

Health Care Guidelines for Individuals with Down Syndrome

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Dedicated to the memories of Chris Pueschel and Thomas E. Elkins M.D., two individuals, who, each in his own way, has inspired us to provide compassionate care for individuals with Down Syndrome

Introduction

Individuals with Down syndrome (DS) need the usual health care screening procedures recommended for the general population. For example, children with DS need the usual immunizations and well child care procedures as recommended by the American Academy of Pediatrics.² Immunization practices are continually evolving: be certain to use the most up-to-date protocols.³ Similarly, adults with DS should have health evaluations using the standard accepted practices. However, children with DS have an increased risk of having certain congenital anomalies. Both children and adults may develop certain medical problems that occur in much higher frequency in individuals with DS. Described below is a checklist of additional tests and evaluations recommended for children and adults with DS. These recommendations should take into consideration available local expertise and referral patterns. They are based on our present level of knowledge and should be modified as new information becomes available. Modern primary health care includes educational and developmental concerns within its domain, and therefore we have included information and recommendations specific to these needs of individuals with DS.

These recommendations are a thoughtful composite of the input of many clinicians involved in the care of people with DS. They reflect current standards and practices of health care in the United States of America. They have been designed for a wide audience: for health care professionals who are providing primary care, such as pediatricians, family physicians, internists, and geneticists, as well as specialists, nursing personnel and other allied health professionals, such as physical and occupational therapists, speech-language pathologists, and audiologists. In addition to educators and early intervention providers, these guidelines are designed for parents and other caregivers to use with the professionals who participate in the care of the individual with DS.

Certain recommendations are clearly supported by current scientific knowledge. This is the case for the recommendations to look for the presence of congenital heart disease, which occurs in some 50% of infants with DS. In other cases, the recommendations represent our educated guesses. Recognizing the increased frequency of thyroid dysfunction in children with DS, we continue to recommend yearly screening for hypothyroidism. However, we are uncertain as to the appropriate periodicity and nature of the screening: how often, and what constitutes an adequate screening. This question, and others, will be answered by the anticipated development of a large-scale clinical database.

Be certain to use the specific DS growth charts in addition to regular charts to record height and weight (for children from birth to 18 years of age), and head circumference (for children birth to 36 months of age).⁴ If a child is below the third percentile, or if falling off the expected percentiles, consider congenital heart disease, endocrine disorders (thyroid or pituitary), or nutritional factors. Because children with DS have a tendency to become overweight, always use the "Weight v. Height" plots on the growth charts for typically developing children; this will give a more realistic picture of appropriateness of a child's weight.

[**Note:** Immediately following the recommendations by age, you will find explanations for the specific medical recommendations listed below, descriptive information about other areas of interest to individuals interested in the needs of individuals with Down syndrome, and an updated bibliography.]

About these Health Care Guidelines (Preventive Medical Check List)

These health guidelines continue the series begun in 1981, by Dr. Mary Coleman, and published in *Down Syndrome Papers and Abstracts for Professionals (DSPAP)*, the predecessor of this *Down Syndrome Quarterly*. The 1992 version was prepared by the members of the Ohio/Western Pennsylvania Down Syndrome Network and published in *DSPAP* (Volume 15, Number 3, 1992, 1-9) and was based on the 1989 version prepared by Dr. Nancy Roizen, University of Chicago. The 1996 version was the first one prepared by the Down Syndrome Medical Interest Group (DSMIG).

In July, 1994, the Committee on Genetics of the American Academy of Pediatrics published "Health Guidelines for Children with Down syndrome."⁵ Members of DSMIG have been fortunate to work with the Committee during their recent review of their "Health Guidelines" for the purpose of coordinating these efforts and removing differences and incongruities. This version therefore reflects the shared screening protocol, which the Committee will publish in *Pediatrics* in early 2000. The editor wishes to express his appreciation to Drs. Marilyn Bull and Nancy Roizen for their liaison work with the Committee, and to Dr. Franklin Desposito, committee chair, and Dr. Tracy Trotter and the other members of the committee for their substantial assistance in this joint effort. The preparation of this revision has been a cooperative effort. The editor has been particularly fortunate to have the expertise of several members of Down Syndrome Medical Interest Group (DSMIG).⁶

This is one of many such compilations. Please see the **References**, Section C, for a selected list of other protocols.

