Down Syndrome

milestone HCQU Northwest

Objectives:

Upon completion of this training, you will:

- Have increased knowledge of Down Syndrome, including the different classifications and characteristics
- Understand the diseases/conditions particularly associated with Down Syndrome
- Understand the differences in aging the person with Down Syndrome experiences

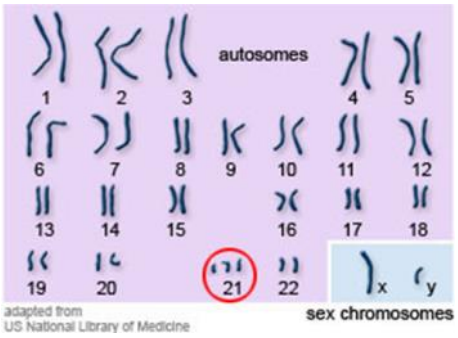
What is Down Syndrome?

The syndrome commonly known as Down Syndrome (DS), specifically the physical features and medical complications associated with it, was first described by John Langdon Haydon Down in 1866. Down Syndrome is also known as Trisomy 21, and is the most common chromosomal condition in humans, as well as the most common genetic cause of intellectual disability (ID). In fact, it is responsible for about 1 in 3 intellectual disabilities.

DS is a genetic condition causing delays in intellectual & physical development. The incidence of DS is approximately 1 in 700 to 900 live births, and the incidence increases as the mother ages. A mother at the age of 20 has a risk of 1 in 2000; at 30, 1 in 900; at 40, 1 in 100; and at age 49, the odds are 1 in 10. Counter-intuitive to that is the fact that 80% of DS births are to women less than 35 years of age. The reason for this is that the majority of children are born to women less than 35 years of age. DS occurs in all races.

Types of DS

There are three genetic versions of DS – each resulting from chromosomal irregularities of chromosome 21. Trisomy 21, which accounts for approximately 95% of cases, is the most common, with Translocation and Mosiacism comprising the other 5%. People with trisomy 21 have 47 chromosomes; three copies of chromosome 21 rather than two. Trisomy 21 is not genetically inherited. To the right is a karotype of a male with trisomy 21 — notice the three chromosomes on 21.



Translocation classification DS accounts for approximately 3-4% of people with DS. Translocation DS has two independent #21 chromosomes and a #21 chromosome that is attached to another chromosome. This is the only form of DS that is sometimes directly inherited. It may be inherited from the male or female. If the mother is the carrier there is a 12% chance, if the father is the carrier the chance is 3%. Mosaicism classification accounts for 1-2% of people with DS. Mosaicism results

(Continued on page 2)

Disclaimer:

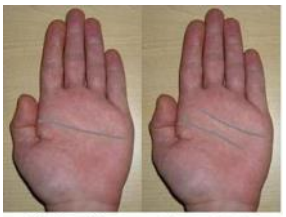
The information presented in this training is intended to provide education and ideas to promote the health and well being of people with intellectual and developmental disabilities; it is in no way meant to replace a doctor's advice or individual agency policies.

from abnormal cell division in only some cells, while others divide normally. When there are more normal than abnormal cells the features are less defined. Symptoms usually are nearly the same as the other forms of DS.

Testing for Down Syndrome

Genetic testing is advised when there is a family history of DS and a pregnancy is planned. This is done to identify if either parent is a carrier of the Translocation chromosome. Testing during pregnancy screening does not diagnose DS, but can help predict if the child might have DS. Tests are not 100% accurate for diagnosing DS. Screening during pregnancy includes fetal ultrasound to confirm gestational age of the fetus, detect some developmental deformities common with DS, and detect serious medical deformities earlier. Quad screen testing measures the levels of four substances in the mother's blood (Alpha-fetoprotein, Estriol, HCG, & Inhibin A) that indicate possibility of DS in the 15-18th week of gestation. False positives & false negatives are possible, making the test unreliable. Amniocentesis is performed by inserting a needle through abdominal wall into the amniotic sac; fluid is removed & genetically tested. This is usually performed in the 14 to 18th week of pregnancy. There are claims the test is 99.8% accurate in detecting DS; however, there is 2.5-3% risk of miscarriage with this test. Chorionic Villus Sampling is done under ultrasound by guiding a small tube inserted through the vagina into the uterus; a small tissue sample is taken from the placenta and genetically tested in the 10-12th weeks of gestation; results can be obtained quicker – usually within two weeks, with a 3-5% or less chance of miscarriage. With Percutaneous Umbilical Blood Sampling (PUBS), blood is taken from a vein in the umbilical cord and examined for chromosomal defects by karyotyping, usually after 18 weeks of gestation. A PUBS carries a greater risk of miscarriage than amniocentesis or chorionic villus sampling it is generally done when speed of diagnosis is essential. These tests are offered to a pregnant woman over 35 years old, or who has an abnormal triple screening test, previous birth of a child with DS, or there is a family history of DS.

