

DOWN SYNDROME

(TRISOMY 21)

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DOWN SYNDROME
(also called Trisomy 21)

Prevalence

Down syndrome occurs in 1 of 800-1000 live births and is the most common genetic cause of developmental delay. The risk increases with the mother's age (1 in 1250 for a 25 year old mother to 1 in 1000 at age 31, 1 in 400 at age 35, and about 1 in 100 at age 40). However, 80% of babies with Down syndrome are born to women under age 35 years. Translocation Down syndrome has a greatly increased risk of occurring again in a subsequent pregnancy.

Condition Description

Down syndrome is a genetic birth defect that is caused at the time of the union of the egg and sperm. For an unknown reason, an extra chromosome (number 21) is added, so that each of the baby's cells has this extra genetic material (called nondisjunction, the cause of 95% of Down syndrome). Two other variations of Down syndrome can occur, but rarely. Translocation Down syndrome (4%) results from having the extra genetic material attached to another chromosome rather than to chromosome #21; this rare kind of Down syndrome can be inherited, unlike the other varieties. The third kind of Down syndrome (1%) is called mosaic, where some cells have the extra chromosome and other cells are normal. Down syndrome results in unique facial features and developmental delay in all the individuals. Generally the facial features will be noted at birth and the diagnosis will be confirmed with chromosomal analysis (the baby's genetic karyotype).

Children with Down syndrome have widely spaced eyes that slightly slant upward with a small nose flattened at the bridge. Microcephaly (small head size), low muscle tone, large tongue, short neck, small hands and short fingers are also characteristic features. Most children with Down syndrome are short in stature.

Common Associated Conditions

The degree of intellectual disability is usually mild or moderate, with less than 10% considered severely developmentally delayed. Typically, children with Down syndrome have social skills that exceed their cognitive skills. Heart defects are common (50%) and can vary from atrial septal defect, ventricular septal defect, tetralogy of Fallot or patent ductus arteriosus. Vision problems such as cataracts and amblyopia occur in 70% of children with Down syndrome, and hearing problems occur in about 60%. About 10% of children will have an intestinal malformation of some type, requiring surgery (typically duodenal or esophageal atresia, Hirschsprung's disease, imperforate anus, tracheoesophageal fistula or pyloric stenosis). Celiac disease occurs in about 15% of children with Down syndrome, and must be treated with a gluten-free diet.



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DOWN SYNDROME – page 2

Low muscle tone and loose ligaments are common with Down syndrome, resulting in dislocated hips, scoliosis, joint/muscle pain and muscle weakness. Some children with Down syndrome (about 10-20%) will have atlantoaxial instability (AAI), a condition with loose ligaments between the first and second cervical vertebrae in the neck that can often be seen on x-ray. AAI puts them at risk of spinal cord injuries if this joint becomes dislocated, and neurologic symptoms from AAI will occur in about 1% of children with Down syndrome. X-ray screening of children with Down syndrome to detect AAI is required before participation in certain contact sports in the Special Olympics.

Short-term Treatment and Outcomes

The number and type of associated conditions will determine treatment for children with Down syndrome. Breast-feeding should be encouraged because of the increased immunity that breast milk offers. However, babies with Down syndrome may have a weak and uncoordinated suck, so infant feeding requires patience, and special help from lactation experts might be recommended.

Coordinated interdisciplinary health and educational services are essential. All children with Down syndrome should be examined by a pediatric cardiologist since half will have cardiac defects; an echocardiogram should also be done in infancy to rule out heart problems. Because over 50% of children will have crossed eyes (called strabismus, that can lead to vision loss called amblyopia), near-sightedness, farsightedness or cataracts, children should be examined by a pediatric ophthalmologist at regular intervals. Glasses, eye patching or perhaps eye surgery will be helpful in maximizing the child's vision.

Children with Down syndrome have decreased resistance to infections and are more prone to dental disease, leukemia, and other infections. Insulin-dependent diabetes is twice as prevalent in children with Down syndrome. The child is also prone to ear infections because of having small Eustachian tubes and may need tympanostomy tubes (also called PE or pressure equalizing tubes) to treat fluid in the middle ear that muffles their hearing. Making sure that children with Down syndrome are seeing and hearing well will be very important so that they are not hindered in their learning and sensory input. Families should be offered genetic counseling to be informed of the risk for recurrence.

Long-term Treatment and Outcomes

Children with Down syndrome are at increased risk for respiratory problems during surgery because of their anatomy and low muscle tone (hypotonia). Stridor and apnea after surgery are common and thus children with Down syndrome should be very carefully assessed pre and post-operatively. Children with Down syndrome may be more prone to infections such as ear infections and pneumonia. All infections should be treated aggressively. Sleep apnea is common and is often relieved by adenoid and tonsil removal.

Immunizations and consistent health care are extremely important. Their growth should be monitored using special growth charts that compare them to other children with Down syndrome. They may be more prone to obesity, and interventions for weight management are important in order to keep them active and healthy. They may be prone to constipation because of their low muscle tone and inactivity and thus a high-fiber diet will be useful.

DOWN SYNDROME – page 3

Puberty usually occurs at the same time as their peers, and children and parents will need help to learn how to handle their sexuality in socially appropriate ways.

Children with Down syndrome are at increased risk for thyroid problems (20% incidence) and leukemia (18-20 times more common in Down syndrome than in the general population). Life expectancy is about 55 years. Most men are sterile but women can conceive. They will have a 50% chance of having a baby with Down syndrome.

Common Complications

About 15-20% of people with Down syndrome develop Alzheimer's dementia in middle age because the gene coding for the amyloid protein of the disease is located on chromosome 21. Premature aging is seen by the age of 30-40 years and their memory and intellectual function often deteriorates at that time. Mitral valve prolapse occurs in 50% of individuals with Down syndrome in later life.

Implications for Children's Development

Children with Down syndrome will walk, talk, become toilet trained and develop many skills of independent living. However they will learn new skills more slowly than most other children. Most will test at an intelligence quotient (IQ) of 40-55. Early intervention programs beginning in infancy can be very helpful to children and families in making sure that they have all the assistance possible to develop and remain healthy. Inclusion of children with their age mates results in developmental gains. Speech therapy may be helpful to encourage children to learn to speak with good articulation.

Most children with Down syndrome can be mainstreamed into regular classrooms for at least part of their education. With special education, most children will develop some reading and writing skills.

There is a range of developmental attainment of children with Down syndrome, but in general, their skills have been underestimated. They can take part in recreational activities with other children if there is some degree of accommodation for their delays. Their social development must be encouraged so that they can develop friendships and some degree of independent living.

Special work programs and semi-independent community living arrangements promise a full life for children with Down syndrome if families are assisted to guide their child in supervised independence that matches the child's abilities and decision-making skills.

Parental education and support are essential, and local, regional and national organizations are very helpful. Safety issues, sexuality, and transition to adult care services are big issues that parents face as the years go by for the child with Down syndrome.

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