

General Information about Down Syndrome

What is Down Syndrome?

Down syndrome (DS), also called Trisomy 21, is a condition in which extra genetic material causes delays in the way a child develops, both mentally and physically. It affects about 1 in every 800 babies.

The physical features and medical problems associated with Down syndrome can vary widely from child to child. While some kids with DS need a lot of medical attention, others lead healthy lives.

Though Down syndrome can't be prevented, it can be detected before a child is born. The health problems that can go along with DS can be treated, and there are many resources within communities to help kids and their families who are living with the condition.\

What Causes It?

Normally, at the time of conception a baby inherits genetic information from its parents in the form of 46 chromosomes: 23 from the mother and 23 from the father. In most cases of Down syndrome, a child gets an extra chromosome 21 — for a total of 47 chromosomes instead of 46. It's this extra genetic material that causes the physical features and developmental delays associated with DS.

Although no one knows for sure why DS occurs and there's no way to prevent the chromosomal error that causes it, scientists do know that women age 35 and older have a significantly higher risk of having a child with the condition. At age 30, for example, a woman has about a 1 in 900 chance of conceiving a child with DS. Those odds increase to about 1 in 350 by age 35. By 40 the risk rises to about 1 in 100.

How Down Syndrome Affects Kids

Kids with Down syndrome tend to share certain physical features such as a flat facial profile, an upward slant to the eyes, small ears, and a protruding tongue.

Low muscle tone (called **hypotonia**) is also characteristic of children with DS, and babies in particular may seem especially "floppy." Though this can and often does improve over time, most children with DS typically reach developmental milestones — like sitting up, crawling, and walking — later than other kids.

At birth, kids with DS are usually of average size, but they tend to grow at a slower rate and remain smaller than their peers. For infants, low muscle tone may contribute to sucking and feeding problems, as well as constipation and other digestive issues. Toddlers and older kids may have delays in speech and self-care skills like feeding, dressing, and toilet teaching.

Down syndrome affects kids' ability to learn in different ways, but most have mild to moderate intellectual impairment. Kids with DS can and do learn, and are capable of developing skills throughout their lives. They simply reach goals at a different pace — which is why it's important not to compare a child with DS against typically developing siblings or even other children with the condition.

Kids with DS have a wide range of abilities, and there's no way to tell at birth what they will be capable of as they grow up.

Medical Problems Associated With DS

While some kids with DS have no significant health problems, others may experience a host of medical issues that require extra care. For example, almost half of all children born with DS will have a congenital heart defect.

Kids with Down syndrome are also at an increased risk of developing pulmonary hypertension, a serious condition that can lead to irreversible damage to the lungs. All infants with Down syndrome should be evaluated by a pediatric cardiologist.

Approximately half of all kids with DS also have problems with hearing and vision. Hearing loss can be related to fluid buildup in the inner ear or to structural problems of the ear itself. Vision problems commonly include amblyopia (lazy eye), near- or farsightedness, and an increased risk of cataracts. Regular evaluations by an audiologist and an ophthalmologist are necessary to detect and correct any problems before they affect language and learning skills.

Other medical conditions that may occur more frequently in kids with DS include thyroid problems, intestinal abnormalities, seizure disorders, respiratory problems, obesity, an increased susceptibility to infection, and a higher risk of childhood leukemia. Upper neck abnormalities are sometimes found and should be evaluated by a physician (these can be detected by cervical spine X-rays). Fortunately, many of these conditions are treatable.