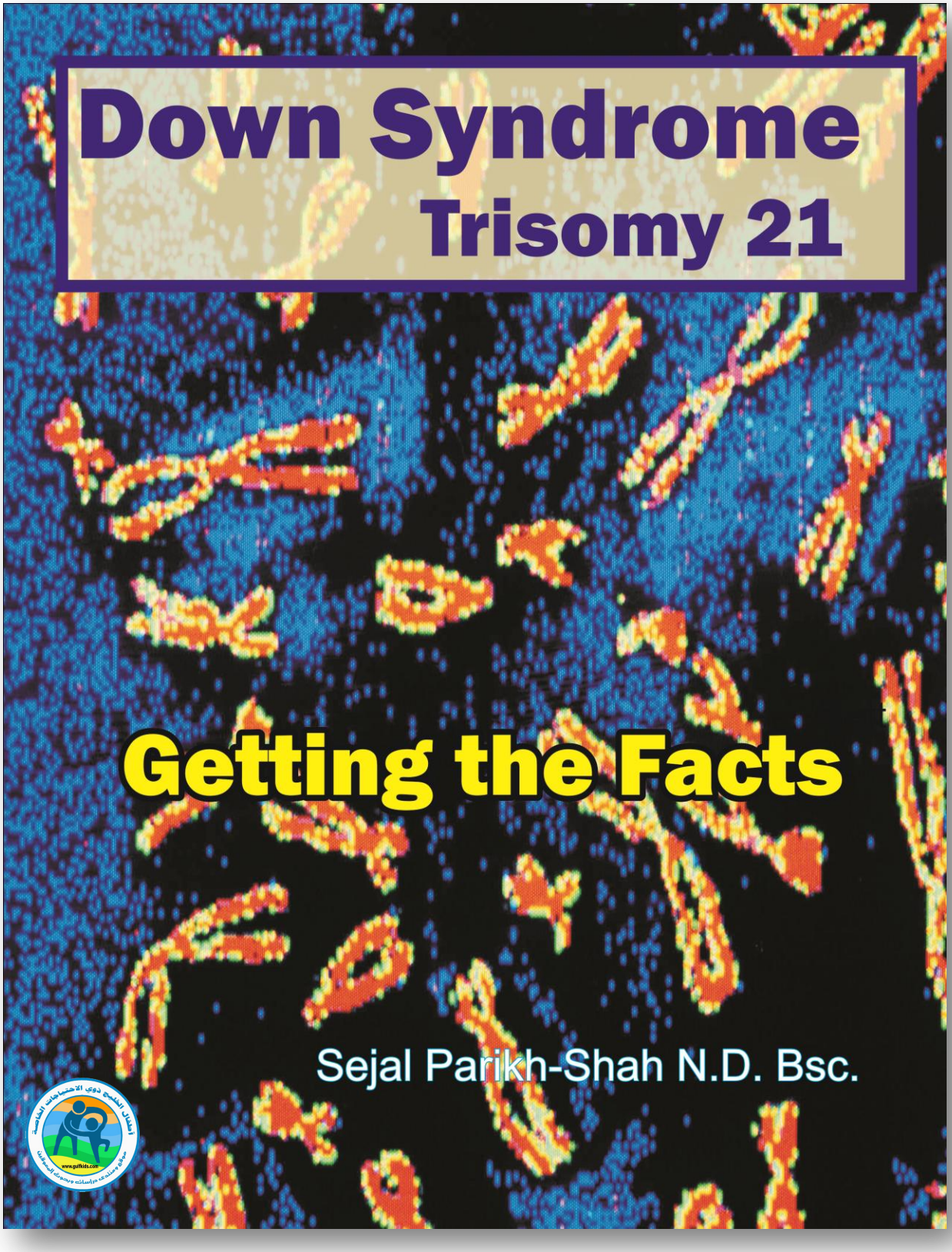
Down Syndrome
Trisomy 21



Getting the Facts

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Down Syndrome (trisomy 21)

Introduction

Down syndrome received its name in 1866 when John Langdon Down initially described this disorder. Down syndrome is more recently known as trisomy 21 which better describes the extra 21st chromosome that is distinctive for 95 percent of individuals with this genetic disorder.

Many individuals and groups prefer the later terminology as it has less of a negative connotation than using the word “Down” and “Syndrome.”

Today, people with Trisomy 21 (T21) enjoy a dramatically better life style and health profile than those of just a few decades ago. This can be attributed to better research, advancements in medicine and greater ability to share information by individuals and societies. It’s also due to greater social responsibility by governments and other organizations.

T21 is more common than most people are aware. It is estimated that 1 in 800 births worldwide are to infants with T21. This number may be significantly higher if genetic testing was not used to identify fetuses with T21 who are more often than not terminated. The high rate of termination is due partly to the very short window in which parents are to make this decision. Often the fetus is past a point at which it is old enough to be considered living and must be provided with a proper funeral making this an even more traumatic time for the parents. Stereotypes of T21 kids from years past often play in the decision of parents as well as the advice of the hospital staff.

