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ORIGIN, TREATMENT AND PREVENTION OF DOWN SYNDROME

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Annotation: Down syndrome is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. This extra genetic material causes the developmental changes and physical features of Down syndrome.

Key words: Down syndrome, children, genetic material.

Down syndrome varies in severity among individuals, causing lifelong intellectual disability and developmental delays. It's the most common genetic chromosomal disorder and cause of learning disabilities in children. It also commonly causes other medical abnormalities, including heart and gastrointestinal disorders.

Better understanding of Down syndrome and early interventions can greatly increase the quality of life for children and adults with this disorder and help them live fulfilling lives.

Each person with Down syndrome is an individual — intellectual and developmental problems may be mild, moderate or severe. Some people are healthy while others have significant health problems such as serious heart defects.

Children and adults with Down syndrome have distinct facial features. Though not all people with Down syndrome have the same features, some of the more common features include:

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet
- Excessive flexibility

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- Tiny white spots on the colored part (iris) of the eye called Brushfield's spots
- Short height

Infants with Down syndrome may be average size, but typically they grow slowly and remain shorter than other children the same age.Most children with Down syndrome have mild to moderate cognitive impairment. Language is delayed, and both short and long-term memory is affected.

Children with Down syndrome usually are diagnosed before or at birth. However, if you have any questions regarding your pregnancy or your child's growth and development, talk with your doctor.

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 an extra partial or full chromosome 21. This extra genetic material 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 person 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, a person has only 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 a portion 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 genetic material from chromosome 21 attached to another chromosome.

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

Most of the time, Down syndrome isn't inherited. It's caused by a mistake in cell division during early development of the fetus.

Translocation Down syndrome can be passed from parent to child. However, only about 3 to 4 percent of children with Down syndrome have translocation and only some of them inherited it from one of their parents. When balanced translocations are inherited, the mother or father has some rearranged genetic material from chromosome 21 on another chromosome, but no extra genetic material. This means he or she has no signs or symptoms of Down syndrome, but can pass an unbalanced translocation on to children, causing Down syndrome in the children.

Some parents have a greater risk of having a baby with Down syndrome. Risk factors include:

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- Advancing maternal age. A woman's chances of giving birth to a child with Down syndrome increase with age because older eggs have a greater risk of improper chromosome division. A woman's risk of conceiving a child with Down syndrome increases after 35 years of age. However, most children with Down syndrome are born to women under age 35 because younger women have far more babies.
- Being carriers of the genetic translocation for Down syndrome. Both men and women can pass the genetic translocation for Down syndrome on to their children.
- Having had one child with Down syndrome. Parents who have one child with Down syndrome and parents who have a translocation themselves are at an increased risk of having another child with Down syndrome. A genetic counselor can help parents assess the risk of having a second child with Down syndrome.

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