Down Syndrome Review

Part 2

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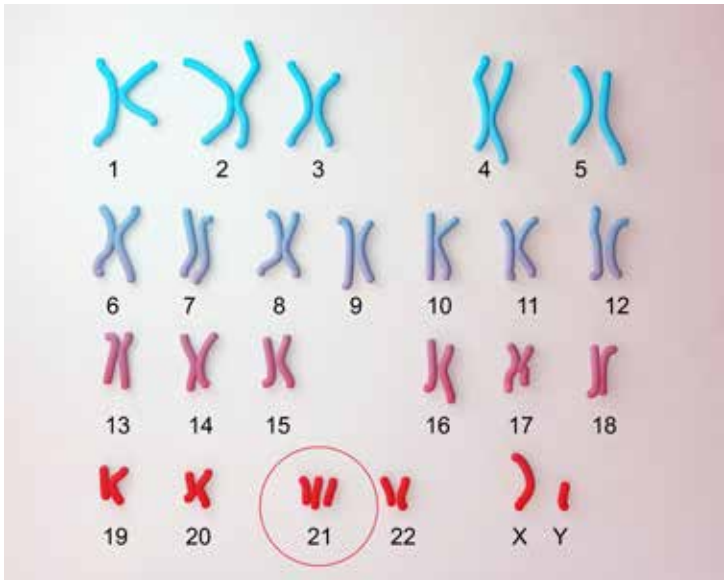


Vision

About seventy percent of children with T21 will have vision problems. Nearsightedness, farsightedness, and crossed eyes (strabismus) are the common visual disturbances found in children with T21. As a result, annual vision tests are very important for children with T21. Most of the conditions mentioned in the previous part of this article (https://www.naturopathiccurrents.com/articles/down-syndrome) are easily correctable.

Antioxidants such as vitamin A and the carotenoids are vital for the health of the vision.[1][2]

Hearing

About forty to sixty percent of babies with T21 will have some form of hearing loss. Most infants in North America are screened for hearing loss shortly after birth. It is unlikely that an infant with T21 will have complete hearing loss. However, the detection of a hearing impediment is important, since any hearing loss will play a large role in language development.

There may be some degree of hearing loss. This is most often due to frequent ear infections. Because of the smaller head

size and small ears, children with T21 are more susceptible to frequent ear infections. Adequate hearing is critical to the development of good language skills. As a result, periodic medical checkups should include an ear examination and hearing tests.[3]

Prenatal Screening

Between the fifteenth and eighteenth week of pregnancy, a woman's blood can be checked for markers. The "triple test" is a combination of *alpha*-fetoprotein (AFP), unconjugated estriol (uE3), and human chorionic gonadotropin (hCG); when inhibin A is added, this test is called the "quadruple screen." These tests are independent measurements and, when taken along with the maternal age, can help calculate the risk of having a baby with T21.

alpha-Fetoprotein is made in the part of the womb called the yolk sac and in the fetal liver. Some amount of AFP will be found in the mother's blood. In neural-tube defect, the skin of the fetus is not intact and as a result, a large amount of AFP is measured in the mother's blood. In T21, the AFP is decreased in the mother's blood; this may be due to fact that the yolk sac and the fetus are smaller than usual.[4]

Estriol is a hormone produced by the placenta, fetal liver, and adrenal gland. Estriol is decreased in a pregnancy carrying a fetus with T21.

Human chorionic gonadotropin hormone is produced by the placenta. This is the hormone that is measured when one tests for pregnancy. A specific smaller part of the hormone, called the *beta* subunit, is increased in a pregnancy with a fetus with T21.

Inhibin A is a protein made by the ovary. Inhibin is produced to reduce the level of FSH, which is made by the mother's pituitary gland. This hormone helps to keep the pregnancy viable. The level of inhibin A is increased in the blood of mothers of fetus with T21.

Pregnancy-associated plasma protein A (PAPP-A) is another hormone that is tested. PAPP-A is produced by the covering of the newly fertilized egg. In the first trimester, low levels of this protein are seen in pregnancies carrying a fetus with T21.

Amniocentesis

This procedure is used to collect amniotic fluid, the liquid that is in the womb. It is usually performed in a hospital setting as an "out-patient." A needle is inserted through the mother's abdomen into the uterus. Ultrasound is used to guide the needle. An ounce of fluid is taken for testing. This fluid contains fetal cells that can be examined for chromosome testing. The test can take from two to five weeks.

Amniocentesis is usually performed between the fourteenth and eighteenth week of pregnancy. Side effects are cramping, bleeding, infection, and leaking of amniotic fluid. There is a slight increase in the risk of miscarriage. Amniocentesis in not recommended before the fourteenth week of pregnancy. Amniocentesis prior to the fourteenth week may increase the risk of complication and miscarriage.

If a woman has a risk of having a child with T21 of one in 250 or greater, amniocentesis is usually offered.