People with T21 are generally characterized as having a combination of impairment in cognition and physical growth along with poorer health and distinctive physical characteristics.

Most individuals have 2 pairs of 23 chromosomes. In individuals with T21, they can have up to 3 copies of chromosome 21. Individuals with T21 can present with one of three types of chromosomal patterns. Ninety-five percent of individuals with T21 have true trisomy 21, this means that they have 3 copies of chromosome 21. Two to three percent of individuals with T21 have a translocation disorder. This means that part of chromosome 21 breaks off and attaches itself to another chromosome, usually chromosome 14. Two-thirds of these

translocations are spontaneous. The rest are inherited from a parent. In individuals with translocation, screening the parents may be of interest to determine whether future pregnancies may result in another pregnancy with a child with T21.

Another 2 percent of individuals with T21 have mosaicism. In this case, there is an error with cellular division. After conception, some cells have 46 chromosomes while others have 47 chromosomes. The percent of cells with 46 chromosomes tends to vary from person to person.

Common Physical Characteristics

Many of the common physical features of T21 may also appear in the general population. Individuals with T21 have some common facial features. Almond shaped eyes due to the presence of epicantal folds (a folding of the skin of the upper eyelid extending from the nose to inner side of the eyebrow) and up slanting palpebral fissures (the separation between the upper and lower eyelids). Light-coloured spots in the eyes, called Brushfield spots, may be present. A small, somewhat flat nose with a protruding tongue (macroglossia) and small ears may also be present. A small chin (microgenia) in an unusually round face may be present. They may also have a smaller head that is somewhat flattened in the back (brachycephaly) and straight hair that is fine and thin.

Other physical features include a single crease across the palms of their hands (Simian crease), short stubby fingers and a fifth finger or pinky that curves inward (clinodactyly). In general they have shorter stature with short limbs. They may have poor muscle tone and a larger than normal space between the big and second toes.

None of these facial or physical features are abnormal by themselves, nor would they cause a baby with T21 any problems. If a doctor sees a number of these features together in one baby, they may suspect that the baby to have T21. These facial and physical features are what cause people with T21 to resemble one another, although they also resemble their own families.

Common Health Concerns

Children with T21 may face health complications beyond the normal childhood illnesses. There are a few congenial (present at birth) malformations that are common to babies with T21. More than 40 percent of children born with T21 have a congenital heart malformation. Because of this high incidence, most babies with T21 should be screened with an echocardiogram at birth. The presence of

a heart problem does not mean that the baby will be seriously ill. The extent of the heart problems will vary greatly. The management of heart defects will vary from watchful waiting to medication to surgery.

Around ten percent of infants with T21 will have gastrointestinal (stomach/digestion) issues. These can range from narrowing or blockage of the intestine (duodenal atresia), an absent anal opening (anal atresia), an obstruction of the outlet of the stomach (pyloric stenosis) or an absence of the nerves in the colon called Hirschsprung's disease. Most of these malformations need to be fixed with surgery.

Hypothyroidism is more common in children with T21. Children with T21 should have regular blood tests to check thyroid function. The thyroid is a small gland located in the neck. Hypothyroidism is when the body makes too little thyroid hormone. Thyroid hormone is necessary to a variety of bodily functions. Metabolism, bowel movements, temperature and cognition are affected by low levels of thyroid hormone. Optimal thyroid function is also important for growth and muscle function. Testing thyroid hormone levels should be done a few times in the first couple of years of life and then annually. Treatment of hypothyroidism is usually in the form of replacement thyroid hormone.

Preventive medical care is important for all individuals. It is especially important for individuals with T21. Working together with a conventional medical doctor and a naturopathic doctor we can enhance the potential of individuals with T21.

The use of nutritional supplementation, especially New Downs in the daily life of a child with T21 can help with the long term health of individual.

Fine motor skills may also be slower. The fine motor helps kids pick up and manipulate toys and objects. Self feeding, drinking from a cup, self dressing are examples of fine motor skills. Writing skills develop as a refinement of the above mentioned skills. These skills are necessary for both learning and self help.

Minerals such as calcium, magnesium and omega 3 fatty acids can be beneficial for increasing fine motor skills. These minerals are found in therapeutic levels in New Downs.

Social development

Kids with T21 usually have age appropriate social skills. Unless there is a dual diagnosis (with autism), children with T21 will interact well with other kids of same age. Encourage social interaction with other children. Because of the T21 diagnosis, some parents may limit social interactions. Do not underestimate their ability to behave in an age appropriate way because of their size or language delays. Kids of all ages require and thrive with interactions with their peers.

