

III. What Do We Know? Medical Facts about Down Syndrome

What is Down Syndrome?

Down syndrome is the most common chromosomal aberration in human beings. Named after the English doctor John Langdon Down, who first described it in 1866, it is caused by an error in the cell division of the 21st chromosome. The exact causes of the chromosomal error in Down syndrome are currently unknown.

There are three chromosomal patterns that result in Down syndrome:

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|----------------------|---|---|
| Trisomy 21 | } | <ul style="list-style-type: none"> – also known as Nondisjunction Trisomy 21 – every cell in the body contains an extra 21st chromosome – occurs in 95 per cent of individuals with Down syndrome; |
| Translocation | } | <ul style="list-style-type: none"> – an extra piece of the 21st chromosome is attached – or “translocated” – to another chromosome – occurs in 3 to 4 per cent of individuals with Down syndrome; |
| Mosaicism | } | <ul style="list-style-type: none"> – is the result of an error in cell division soon after fertilization (in all other forms of Down syndrome, the error occurs at or before fertilization) – instead of having an extra 21st chromosome in every cell, individuals have 46 chromosomes in some cells and 47 in others – fewer of the typical physical features of Down syndrome may be evident.
(Rynders and Horrobin, 1996; Selikowitz, 1990) |

The incidence of Down syndrome in Canada is approximately **1 in every 700 live births** and is independent of the ethnic background, educational level or socio-economic status of the parents. More than 80 per cent of children with Down syndrome are born to women under the age of 35 with the average maternal age being 28 years. (Alberta Children’s Hospital, Calgary, Alberta, 1999)

For the majority of children with Down syndrome, the diagnosis is made at birth or shortly thereafter. Detection of the syndrome at this time allows for early intervention and aggressive management of associated health problems.



As in the general population, people with Down syndrome have a wide variation of cognitive, social and behavioural abilities and will develop their own unique personalities and talents.

What are the Associated Health Concerns?

The extra genetic material present in people with Down syndrome can cause specific medical conditions, some of which may influence a student's learning and participation in activities. It is incumbent for parents to share the necessary information about their child's health status with the school. In addition, teachers are obliged to ensure that the student's school file continually reflects current medical information. Changes in a student's behaviour or activity level at home or at school should be monitored as these could indicate a change in health status.

Although individuals with Down syndrome may have many of the following conditions, they can still be expected to lead relatively healthy, active lives.

Hypotonia. All individuals with Down syndrome have varying degrees of hypotonia or "low muscle tone" (i.e., muscle strength, stability and readiness for action) which impacts their fine and gross motor movements as well as balance and coordination skills. Due to hypotonia, developmental motor milestones (e.g., sitting, walking, toilet training, speaking, running) are reached at later ages than those of their peers. The impact of hypotonia can be diminished by regular exercise – through both gross and fine motor activities – as well as providing appropriate adaptive supports.

Heart. Approximately one-third of babies with Down syndrome are born with a congenital heart abnormality. Today, through pediatric cardiac surgery, most anomalies are corrected during the preschool years. Cardiac conditions can preclude a child to frequent chest infections and a decline in energy level. Regular checkups should be maintained until cardiac difficulties are ruled out. (Selikowitz, 1990)

Vision. The incidence of visual impairment is high for people with Down syndrome, so it is more common than not for them to need glasses. Yearly examinations by a pediatric ophthalmologist are recommended from four months of age until ten years. Thereafter, checks should be done every second year. The most common forms of visual difficulties include:

- Hypermetropia*** – medical term for long-sightedness
 - being able to see things at a distance but not up close
 - corrected by glasses;
- Myopia*** – medical term for shortsightedness
 - being able to see things up close but not at a distance
 - corrected by glasses;
- Squint*** – medical term for cross-eye
 - eyes may go out of alignment with one or both eyes converging inward to the nose
 - glasses, eye patch or surgery corrects muscle alignment;
- Nystagmus*** – eyes are unable to hold steady and move back and forth at a rapid rate
 - can reduce visual ability of the child
 - may be improved by glasses. (Selikowitz, 1990; Alberta Children's Hospital, Calgary, Alberta, 1999 information)