A NOTE ABOUT FLOW CHARTS: These "Health Guidelines" were prepared with the goal of providing both depth and breadth to the topic of health promotion for individuals with Down syndrome. We trust that this will serve as a reference for families, educators, agencies, and, of course, health care providers. Nevertheless, we recognize the ease and simplicity of using a summary of these guidelines in a one-page graphic format. Such a summary can be placed in the front of a family's medical record book, and likewise, in the front of a medical chart for rapid consultation. Several members of *DSMIG* have developed such forms. In 1989, Dr. Allen Crocker prepared a "Healthwatch for Persons with Down Syndrome", which is reprinted in Dr. W. Carl Cooley's chapter in Van Dyke, D. C. et al, Ed., *Medical and Surgical Care for Children with Down Syndrome*.

We have included with this document a Flow Chart adapted from Dr. David Smith's document, used at the DS Clinic of Wisconsin in Milwaukee. (See Appendix I) Dr. Brian Chicoine has prepared a variety of materials for providing health care to adults with Down syndrome. These include history questionnaires, review of systems checklists, physical examination forms and an assessment/plan form which includes screening information. You can contact him at the Adult Down Syndrome Clinic at Lutheran General Hospital, Park Ridge, IL at 847-795-2303 if you wish to obtain this material.

What's New for 1999

- Additional thyroid screening at 6 months of age.
- Hearing evaluations at birth, and every 6 months thereafter until 3 years.
- Eye evaluations by 6 months of age and yearly.
- Celiac disease screening between 2 and 3 years of age.
- Radiographic screening for atlanto-axial instability once between 3 and 5 years, and then as needed for Special Olympics

Neonatal (Birth to one month)

History: Review parental concerns. Was there a prenatal diagnosis of DS? With vomiting or absence of stools, check for gastrointestinal tract blockage (duodenal web or atresia, or Hirschsprung disease); review feeding history to ensure adequate caloric intake; any concerns about hearing or vision? Inquire about family support.

Exam: Pay special attention to cardiac examination; cataracts (refer immediately to an ophthalmologist if the red reflex is not seen); otitis media; subjective assessment of hearing; and fontanelles (widely open posterior fontanelle may signify hypothyroidism). Exam for plethora, thrombocytopenia.

Lab and Consults: Chromosomal karyotype; genetic counseling; hematocrit or complete blood count to investigate plethora (polycythemia) or thrombocytopenia (possible myeloproliferative disorders); thyroid function test - check on results of state-mandated screening; evaluation by a pediatric cardiologist including echocardiogram (even in the absence of a murmur); reinforce the need for subacute bacterial endocarditis (SBE) prophylaxis in susceptible children with cardiac disease; refer for auditory brainstem response (ABR) or otoacoustic emission (OAE) test to assess congenital sensorineural hearing at birth or by 3 months of age. Refer for a pediatric ophthalmological evaluation by six months of age for screening purposes. Refer immediately if there are any indications of nystagmus, strabismus or poor vision. If feeding difficulties are noted, consultation with feeding specialist (occupational therapist or lactation nurse) is advised.

Developmental: Discuss value of Early Intervention (infant stimulation) and refer for enrollment in local program. Parents at this stage often ask for predictions of their child's abilities: "Can you tell how severe it is?" This is an opportunity to discuss the unfolding nature of their child's development, the importance of developmental programming, and our expectation of being able to answer that question closer to two years of age.

Recommendations: Referral to local DS parent group for family support, as indicated.

Infancy (1 - 12 months)

History: Review parental concerns. Question about respiratory infections (especially otitis media); for constipation, use aggressive dietary management and consider Hirschsprung disease if resistant to dietary changes and stool softeners. Solicit parental concerns regarding vision and hearing.

Exam: General neurological, neuromotor, and musculoskeletal examination; must visualize tympanic membranes or refer to ear, nose and throat (ENT) specialist, especially if suspicious of otitis media.

Lab and Consults: Evaluation by a pediatric cardiologist including echocardiogram (if not done in newborn period): remember to consider progressive pulmonary hypertension in DS patients with a VSD or atrioventricular septal defect who are having little or no symptoms of heart failure in this age group. Auditory brainstem response test (ABR) by 3 months of age if not performed previously or if previous results are suspicious. Pediatric ophthalmology evaluation by six months of age (earlier if nystagmus, strabismus or indications of poor vision are present). Thyroid function test (TSH and T4), at 6 and 12 months of age. Evaluation by ENT specialist for recurrent otitis media as needed.

Developmental: Discuss early intervention and refer for enrollment in local program (if not done during the neonatal period). This usually includes physical and occupational therapy evaluations and a developmental assessment.

Recommendations: Application for Supplemental Security Income (SSI) (depending on family income); consider estate planning and custody arrangements; continue family support; continue SBE prophylaxis for children with cardiac defects.