It is very beneficial for all kids to interact with a variety of children of different ages and a wide range of adults. This encourages communication skills and social behaviour. A parent and tot play group or preschool situation will have develop independence and social interactions. This will also encourage play skills and cognition. Do not feel that the language delay will prevent the child from interacting with others or watching and imitating other children.

Being in a preschool or play group will also encourage turn taking, to share, how to follow instructions from teachers and have appropriate behaviour. All kids go through the ‘terrible two’. This is when children go though a period of tantrums as they want to be independent and not conform. Luckily, all kids out grow this behaviour, latest by 3-4 years of age. Having a child with T21 mainstreamed will aid in age appropriate behaviour.

Self-help skills and independence will develop in all children. During preschool years, children become independent. They are able to self feed, self dress and use the bathroom. These skills develop in children with T21. Most children with T21 will be able to achieve self-help skills by the time they are 4-6 years of age. The speed with which children become independent in these areas is influenced by the expectations of their parents.

Cognition

Cognition includes learning and mental processing; thinking, reasoning, remembering and learning skills. In typical development, speech and language skills play a central role. Imaginative play is an

important part of cognition. This is where the child thinks out loud. Language skills are practiced and social interactions are modeled.

Cognitive development also refers to acquiring knowledge about the world and understanding the social and physical world. Knowledge is obtained via the senses. Babies watch all activities around them. Exploration with toy and objects within their reach encourages cognition. The way in which a baby or young child plays with toys is usually a good indication of their level of understanding. For instance, is s/he using the toy appropriately? Is s/he stacking blocks? Is s/he opening up a book and pretending to read it?

Imaginative play starts in the second year. This is when they begin to show how they understand their world. They act out behaviours and actions of those around them. They may pretend to make meals, put a doll to bed, drive a car, and go grocery shopping. This type of play is very important. This type of play can be used to teach many things. Vocabulary can be introduced, demonstrated, repeated and learned through imaginative play. An example would be matching and selecting pictures. Children can learn the names of animals and common household items. Colours and numbers can also be introduced and taught. Structured teaching can also help children to follow instructions.

During daily activities, adults can help children to reach the next step in play and understanding their environment. Modeling (showing children what to do and explaining activities) is very educational. Folding clothes, loading a dishwasher, combining ingredients for a cake can be fun and educational activities for all children.

In the first two years of life, knowledge is acquired by a child's ability to play with toys. This will encourage an understanding of their world around them. This knowledge will be displayed through imitative and imaginative play. As language increases, the child will learn more about the characteristics of objects and events in their world. Rather than say ball, s/he will say big, red ball. Action words are also learned: running, swimming, washing. Object placement is also learned at this time; on, under, behind.

Speech and language development is linking to cognition. In children who have speech delays, it is important to recognize the impact of this and still try to teach as many concepts as possible with toys and real

objects. This is why play activities and giving opportunities for interaction is very important for children with T21.

By preschool (3-5 years of age), children start to learn to count and start to gain wide experiences of books. They are ready to learn and read. Fine motor skills will also start developing now. They will start to gain control of writing, by colouring and drawing. Children with T21 can begin to learn all these same things. The acquisition of these skills may be at a slower pace.

Nutrients such as B-complex, amino acids, magnesium and omega 3 fatty acids are vital to the development of cognition. New Downs contains there nutrients in therapeutic doses.

Attention and memory

Children have to look and listen or touch things in their world in order to gain knowledge of their world. They also need to attend long enough to take in the information and to remember it. If attention is not present, a memory of the object or action will not be retained.

Attention

Most children with T21 do not have any attention difficulties. Engagement of children in activities which require them to pay attention is important. Using play activities and picture book reading to engage children's attention is very important. It is important to encourage babies to engage in early 'face-to-face' babble games and to continue from this to playing with toys and looking at books. When a child is able to see an adult's face with babble games and reading, they can visualize the facial expression and will start to imitate these expressions.

If a child's attention is limited, it is helpful to find activities that they enjoy. Noisy toys or toys with moving parts can be motivating. Turn taking will help keep these kids engaged. Looking at books is a good way to move towards a sitting still and formal learning experience. To encourage a child to sit at the table, choose activities that are fun, that the child enjoys. A table activity involving other children and adults can be motivating for children who have difficulty with attention.

Attention and motivation are usually linked. A child will pay attention and concentrate for longer periods when the activity is one that they enjoy.

Memory

Memory can be divided into 2 types; long term and short term. Long term memory refers to all the information and learned skills that can be accessed. Short term memory refers to the recall of information that has been retained for a short period of time; remembering a telephone number while dialing it. Children with T21 have adequate long term memory. Information and skills once learned are retained once learned. Short term memory (working memory) does not develop at the expected rate. There is a difficulty with short term memory especially when it is verbal information. This makes learning to talk and process speech in everyday situations particularly difficult for most children with T21. However, their ability to process visual and spatial information in short-term memory is better. As a result, the use of pictures and visual information to supplement spoken information is very important.