Hearing. Many people with Down syndrome experience hearing loss that can be temporary or permanent. Temporary fluctuating hearing losses are common and are usually brought on by ear infections caused by a smaller-than-normal middle ear and narrow eustachian tubes that block the drainage of fluid. Antibiotics and/or the insertion of drainage tubes will remedy most infections. However, if left unattended, temporary hearing losses can become permanent. Sensorineural hearing losses (i.e., congenital, permanent loss due to damage of the nerves or inner ear) are less common and a hearing aid may be prescribed. (Nicolosi, Harryman and Kresheck, 1989; Roizen, 1997 in the Down Syndrome Quarterly)

Early and accurate identification of hearing loss is essential to minimize the adverse effect on language comprehension, speech acquisition and learning ability. It's recommended that aggressive monitoring of hearing status occur every three months until three years of age, every six months until eight years of age, then once yearly throughout life. (Roizen, 1997)

Sinusitis and Upper Respiratory Infections. Individuals with Down syndrome are highly susceptible to upper respiratory infections and recurrent sinus infections and chest colds. Symptoms include a runny nose, watery eyes, chest congestion, coughing, fatigue and possible mood changes. A secondary characteristic of sinusitis may include intermittent teeth grinding which relieves the pressure and pain. These conditions do affect a student's stamina and overall health which can, in turn, impact his learning. Therefore, careful monitoring and prompt treatment of these conditions is necessary. (Rynders and Horrobin, 1996; Selikowitz, 1990)

Atlantoaxial Instability (AAI). This medical term describes an increased flexibility between the first and second vertebrae of the neck that increases the risk of spinal cord injury. Physicians recommend X-ray screening of all children with Down syndrome between three and four years of age and then re-screening at ten to twelve years. Although only one to two per cent of individuals with Down syndrome have symptoms that may require treatment, those with AAI should avoid activities that could put extra strain on the neck (i.e., gymnastics – especially somersaulting, tumbling and trampoline – diving, butterfly stroke, high jump, soccer and football). All children with Down syndrome should have yearly examinations to detect any signs of spinal cord compression. (Selikowitz, 1990)



Hypothyroidism. The thyroid is a tiny gland in the neck that secretes hormones into the blood that help regulate the body's use of sugar, fat and vitamins. Without this hormone, certain bodily functions suffer. Hypothyroidism occurs when the thyroid gland decreases or stops producing hormones; a condition present in up to thirty per cent of individuals with Down syndrome. (Trumble, 1993) Symptoms include decreased energy level, sleepiness, thickening of the skin, constipation, disproportionate weight gain and a slower rate of growth in height. Blood tests can reveal hypothyroidism before symptoms appear so should be done yearly. Hypothyroidism can be managed through oral medications. (Rynders and Horrobin, 1996; Selikowitz, 1990; Alberta Children's Hospital, Calgary, Alberta, 1999)

Diabetes. Diabetes is an endocrine disorder caused by the pancreas's failure to produce enough insulin; a hormone vital to carbohydrate metabolism. It is common in people with Down syndrome. The onset of diabetes in children is relatively quick and often presents itself through symptoms of persistent thirst, increased urination and weight loss. Diabetes is managed through the use of insulin and diet monitoring. (Rynders and Horrobin, 1996)

Dental. Usually, the onset of teeth in children with Down syndrome is delayed. But, once they've come in, hypotonia, which affects the muscles of the mouth, results in an imbalance of lip and tongue muscle pressure on those teeth. The tongue's influence is greater, thereby causing an open bite. This condition can be further aggravated by an oral cavity that is smaller than normal and by the side walls of the palate being thick. These factors, in addition to possible jaw misalignments or missing and malpositioned teeth, interfere with the articulation of sounds and the chewing of foods. They also negatively affect appearance. Periodontal disease (i.e., affecting the gums surrounding the tooth) is very common in teens and early, aggressive treatment is needed. Regular pediatric dental care, including orthodontics and possibly reconstructive surgery, is essential to minimize these complications. Finally, good dental care practices should be taught from an early age and monitored throughout adulthood. (Pilscher, 1998)