After delivery, diagnosis is done by physical exam of the infant, detailed medical family history, and genetic blood testing of the infant. Characteristics such as small & low set ears, mouth – palate high and arched with downward curve, a protruding tongue, a flat nasal bridge, and upward slanting eyes. General characteristics are: short stature, broad neck, short broad extremities, a single crease across center of palms — called a simian line (shown at right), asymmetrical or odd shaped head, small skull —rounded and flat in back, unusually shaped and shorter than normal rib cage, and a protruding abdomen. All babies born with DS have some of these same characteristics in common, but most also resemble their family members.



Approximately 50% of infants with DS are born with heart conditions or heart defects, including atrial-septal defects, ventricular-septal defects, atrio-ventricular septal defects, patent Ductus Arteriosus, and Tetralogy of Fallot. All of these conditions affect the way blood flows through and around the heart abnormally. All infants born with DS should have an echocardiogram at 2-3 months. Survival rate has increased greatly with advent of better diagnosing and heart surgeries to correct or repair the abnormalities.

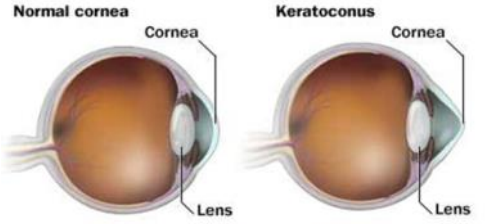
Co-existing Conditions

Some individuals are affected with endocrine disorders. Hypothyroidism is present in 10-40% of people with DS. Later in life this could masquerade as early-onset dementia. With hypothyroidism, the thyroid gland does not secrete enough thyroid hormone, causing the need to replace the hormone with medication. Hypothyroidism is managed by medications known as Synthroid (levothyroxine). Synthroid should be taken at same time every day, one hour before eating or two hours after eating. People with DS have a lower basal metabolic rate, which can lead to easier weight gain. To help them manage weight gain that can accompany hypothyroidism, individuals should increase physical activity to build stamina and limit sedentary activities.

People with DS are also at an increased risk of Type 1 diabetes. It's believed that the higher incidence is due to susceptibility to auto-immune disorders. Individuals with DS also suffer from increased risk of Type 2 diabetes, and at an earlier age – mean age being 24 years old.

Possible GI disorders include celiac disease in approximately 5-10% of people with DS. Celiac disease is a condition that damages the lining of the small intestine and prevents it from absorbing parts of food that are important for staying healthy. The damage is due to a reaction to eating gluten, which is found in wheat, barley, rye, and possibly oats. Approximately 30-50% are at an increased risk of aspiration pneumonia caused by swallowing problems. One to five percent have Gastro Esophageal Reflux Disease (GERD). Two to 15% have Hirschsprung disease, where the last part of large intestine does not function properly due to lack of certain nerve cells, causing extreme constipation which worsens with age.

People with DS may suffer from several eye conditions, such as: congenital cataracts, glaucoma, strabismus (eyes out of alignment/crossed eyes), and major refractory problems such as hyperopia (farsightedness) and myopia (nearsightedness). Keratoconus is a thinning of the cornea forming a cone shape as shown above. Correction of Keratoconus, as well as the refractory problems, may be corrected by glasses, contacts, or corneal transplants.