Speech

Speech typically starts developing at 12 months of age. All babies with T21 benefit from speech and language therapist from birth. Even though words begin in the second year of life, the foundation of non-verbal communication skills and babble starts much earlier. Babies begin to understand the words used around them and will start to point to objects around them around 9 months of age. Babies with T21 may have hearing difficulties and the use of signing can be helpful. Parents can utilize sign language starting at 9 months of age.

Babble is a very important step towards speech. It starts developing in the first year of life. In the second year, babies begin using single words and then join words together. Babies with T21 can often sign words before they can say them. Working with a speech and language therapist can help encourage articulation and production of words and sentences. This also needs to be practiced at home with all members of the household.

Nutrients such as B complex, minerals such as calcium and magnesium and omega 3 fatty acids are vital for the development of speech. New Downs contains these nutrients in therapeutic levels.

Learning Potential

The learning potential in individuals with T21 can be maximized through early intervention, good education, higher expectations, encouragement and optimal nutritional supplementation.

Muscle tone

Almost all infants with T21 will have some degree of low muscle tone (hypotonia). Low muscle tone affects all muscle groups. Gross motor and fine motor skills are affected due to hypotonia as in speech. Some infants with T21 have more hypotonia which may make them look weak or 'floppy'. Hypotonia will affect a child's ability to learn and grow. It may make it difficult to learn to roll over, sit-up, crawl, stand and speak. In infants, low muscle tone can also affect feeding. Some infants with T21 may have a difficult time with breastfeeding. This does not mean that all infants with T21 do not breastfeed; it means that it may take them longer to breastfeed.

Delays in motor milestones are usually due to hypotonia. As the child grows the hypotonia decreases. Working with a physical therapist or physiotherapist will greatly help increase the muscle tone. The development of gross motor skills aid in the development of independence and as a result will influence social, language and cognitive development.

Minerals are important for muscle tone. New Downs contains minerals in therapeutic doses to aid in muscle tone development.

Vision

About seventy percent of children with T21 will have vision problems. Nearsightedness (myopia), farsightedness (hypermetropia), crossed-eyes (strabismus) is 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 above mentioned conditions are easily correctable.

Antioxidants such as vitamin A and the carotonoids are vital for the health of the visual system. New Downs contains therapeutic doses of the above mentioned nutrients.

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 small head 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 check-ups should include an otoscopic examination (visualization of the ear canals and the tympanic membrane, to ensure that there is no infection present) and hearing examination.

Chronic Health Concerns

Individuals with T21 may have concerns with the following chronic health conditions.

ADHD (Attention Deficit, Hyperactive Disorder)

The diagnosis of ADHD is made by a psychiatrist. A child may have problems with concentrating 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.

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
- 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. In a typical child, a judgment is made as to whether the child is more inattentive, active or impulsive than would be expected for his or he age. In a child with T21, allowances need to be made for the developmental delay and speech and language delay.

New Downs contains nutritional supplements that can help to decrease the incidence and intensity of ADHD symptoms. Nutrients including B-complex, Magnesium, omega 3 fatty acids and methyl donor amino acids have been shown to be effective in the treatment of ADHD.

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. Children with ASD display 'atypical' behaviour. 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
- 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),
- repetitive body movements or behaviour patterns

Children who have ASD may or may not exhibit all of these characteristics at any one time nor will they consistently demonstrate their abilities across similar circumstances. The following are some characteristics of ASD common with kids with T21-ASD:

- unusual response to sensations (especially sounds, lights, touch or pain,
- food refusal (preferred textures or tastes),
- difficulty with changes in routine or familiar surroundings,
- little or no meaningful communication,
- disruptive behaviours (aggression, throwing tantrum, or extreme non-compliance),
- hyperactivity, short attention, and impulsivity,
- sleep disturbances,
- history of developmental regression (especially language and social skills,
- unusual play with toys and other objects,

- self-injurious behaviour (skin picking, head hitting or banging, eye poking, or biting).

The incidence of a dual diagnosis of T21 and ASD is between 1 and 10%. 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.

New Downs contains nutritional supplements that can help to decrease the incidence and intensity of AD symptoms. Nutrients including methyl cobalamin, folinic acid, B-complex, omega 3 fatty acids and methyl donor amino acids have been shown to be effective in the treatment of AD.

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.

New Downs contains nutritional supplements that can help to decrease the incidence of leukemia. Nutrients including anti-oxidants, B-complex, omega 3 fatty acids and methyl donor amino acids have been shown to be effective in the treatment of leukemia.

Alzheimer's disease

Alzheimer's disease (AD) is the most common form of dementia (loss of short and eventual 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 of AD. Individuals with T21 will develop these symptoms in their late forties or early fifties. AD is normally diagnosed in individuals who are sixty to seventy 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 forty to forty-nine. Twenty to fifty percent have AD at the age of fifty to fifty-nine, and sixty to seventy five percent have AD when they are older than sixty 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.