Chronic Health Concerns

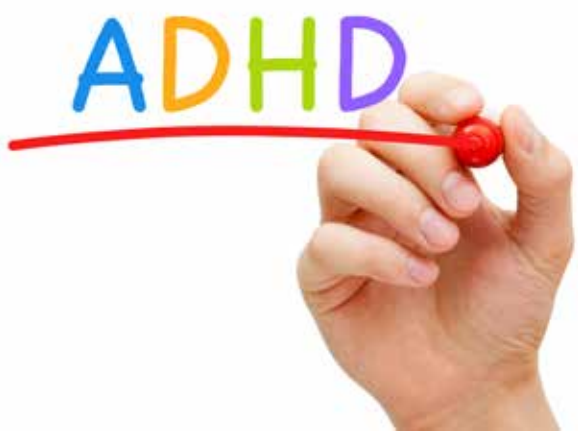
Individuals with T21 may have concerns with the following chronic health conditions. Some children may also have dual diagnosis (be diagnosed with Down syndrome and another diagnosis such as ADHD or autism).[5]

ADHD (Attention Deficit, Hyperactive Disorder)

The diagnosis of ADHD is made by a psychiatrist. A child may have problems with concentration or paying attention (attention deficit), may be very active (hyperactivity), or may act before thinking (impulsivity). There is no blood test or truly objective way of agreeing that a child has ADHD. Around three to eight percent of children may have ADHD in the USA. Boys are about three times more likely to have ADHD than girls. Some research indicates that there may be a genetic component to the condition.[6]

ADHD can be divided into three subtypes:

- inattentive type, where attention and staying on task is the main problem;
- hyperactive-impulsive type, where the child is very active and often acts without thinking; and
- combined type, where the child is inattentive, impulsive, and too active.



A diagnosis of ADHD may be difficult to make in a child with T21.

Autism

In rare cases, a child with T21 may also have a diagnosis of autism. Autism is a spectrum disorder (ASD). It may be mild or severe. Many of the symptoms overlap with other conditions such as obsessive compulsive disorder (OCD) or ADHD. ASD is a developmental diagnosis: Expression of the syndrome varies with a child's age and developmental level. Autism is a life-long diagnosis. During infancy or toddler years, the child may display:

- repetitive motor behaviours (fingers in mouth, hand flapping);
- fascination with and staring at lights, ceiling fans, or fingers;
- extreme food refusal—limited variety of food intake,
- receptive language problems (poor understanding and use of gestures), possible giving the appearance the child does not hear; and
- spoken language may be highly repetitive or absent.

The most commonly described areas of concern for children with ASD include:

- communication (using and understanding spoken words or signs);
- social skills (relating to people and social circumstances); and
- repetitive body movements or behaviour patterns.

The incidence of a dual diagnosis of T21 and ASD is between one and ten percent. The occurrence of trisomy 21 lowers the threshold for the appearance of ASD in some children. This may be due to other genetic or other biological influences on brain development.[7]

Leukemia

One percent of individuals with T21 can develop leukemia. This is a type of cancer that affects the white blood cells in the body. Symptoms of leukemia include easy bruising, tiredness, paleness, and unexplained fevers. Although leukemia is a serious disease, around ninety percent of children that develop leukemia survive. The treatment of leukemia involves chemotherapy or radiation. Occasionally, a bone-marrow transplant is performed.

Nutrients including antioxidants, B-complex vitamins, omega-3 fatty acids, and methyl-donor amino acids have been shown to be effective in the treatment of leukemia.[8]

Alzheimer's Disease

Alzheimer's disease (AD) is the most common form of dementia (loss of short and eventually long-term memory). This disease is progressive, and the brain degenerates. Alzheimer's disease is associated with old age; however, it is not considered a normal part of aging.

Individuals with T21 develop a syndrome of dementia that has the same characteristics as AD.[9] Individuals with T21 will develop these symptoms in their late 40s or early 50s. AD is normally diagnosed in individuals who are 60 to 70 years of age. Most individuals with T21 develop the brain changes associated with AD. An estimated ten to twenty-five percent of individuals with T21 have AD at the age of 40 to 49, twenty to fifty percent have AD at the age of 50 to 50, and sixty to seventy five percent have AD when they are older than 60 years of age.

The reason AD is more common in people with T21 is not completely known. AD is associated with increased production of a compound called amyloid *beta* in the brain. Amyloid *beta* builds up and causes the loss of brain cells (neurons). How the neuron loss occurs is not well understood. The higher risk for AD is individuals with T21 may be related to the extra copy of chromosome 21. The gene that codes for amyloid *beta* is found on chromosome 21. Individuals with T21 have three copies of this gene compared to the normal population.

The age when symptoms of AD develop may be related to a person's mental capacity (cognitive reserve). This means that people with greater brain weight, more brain cells (neurons), and more education may not have symptoms of AD as early as people with less cognitive reserve. Because of this, individuals with T21 need to be encouraged to pursue higher education, and supplement with nutrients that encourage neuron production.

The main symptoms are confusion, disorientation, and wandering. These early signs are not usually recognized and are commonly misdiagnosed.

Behaviour changes also occur. Early behaviour changes that are truly related to AD are often seen as an exaggeration of the person's normal traits, e.g. the person may refuse to follow certain directions or to do chores. This refusal may be perceived as stubbornness.

Because the early changes are hard to recognize, only those familiar with the individual notice these changes. Changes can include changes in daily routine, change in sleeping or eating habits, inability to make decisions about clothing, getting lost in familiar environments, and inability to remember the names of familiar people. Another early sign of AD in highly functional individuals with T21 is the inability to perform job duties.

Visual problems can develop in the early stages of AD. Combining these visual problems with the cognitive and memory deficits will put individuals with T21 at greater risk for the following activities: get lost in familiar environments, may not be able to perform certain activities, may have accidents and falls, and may have difficulty with learning new tasks.

The ability to learn new tasks will be impaired. It may be more difficult to demonstrate new activities to an individual with T21 and AD.

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