What is chromosome 21?

Health & Medicine, Cancer

Down syndrome (DS) is a condition in which extra genetic material causes delays in the way a child develops, and often leads to mental retardation. It affects 1 in every 800 babies born. The symptoms of Down syndrome can vary widely from child to child. While some kids with DS need a lot of medical attention, others lead very healthy and independent lives. Though Down syndrome can't be prevented, it can be detected before a child is born. Thehealthproblems 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.

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, however, a child gets an extra chromosome - for a total of 47 chromosomes instead of 46. It's this extra genetic material that causes the physical and cognitive 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 less than a 1 in 1, 000 chance of conceiving a child with DS. Those odds increase to 1 in 400 by age 35. By 42, it jumps to about 1 in 60. How Does Down Syndrome Affect a Child? Kids with Down syndrome tend to share certain physical features such as a flat facial profile, an upward slant to the eyes, small ears, a single crease across the center of the palms, and an enlarged tongue. Adoctorcan usually tell if a newborn has the condition through a physical exam. Low muscle tone and

loose joints are 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. In toddlers and older children, there may be delays in speech and self-care skills like feeding, dressing, and toilet teaching.

Down syndrome affects kids' cognitive abilities in different ways, but most have mild to moderate mental retardation. Kids with DS can and do learn, and are capable of developing skills throughout their lives. They simply reachgoalsat a different pace - which is why it's important not to compare a child with DS with 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 Down Syndrome. While some kids with DS have no other health problems, others may experience a host of medical issues that require extra care. For example, half of all children born with DS also have congenital heart defects and are prone to developing pulmonary hypertension (high blood pressure in the lungs). A pediatric cardiologist can monitor these types of problems, many of which can be treated with medication or surgery. 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 a child's language and learning skills. Other medical conditions that may occur more frequently in children with DS include thyroid problems, intestinal abnormalities, seizure disorders, respiratory problems, obesity, an increased susceptibility to infection, and a higher risk ofchildhoodleukemia. Fortunately, many of these conditions are treatable. What is Chromosome 21?

According to wikipedia. org, Chromosome 21 is one of the 23 pairs of chromosomes in humans. People normally have two copies of this chromosome. The trisomy of the 21 causes Down Syndrome. Chromosome 21 is the smallest human chromosome, pning almost 47 million nucleotides (the building material of DNA) and representing about 1. 5 percent of the total DNA in cells. The Human Genome Project announced that they had determined the sequence of base pairs that make up this chromosome. Chromosome 21 was the second human chromosome to be fully sequenced. Chromosome 21 likely contains between 200 and 400 genes.

These include: • APP: amyloid beta (A4) precursor protein (peptidase nexin-II, Alzheimer disease) • CBS: cystathionine-beta-synthase • CLDN14: claudin 14 • HLCS: holocarboxylase synthetase (biotin-(proprionyl-Coenzyme A-carboxylase (ATP-hydrolysing)) ligase) • KCNE1: potassium voltage-gated

channel, Isk-relatedfamily, member 1 • KCNE2: potassium voltage-gated channel, Isk-related family, member 2 • LAD: leukocyte adhesion deficiency (symbols are ITGB2, CD18, LCAMB) • SOD1: superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult)) • TMPRSS3: transmembrane protease, serine 3.

Effects of Chromosome 21 The effects of chromosome 21 is best felt by the conditions that are associated with its lack or mutation. These include: Cancer Rearrangements (translocations) of genetic material between chromosome 21 and other chromosomes have been associated with several types of cancer. For example, acute lymphoblastic leukemia (a type of blood cancer most often diagnosed in childhood) has been associated with a translocation between chromosomes 12 and 21. Another form of leukemia, acute myeloid leukemia, has been associated with a translocation between chromosomes 8 and 21.

Down Syndrome In a small percentage of cases, Down syndrome is caused by a rearrangement of chromosomal material between chromosome 21 and another chromosome. As a result, a person has the usual two copies of chromosome 21, plus extra material from chromosome 21 attached to another chromosome. These cases are called translocation Down syndrome. Researchers believe that extra copies of genes on chromosome 21 disrupt the course of normal development, causing the characteristic features of Down syndrome and the increased risk of medical problems associated with this disorder.

Mental Retardation Other changes in the number or structure of chromosome 21 can have a variety of effects, including mental retardation, delayed development, and characteristic facial features. In some cases, the signs and symptoms are similar to those of Down syndrome. Changes to chromosome 21 include a missing segment of the chromosome in each cell (partial monosomy 21) and a circular structure called ring chromosome 21. A ring chromosome occurs when both ends of a broken chromosome are reunited.

Alzheimer’s Disease. Duplication in Amyloid precursor protein (APP) locus (duplicated segment varies in length but includes APP) on Chromosome 21 was found to cause early onset familial Alzheimer's disease (AD) in a french family set (Rovelet-Lecrux et al) and a dutch family set (Sleegers et al). Compared to AD caused by missense mutations in APP, the frequency of the AD caused by APP duplications is significant. ALL the patients that have an extra copy of APP gene due to the locus duplication show AD with severe Cerebral amyloid angiopathy (CAA).