In people with T21, the first symptom of AD develops at the age of 50. The disease is usually diagnosed by the age of 52. Death occurs at an average of 60 years. The time from the first symptoms of AD to death is usually about 9 years.

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, i.e. 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.

Other early signs include loss of language and other communication skills, impairment of social skills and progressive loss of 'activities of daily living' (personal hygiene, dining skills, and bathroom skills).

As AD progresses to the middle age, the individual may totally depend on others for activities such as dressing, eating, walking and toilet needs. Communication also reduces. If behaviour problems are present, they may be exaggerated and psychotic behaviour may develop. Social activities may be reduced to a minimum.

As AD advances, the individual may seem to be in a coma; totally depend on others and may interact minimally with the environment.

Physical symptoms of AD may be observed in the early stage but become obvious in the middle stage of the disease. Walking becomes difficult and in the advanced stage, the person is confined to bed and has almost no voluntary movements. Eating disorders may be observed at the beginning of the disease but are more obvious in the middle stage. The person may have problems swallowing and may frequently choke.

New Downs contains nutritional supplements that can help to decrease the incidence and intensity of Alzheimer's symptoms. Nutrients including anti-oxidants, B-complex, omega 3 fatty acids and methyl donor amino acids have been shown to be effective in the treatment of Alzheimer's disease. In adults, the use of naturopathic medicine, utilizing annual detoxification can be helpful in prevention of Alzheimer's disease.

Prenatal Screening

Between the 15th and 18th 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.

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.

PAPP-A is another hormone that is tested. It is pregnancy-associated plasma protein A. PAPP-A is produced 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. One ounce of fluid is taken for

testing. This fluid contains fetal cells that can be examined for chromosome testing. The test can take from 2-5 weeks.

Amniocentesis is usually performed between the 14th and 18th week of pregnancy. Side effects are cramping, bleeding, infection and leaking of amniotic fluid. There is a slight increase in risk of miscarriage. Amniocentesis in not recommended before the 14th week of pregnancy. Amniocentesis prior to the 14th week may increase the risk of complication and miscarriage.

If a woman has a risk of having a child with T21 of 1 in 250 or greater, amniocentesis is usually offered.

Jai's story

We had 2 children before I became pregnant with Jai. Anjali was 8 and Tarun was 6, when we learned about Jai's arrival. The pregnancy was somewhat different than my other two pregnancies. A little less movement, a bit more fatigue. I had just turned 35 when I become pregnant with Jai.

The antenatal visits were always normal. We performed the maternal serum test with this pregnancy as we had done with the other two pregnancies. I never gave it a second thought.

I was in my office at the naturopathic college when I received the call from Judy, my midwife. She told me that the maternal serum testing identified me as having a risk of 1 in 214 whereas, women my age, the risk was 1 in 250 for carrying a child with Down Syndrome. This doesn't seem like a huge difference, but call it woman's intuition, I needed to know.

We scheduled an appointment for amniocentesis. The four weeks that followed were the hardest four weeks of our lives. The waiting and not knowing was very difficult. Friday evening, Judy called me at home, confirming the Down Syndrome diagnosis. We were devastated, now what? We were given the option of carrying through with the pregnancy or terminating it. Please realize that growing up, we did not know anyone with special needs. Not knowing what to expect and being thrown into genetic counseling (which pretty much told us to terminate the pregnancy) was grueling. I thought the waiting for the amniocentesis results was difficult, trying to make a decision about the pregnancy was even worse. The thought about terminating a pregnancy was never one that we would ever consider, but we were given this option.

The decision to keep the pregnancy was made by my daughter. When I was at my lowest, Anjali came to me and gave me and my belly a huge hug. She said that she loved me and loved the baby. At that moment, the decision was made, how I could take her sibling away from her.

Once the decision to keep the pregnancy had been made, my quest for knowledge began. I needed to do everything possible to ensure that my child will be healthy.

I spent hours researching about Down Syndrome. I asked colleagues, my students, friends for information about Down Syndrome. I became a sponge. People came out from everywhere offering support, knowledge and love. We were overwhelmed. It felt as if everyone that we encountered from then on only had best wishes for us.

I started taking supplements during my pregnancy, more antioxidants, more B complex, more protein. I started seeing a colleague who prescribed a homeopathic remedy. My father-in-law encouraged me to start meditation.