Childhood (1 year to 12 years)

History: Review parental concerns; current level of functioning; review current programming (early intervention, preschool, school); ear problems; sleep problems (snoring or restless sleep might indicate obstructive sleep apnea); constipation; review audiologic and thyroid function tests; review ophthalmologic and dental care. Monitor for behavior problems.

Exam: General pediatric and neurological exam including evaluation for signs of spinal cord compression: deep tendon reflexes, gait, Babinski sign. Include a brief vulvar exam for girls. Use Down syndrome growth charts, as well as growth charts for typically developing children. Be sure to plot height for weight on the latter chart.

Lab and Consults: Echocardiogram by a pediatric cardiologist if not done previously; Thyroid function test (TSH and T4) yearly; behavioral auditory testing every 6 months until 3 years of age, then yearly. Continue regular eye exams every year if normal, or more frequently as indicated. Between 3 years and 5 years of age, lateral cervical spine x-rays (neutral view, flexion and extension) to rule out atlanto-axial instability: have the radiologist measure the atlanto-dens distance and the neural canal width. X-rays should be performed at an institution accustomed to taking and reading these x-rays. Initial dental evaluation at two years of age with follow-ups every six months. At 2-3 years of age, screen for celiac disease with IgA antiendomysium antibodies, as well as total IgA. Administer Pneumococcal vaccine at 2 years of age.

Developmental: Enrollment in appropriate developmental or educational program; complete educational assessment yearly, as part of Individualized Family Service Plan (IFSP) for children from birth to 3 years of age, or Individualized Educational Plan (IEP) from age four until the end of formal schooling. Evaluation by a speech and language pathologist is strongly recommended to maximize language development and verbal communication. An individual with significant communication deficits may be a candidate for an augmentive communication device.

Recommendations: Twice daily teeth brushing. Total caloric intake should be below recommended daily allowance (RDA) for children of similar height and age. Monitor for well-balanced, high fiber diet. Regular exercise and recreational programs should be established early. Continue speech therapy and physical therapy as needed. Continue SBE prophylaxis for children with cardiac defects. Monitor the family's need for respite care, supportive counseling and behavior management techniques. Reinforce the importance of good self-care skills (grooming, dressing, and money handling skills).

Adolescence (12 to 18 years)

History: Review interval medical history, questioning specifically about the possibility of obstructive airway disease and sleep apnea; check sensory functioning (vision and hearing); assess for behavioral problems; address sexuality issues.

Exam: General physical and neurological examination (with reference to atlanto-axial dislocation). Monitor for obesity by plotting height for weight on the growth charts for typical children. Pelvic exam if sexually active, only. (See Consults, below.) Perform a careful cardiac exam in adolescents, looking for evidence of valvular disease. Lab and consults: Thyroid function testing (TSH and T4) yearly. Hearing and vision evaluations every year. Repeat screening cervical spine x-rays as needed for Special Olympic participation. Echocardiogram if evidence of valvular disease on clinical exam. Consult with Adolescent Medicine practitioner or a gynecologist experienced in working with individuals with developmental disabilities to address issues of sexuality and/or for pelvic examination for sexually active teenager. Continue twice-yearly dental exams.

Developmental: Repeat psycho-educational evaluations every two years as part of Individualized Educational Plan (IEP). Monitor independent functioning. Continue speech/language therapy as needed. Health and sex education, including counseling regarding abuse prevention. Smoking, drug, and alcohol education.

Recommendations: Begin functional transition planning (age 16). Consider enrollment for SSI depending on family income. SBE prophylaxis needed for individuals with cardiac disease. Continue dietary and exercise recommendations (see childhood, above). Update estate planning and custody arrangements. Encourage social and recreational programs with friends. Register for voting and selective service at age 18. Discuss plans for alternative long term living arrangements such as community living arrangements (CLA). Reinforce the importance of good self-care skills (grooming, dressing, and money handling skills).

Adults (over 18 years)

History: Interval medical history. Ask about sleep apnea symptoms. Monitor for loss of independence in living skills, behavioral changes and/or mental health problems. Symptoms of dementia (decline in function, memory loss, ataxia, seizures and incontinence of urine and/or stool). This may also represent spinal cord compression from atlanto-axial subluxation.