Obstructive Sleep Apnea (OSA). Sleep apnea is a condition where breathing is interrupted several times during sleep. This results in persistent daytime fatigue which can affect learning and participation. In people with Down syndrome, a narrowed throat area, hypotonia of the upper airway and enlarged adenoids and/or tonsils can cause obstructive sleep apnea. Usual treatment may include removal of adenoids and/or tonsils and/or use of "continuous positive airway pressure" (CPAP) which administers pressurized air through a mask during sleep. The most recent studies reveal that 45 percent of individuals with Down syndrome have OSA. (Dr. Len Leshin, MD, FAAP, 1997-8)

Weight Gain. A balanced diet and regular exercise ensures children have the energy to learn and be active participants. Yet dental complications, digestive problems and hypersensitivity in the mouth can make eating a balanced diet challenging. Furthermore, thyroid function as well as a sedentary lifestyle can cause weight gain during the teenage years. In addition to regular thyroid monitoring, provision should be made for fitness counseling and periodic consultations with a dietician. Likewise, an occupational or speech-language therapist is qualified to suggest ways to correct swallowing and eating disorders.

As society's attitude towards individuals with special needs evolves to one of value and respect, better health care is made available to them.

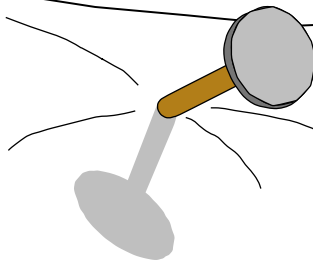
Today, the majority of people with Down syndrome grow up healthier and lead more active lives than previous generations.



Be Aware – Show You Care!

The purpose of the following chart is to help educators identify possible explanations for a student's behaviour. Significant behavioural changes may indicate a decline in health status and a need for further medical evaluation.

If you notice:	It could be a sign of:	And you should:
<i>Your student is lethargic and sleepy and may lack energy</i>	<ul style="list-style-type: none"> – hypothyroidism – low blood sugar – poor sleep habits 	<ul style="list-style-type: none"> – notify parents of behaviour – suggest a medical checkup
<i>Your student puts items very close to face to see</i>	<ul style="list-style-type: none"> – shortsightedness 	<ul style="list-style-type: none"> – notify parents of behaviour – suggest a vision check
<i>Your student's eye(s) turns inward</i>	<ul style="list-style-type: none"> – squint (cross-eye) 	<ul style="list-style-type: none"> – notify parents of observation – suggest a vision check
<i>Your student does not respond to loud noises or being called</i>	<ul style="list-style-type: none"> – fluid in middle ear – ear infection 	<ul style="list-style-type: none"> – notify parents of observation – suggest a hearing check – document when the student does not respond
<i>Your student is grumpy, has reddened ears and is rubbing ears</i>	<ul style="list-style-type: none"> – middle ear infection 	<ul style="list-style-type: none"> – notify parents of observation – document information – suggest a doctor's appointment and a hearing check
<i>Your student's nose is running and excretion is green or yellow; eyes are red; student is lethargic</i>	<ul style="list-style-type: none"> – sinus infection 	<ul style="list-style-type: none"> – notify parents of observation – suggest a medical checkup – send student home
<i>Your student is grinding teeth</i>	<ul style="list-style-type: none"> – sinus infection <i>or</i> – stress reaction 	<ul style="list-style-type: none"> – notify parents of behaviour – suggest a medical checkup



Someone once asked me after Murray was born if I was going to have another child so I could have a “real” boy! I wish they could now see my “real” seven-year-old boy in his grade 2 class. He has “real” friends who save him a seat at lunch so he can sit at their table. Friends from school and Beavers who drop by to play basketball and ball hockey and who invite him to movies, sleepovers and birthday parties. They invite him not because he is different or has Down syndrome, but because he is their peer and they can have fun together.

None of this would happen without full integration so he can be in class participating with his own age group. He learns from them and they learn from him. Other students see Murray working hard alongside them and they all celebrate their successes together. His peers have come to treat him as an equal who sometimes just needs a little more help. In gym and drama especially, classmates will rush over to be his partner that day!

None of these actions would be possible without his being welcomed into our community school beginning in ECS. Without the support and belief by administration and teachers that inclusion can work, Murray would not be perceived by classmates as a “real” boy or “real” friend but, like poor Pinocchio, would sit on a shelf and watch others join in life’s activities until someone decided for him what he could or could not do.