The pregnancy after this was uneventful, life carried on. My due date was January 25th. Anjali's birthday is February 8th. We decided to have her birthday party 3 weeks before the due date. My other two pregnancies were 2 weeks early, I thought that Jai would be similar. Anjali's birthday party was scheduled on January 10th, a sleepover. Little did we know that Jai was planning on crashing on the birthday party. My water broke at noon on January 10th and the birthday party was canceled. Jai was born in the early hours of January 11. Because we were carrying a child with a diagnosis of Down Syndrome, a neonatologist was consulted. He took a look at Jai and questioned the DS diagnosis. Jai had no physical features of an infant with DS. He started nursing with no problem. We were asked to see a neonatal cardiologist within a few days to have an echocardiogram to ensure that there were no problems with his heart.

When we brought Jai home, I apologized to Anjali for canceling her birthday party. She replied that she had the best birthday present that she could ever ask for, a baby brother.

The first few months were quite normal for our family of five. Jai was eating, sleeping, interacting like any newborn. We registered him with Early Intervention. With this we had access to physiotherapists who helped develop exercise routines. Things that would help us help Jai to sit, crawl, walk, and self feed.

Through my research, I started increasing my supplements and started introducing probiotics with Jai's feedings. We were also exposed to

Medec, a form of physiotherapy that helps increase core strength in infants who have low muscle tone.

As time progressed, Jai started sitting up, rolling over, crawling and walking. He achieved all these milestones within the normal ranges. As time continued, I started incorporating supplements into his food, trying to reach therapeutic doses with different supplements. My kitchen looked like a pharmacy with all types of vitamins, minerals and protein powders.

It was taking quite a bit of time to administer all the various forms of supplements. I was not satisfied with the dosages of each nutrient that I was giving him. I was not happy with the form or the quantity. This is when I started to formulate my ideal supplement for children with DS. The idea of Newdowns emerged.

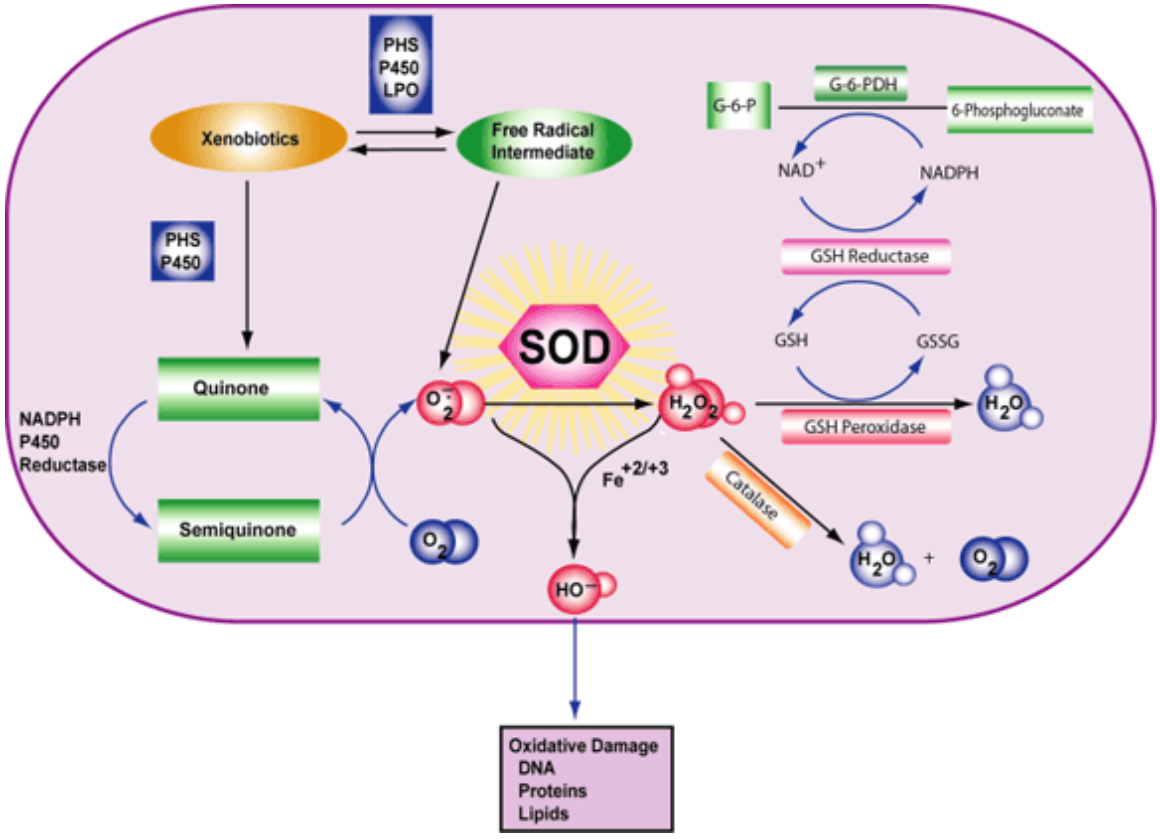
We started meeting with nutraceutical companies. We interviewed various companies to enquire whether they could manufacture newdowns. We finally decided on our current manufacturer. They are an ISO90001 company based in the US. They were able to organize and obtain the correct forms and dosages of the vitamins, minerals and amino acids that I wanted in my formula.