Persons with DS are more susceptible to respiratory infections due to a weaker immune system. Many suffer from obstructive sleep apnea (OSA), a condition in which the flow of air pauses or decreases during breathing while asleep because the airway has become narrowed, blocked, or floppy. Approximately 30-50% of people with DS have OSA caused by narrowed nasopharynx, low muscle tone of the upper airway, or enlarged adenoids and/or tonsils. Snoring is usually but not always present. Restlessness, disturbed sleep, frequent partial or total awakenings, daytime mouth breathing, and daytime sleepiness may all be caused by OSA. Treatment of OSA is usually by Continuous Positive Airway Pressure (CPAP) machine worn during sleep. Air pressure is delivered through a mask keeping the airway open while they sleep. For people who can't tolerate the CPAP, treatment is sometimes by an oral device, made by a dentist, that moves the bottom jaw and tongue forward, keeping the upper airway open. Surgery and removal of the tonsils and adenoids is a last resort, and is not always successful. Many people with DS also have irregular, crooked or missing teeth.

Chronic ear infections in childhood are common as well as chronic rhinitis and sinusitis. Some suffer from hearing loss caused by malformation of the bones of the middle and inner ear. **Hearing loss affects more than 50% of people with DS**. Audiometry (hearing test) should be performed at least every two years. Some people require routine removal of ear wax by a professional.

Musculoskeletal disorders such as scoliosis (curvature of the spine) occurs in about 8.7% of people with DS. Some suffer from dislocation of the hip when the head of the femur moves out of the socket formed by the pelvis. They are prone to poor muscle tone (hypertonia), instability of the kneecaps, flat feet, arthritis, osteopenia (marginal thinning of the bones), osteoporosis (thinning of the bones), and atlanto-axial instability — a condition characterized by excessive movement at the junction between the atlas (C1) and axis (C2) in the cervical spine. Leukemia occurs at increased incidence of 10 to 20 times greater than the general population. Ninety-two percent of people with DS who are diagnosed with leukemia are less than 20 years old. It accounts for more than 50% of all cancers affecting people with DS. Males with DS have a higher prevalence for testicular cancer. Doctors recommend yearly testicular exams for people between 15 and 25 years old. People with DS seem to be at a decreased risk for most other solid cancers (tumerous).

Alzheimer's Disease (AD) causing dementia is common in adults with Down syndrome. The onset of dementia symptoms may begin up to 20 years earlier than in the general population. Alzheimer's disease is a progressive, degenerative, neurological disorder that affects the person's ability to think, reason, remember, use and process language, and be oriented to per-

son, place, and time. It is estimated that up to 20% of people with DS over age 35 have signs & symptoms of Alzheimer's disease. The incidence of people with DS getting Alzheimer's Disease is three to five times greater than the general population; symptoms begin much earlier and the disease course, usually leading to death, occurs more rapidly. If the onset of Dementia-type symptoms is quick, it is imperative to rule out any other possible causes of the symptoms. Conditions such as thyroid disease, depression, brain tumor, recurrent strokes, metabolic imbalances, and many others can mimic the symptoms of AD. The hallmark lesions (neurofibrillary plaques and tangles) of Alzheimer's disease are present in all people with DS by the age 40. These plaques & tangles do not necessarily mean the person will develop AD. Some of the clinical features of AD are personality changes, irritability, apathy, loss of speech or change in language skills, disorientation to time & place, decline in self care skills, abrupt onset of seizure activity (**seizures may occur in up to 46% of people with DS over the age of 50**), abrupt incontinence, short-term memory loss, and sleep/wake cycle disruption. When diagnosing AD it is important to have a baseline assessment of skills and functional status. AD has a slow gradual onset with a documentable decline. The person with DS should be re-evaluated yearly after age 40, so that changes that occur can be managed and documented. When doing care plans or Individual Service Plans (ISP's) the goals should be realistic, and appropriate to the abilities and strengths of the individual. Care Plans/ISP's should include legal and financial planning and preparing advanced directives, and should be done while the person still has the ability to make their wishes known. Care plans should pay particular attention to physical health in those with AD because there is proof that the more physically healthy they are the better they fare with AD. Caregivers need training and support throughout the course of the progression of the disease.

