sample. This is checked in a lab to find the extra chromosome.

Can Down syndrome be prevented?

Researchers don’t know how to prevent the chromosome errors that cause this disorder. There is no known way parents can cause or prevent Down syndrome in their child.

For women who have had 1 child with Down syndrome, the chance of having another baby with Down syndrome depends on several things. Age is one factor.

Your healthcare provider may refer you to a genetic counselor. This expert can explain the results of chromosome tests in detail. They can talk about risks for future pregnancies. They can order tests to diagnose chromosome problems before a baby is born.

Some medical organizations advise that all pregnant people of any age be offered screening for Down syndrome. Talk with your healthcare provider about this prenatal screening test.