When Jai turned 3, Anjali started teaching him to swim. Currently, she is an assistant swim instructor. Over the summer, Jai went from swimming with a life jacket to jumping off the diving board without a life jacket. Of course, there is always an adult close to him in the pool when he jumps. See our u-tube video.

Currently, Jai is registered in senior kindergarten. He has quite a few friends and interacting and learning alongside his peers.

The New Downs Formula

The vitamin/mineral has the maximum amount of antioxidants specific for Down's syndrome. Individuals with Down's syndrome have a greater need for antioxidants because of the extra chromosome 21. This chromosome carries the gene for superoxide dismutase (SOD) which decreases the free radical, singlet oxygen (O*). When there are 3 copies of the SOD gene, there is more hydrogen peroxide molecules generated which the body can not handle. (See diagram below) This leads to faster depletion of glutathione and other antioxidants. This leads to faster aging, decreased cognition, decreased growth, increased risk of cancers, Alzheimer's and Parkinson's. An elevation of SOD activity interferes with the transport of biogenic amines into chromaffin granules. Since neurotransmitter uptake plays an important role in many processes of the central nervous system, SOD gene-dosage may contribute to the neurobiological abnormalities of Down's syndrome.



Cell damage is induced by reactive oxygen species (ROS). ROS are either free radicals, reactive anions containing oxygen atoms, or molecules containing oxygen atoms that can either produce free radicals or are chemically activated by them. Examples are hydroxyl radical, superoxide, hydrogen peroxide, and peroxynitrite. The main source of ROS *in vivo* is aerobic respiration, although ROS are also produced by peroxisomal β-oxidation of fatty acids, microsomal cytochrome P450 metabolism of xenobiotic compounds, stimulation of phagocytosis by pathogens or lipopolysaccharides, arginine metabolism, and tissue specific enzymes.

Under normal conditions, ROS are cleared from the cell by the action of superoxide dismutase (SOD), catalase, or glutathione (GSH) peroxidase. The main damage to cells results from the ROS-induced alteration of macromolecules such as polyunsaturated fatty acids in membrane lipids, essential proteins, and DNA. Additionally, oxidative stress and ROS have been implicated in disease states, such as Alzheimer's disease, Parkinson’s disease, cancer, and aging.

Superoxide Dismutase (SOD) catalyzes the reduction of superoxide anions to hydrogen peroxide.

Excessive hydrogen peroxide results in a reduction in glutathione levels. As cellular GSH is depleted, first individual cells die in those areas most affected. Then zones of tissue damage begin to appear; those tissues with the highest content of polyunsaturated lipids and/or the most meager antioxidant defenses are generally the most vulnerable. Localized free-radical damage spreads across the tissue in an ever-widening, self-propagating wave. If this spreading wave of tissue degeneration is to be halted, the antioxidant defenses must be augmented. Repletion of glutathione appears to be central to intrinsic adaptive strategies for meeting the challenge of sustained (or acute) oxidative stress.

An enzyme called cystathionine-beta-synethase is found on the 21st chromosome; the increased activity of this enzyme drives the SAM cycle away from normal methylation processes

Increase growth to realize best potential height

Individuals with Down syndrome have a separate growth chart. The shorter height may be due to decreased thyroid function and/or poor intake or absorption of nutrition. When a good quality nutritional supplement, such as New Downs is incorporated into a dietary regime, growth can increase.

Support complete digestive cycle

Digestive issues are common in people with Down syndrome. The symptoms can be constipation, weight gain or food intolerances. The high quality of ingredients in this formulation ensures that the supplement is easily absorbed by the digestive system. Once these nutritional supplements are absorbed, they can be delivered to the organ system that requires them.

Boost immune system defense

Upper respiratory infections and ear infections are common in individuals with Down syndrome. Immune supporting nutrients such as: vitamin C, magnesium, zinc and carotenoids will provide the necessary tools for the immune system to help prevent/treat these infections.

Accelerate cognitive development

Intellect, speech, social skills are all aspects of cognition. DHA and EPA have been shown to enhance aspects of cognition.

Enhance nervous system

Gross motor, fine motor and speech are all aspects of the nervous system. Nutrients that support the nervous system, are folinic acid, methylcobalamin, zinc, magnesium, DHA and EPA are an integral part of the New Downs formulation.

Delay the onset of neurological diseases

Individuals with Down syndrome are at risk for early aging and early onset of neurological diseases such as Alzheimer's and Parkinson's. By supporting the nervous system, with the above mentioned nutrients, New Downs will provide the necessary tools to prevent the early onset of these neurological diseases.