Many people with DS have behavior Issues, including: temper tantrums, oppositional defiance, depression, self- talk, compulsive behavior, anxiety disorders, autistic disorders, and ADHD. Acute illness or death of a loved one can trigger behavioral & emotional difficulties. Many have difficulty talking about things that make them sad or angry. Their receptive language skills are stronger than their expressive language.

DS and Aging

People with DS age quicker than the general population, although they are experiencing longer lifespans than ever before. People with DS actually age more rapidly at the cellular level, affecting all body systems. Normal aging occurs about 20 years earlier. In the 1960's the life expectancy of a person with DS was 25. Today many are living into their sixties, probably due to better heart surgeries to correct congenital heart defects and better antibiotics. Many researchers feel strongly that it will be possible to improve, correct, or prevent many of the problems associated with Down syndrome in the future.

People with Down Syndrome Can Lead EveryDay Lives

Every person with Down Syndrome is a unique individual and possesses the characteristics of the syndrome to varying degrees. People with Down syndrome attend school, they work, they participate in decisions that affect them, and some contribute to society in wonderful ways. Education programs, a supportive home environment, good health care, and positive supports from family, friends and the community enable people with Down syndrome to develop full potential and

Down Syndrome Test

Name: _____ Role/Title: _____

Agency: _____ Date: _____

Please provide contact information (email address, fax number, or mailing address) where you would like your certificate to be sent: _____

_____

You must submit your completed test, with at least a score of 80%, to receive **1 hour of credit** for this course.

To submit via fax, please fax the test and evaluation to 814-728-8887.

To submit via email, please send an email to HCQUNW@MilestonePA.org. Please put "Down Syndrome Test" in the subject line, and the numbers 1—5, along with your answers, in the body of the email.

To submit via mail, please send to Lynn Carnahan, Milestone HCQU Northwest, 247 Hospital Drive, Warren, PA 16365

Circle the best answer.

1. Which of the following increases the risk of a child having Down Syndrome?
   a. older father
   b. younger mother
   c. older mother
   d. younger father
2. Although the risk increases with age, 80% of children with Down Syndrome are born to mothers in which age group?
   a. >40
   b. >30
   c. <20
   d. <35
3. There are how many different genetic versions of DS, though the characteristics are the same?
   a. 1
   b. 2
   c. 3
   d. Many
4. Which of the following is not a health problem mentioned in this article?
   a. congenital heart conditions
   b. endocrine disorders
   c. premature aging
   d. gout
5. People with Down Syndrome are more likely to have obstructive sleep apnea (OSA). Which of the following is not a symptom of OSA?
   a. repeated awakenings
   b. daytime sleepiness
   c. snoring
   d. blurry vision

EVALUATION OF TRAINING

Training Title: Down Syndrome Home Study

Date: _____

Please check the box that best describes your role:

- ☐ Direct Support Professional
- ☐ Provider Administrator/Supervisor
- ☐ Program Specialist
- ☐ Provider Clinical Staff
- ☐ Consumer/Self-Advocate
- ☐ Family Member
- ☐ Support Coordinator
- ☐ Support Coordinator Supervisor
- ☐ PCH Staff/Administrator
- ☐ FLP/LSP
- ☐ County MH/MR/IDD
- ☐ Other (please list): _____

Please circle your PRIMARY reason for completing this home-study training:

☐ It's mandatory ☐ interested in subject matter ☐ need training hours ☐ convenience

Please circle the best response to each question.

5 = Strongly Agree 4 = Agree 3 = Undecided 2 = Disagree 1 = Strongly Disagree

1.	As a result of this training, I have increased my knowledge.	5	4	3	2	1
2.	I learned something I can use in my own situation.	5	4	3	2	1
3.	This training provided needed information.	5	4	3	2	1
4.	The training material was helpful and effective.	5	4	3	2	1
5.	Overall, I am satisfied with this training.	5	4	3	2	1
6.	I am glad I completed this training.	5	4	3	2	1

Suggestions for improvement: _____

Additional information I feel should have been included in this training: _____

I would like to see these topics/conditions developed into home-study trainings: _____