Vitamins and Minerals and their Purpose

Vitamin A and carotenoids:

This group of vitamins are essential as an antioxidant. Their purpose is to aid detoxification via the hepatic P450 pathway and aid the immune system. Individuals with Down Syndrome (DS) have an increased risk of upper respiratory tract infections. In infants and children, this is especially important to avoid since, every time a child gets sick, s/he decreases their ability to learn at that period. Individuals with DS also have difficulty with their digestive system. Vitamin A supports the microvilli in the digestive system, helping with new growth of the microvilli.

This group of vitamins are also necessary for the health of the thyroid gland. Many individuals with DS also have a tendency towards hypothyroidism. This condition predisposes them to constipation, weight gain, fatigue, decreased mental alertness.

B-complex:

vitamins B1, B2, B3, B5, B6, B12, biotin, choline and folinic acid are especially important for energy production. These vitamins work together as a group. B3, B5, B6, folinic acid and B12 (methylcobalamin) is especially important for stress management and the detoxification pathway and recycling of glutathione. Folinic acid and methylcobalamin are necessary for maintenance of the methyl pool which is necessary for the recycling of glutathione and maintain the proper activity of SAM cycle. These two vitamins are also needed for neurological development and prevention of free radical damage to the brain. They are especially important for the prevention of Alzheimer's and Parkinson's. Biotin and choline are necessary for neurotransmitter synthesis and fatty acid metabolism.

Vitamin C:

This vitamin is necessary for all aspects of detoxification, immune function, stress management and the regeneration of healthy tissue. Vitamin C is needed for the glutathione regeneration, maintenance of the immune system, digestive system, endocrine and nervous system.

Vitamin D:

This vitamin is necessary for bone and connective tissue health. It is also an important vitamin necessary for the immune system. This vitamin is usually low in individuals who live above the 48th parallel. Vitamin D has recently been shown beneficial in the treatment of Alzheimer's and MS. Vitamin D is necessary for the normal placement of calcium into bones. Vitamin D has also been shown to have antioxidant properties.

Vitamin E:

This formula contains the vitamin E complex, not just alpha tocopherol. Vitamin E has been shown to be deficient in DS individuals. Vitamin E supplementation has been shown to be cardio protective and neuroprotective. Vitamin E is a powerful antioxidant.

CoQ10

Is added to this formula to decrease oxidative damage especially to the fatty acids. CoQ10 is necessary for the prevention of cardiovascular disease. Individuals with DS have a great risk of congenital cardiovascular malformation. CoQ10 helps to regenerate healthy cardiovascular tissue.

The Minerals

Iron:

This mineral is necessary for the energy production pathway. Individuals with DS tend to have lower intake of iron. Iron is necessary for memory and attention.

Manganese:

This mineral is involved in many enzymatic reactions. It is also involved with the neutralization of super oxide radicals.

Copper:

This mineral is involved with the iron in the formation of red blood cells, hence energy, mental alertness.

Molybdenum:

This trace mineral is necessary for the P450 pathway and red blood cell production

Selenium:

This trace mineral is necessary for proper thyroid hormone production. It is an antioxidant needed with vitamin E.

Zinc:

This mineral is necessary for DNA synthesis, i.e. growth. It is also works as an antioxidant, necessary for greater than 200 metabolic pathways. Zinc is necessary for energy production and regulation of glutathione recycling.

Iodine:

This trace mineral is necessary for proper thyroid function. Most individuals with DS have an underactive (subclinical) thyroid.

Calcium:

This mineral is needed for bone and connective tissue production, i.e. growth

Chloride:

Necessary for proper acid/base balance, decrease risk for inflammatory diseases.

Chromium:

Necessary for insulin regulation and weight management

Magnesium:

An important mineral involved in 100s of metabolic pathways. Necessary for detoxification and energy production.

Phosphorous:

Necessary for bone and connective tissue production.

Potassium:

necessary for proper acid/base balance

The amino acids:

Glutamine:

necessary for digestive system health. Food for the microvilli

Taurine:

necessary for heart health

Tyrosine:

necessary for thyroid hormone production

Methionine, Cysteine, Ornitine:

sulfur containing amino acids necessary for the detoxification pathways, maintenance of the methyl pool and glutathione recycling

Proline:

necessary for connective tissue production

Serine:

necessary for DNA synthesis, phosphotidylserine helps with cognitive function

Alpha-ketoglutaric acid:

necessary for the energy production pathway and urea cycle

Carnitine:

necessary for heart health and fatty acid metabolism

Lipoic acid:

antioxidant, scavenge reactive oxygen species and helps reduce glutathione

Glutathione:

antioxidant, scavenges reactive oxygen species, depleted due to increase SOD transcription with trisomy 21

Other nutrients:

- **Betaine HCl:** necessary for adequate digestion
- **Bromelain:** necessary for adequate digestion and decrease inflammation
- **DHA/EPA:** necessary for brain function