Exam: General physical and neurological examination (with reference to atlanto-axial dislocation). Monitor for obesity by plotting height for weight. Cardiac exam: listen for evidence of mitral valve prolapse and aortic regurgitation: confirm suspicions with echocardiogram. Sexually active women will need Pap smears every 1-3 years following the age of first intercourse. For women who are not sexually active, single-finger bimanual examination with finger-directed cytology exam. Screening pelvic ultrasound every 2-3 years for women who refuse or have inadequate follow-up bimanual examinations. This may require referral to an Adolescent Medicine practitioner or a gynecologist with experience with individuals with special needs. Otherwise, pelvic ultrasound may be considered in place of pelvic examinations. Breast exam yearly by physician.

Lab and consults: Annual thyroid screening (TSH and T4). Ophthalmologic evaluation every two years (looking especially for keratoconus and cataracts). Repeat cervical spine x-rays as needed for Special Olympic participation. Continue auditory testing every two years. There are two different suggestions for mammography: Dr. Heaton recommends yearly study after age 50; begin at age 40 for women with a first-degree relative with breast cancer. Dr. Chicoine suggests a mammogram every other year beginning at 40, and yearly beginning at 50. Continue twice-yearly dental visits. Mental health referral for individuals with emotional and behavioral changes.

Developmental: Continue speech and language therapy, as indicated. For individuals with poor expressive language skills, consider referral for augmentive communication device. Discuss plans for further programming/vocational opportunities at age 21 or when formal schooling ends. Be aware that accelerated aging may affect functional abilities of adults with DS, more so than Alzheimer disease.

Recommendations: Discuss plans for alternative long term living arrangements such as community living arrangements (CLA). SBE prophylaxis needed for individuals with cardiac disease. Continue dietary and exercise recommendations (see childhood, above). Update estate planning and custody arrangements. Encourage social and recreational programs with friends. Register for voting and selective service at age 18. Reinforce the importance of good self-care skills (grooming, dressing, and money handling skills). Bereavement counseling for individuals who have experienced the loss of an important person in their life, either via death or by other circumstances: sibling moves away after marriage, or goes off to college.

The following are an elaboration of the recommendations made above, as well as other information designed to promote optimal health care for individuals with Down syndrome:

Cardiac: Congenital heart disease is reported to occur in 30 - 60% of children with DS. Ventricular septal defects and complete atrioventricular septal defects are among the most common. A serious cardiac defect may be present in the absence of a murmur because of the increased tendency of children with DS to develop early increases in pulmonary vascular resistance which reduces the left to right intracardiac shunt, minimizes the heart murmur, and prevents symptoms of heart failure and respiratory problems. Children with DS with a significant cardiac defect who seem to be doing clinically well or getting better, especially during the first 8 months of life, may be developing serious pulmonary vascular changes. Timely surgery, frequently during the first 6 months of life, may be necessary to prevent serious complications. **Therefore, all infants and children need to have an evaluation by a pediatric cardiologist, preferably before three months of age, which should include an echocardiogram.** In some tertiary care centers, an echocardiogram alone is satisfactory when it will be evaluated by a pediatric cardiologist. If this is not available, a full evaluation by a pediatric cardiologist is mandatory. For the older child, who has never had a cardiac evaluation and who has no signs of cardiac disease, a screening echocardiogram is recommended. Adolescents and young adults with no known intracardiac disease can develop valve dysfunction and should be evaluated by clinical examination at age 18, especially prior to dental or surgical procedures. [See References, Section G, Geggel RL, et al.] There is a 57% incidence of mitral valve prolapse and approximately a 10% risk of aortic regurgitation. The finding of a click or murmur should be followed by an echocardiogram. Susceptible individuals will need SBE prophylaxis.

Dental Care: The orofacial features of individuals with DS contribute to a variety of potential problems in regards to dental care. For example, the eruption of teeth is usually delayed and often occurs in an unusual order. Primary and permanent teeth may be missing. Small or misshapen teeth are found, and severe crowding can occur because of the small oral cavity. Orthodontic treatment may be necessary. Mouth breathing, related to the small nasal airway, contributes to fissured tongue and lips. Periodontal disease can occur as early as the teen years, and routine brushing combined with dental visits every 6 months play a key role in preventing tooth loss.

Ears/Audiology: Hearing loss is a significant area of concern for individuals with DS. Infants and children may have a sensorineural loss, a conductive loss (related to middle ear effusions) or both. All infants with DS should have an objective measure of hearing performed at birth, if possible, or within the first 3 months of life. The most common method in widespread use is the measurement of auditory brainstem responses (ABR), also known as brainstem auditory evoked response (BAER). Two screening methods include ABR screening in the newborn nursery, and evoked oto-acoustic emission testing. The typical behavioral audiology requires a developmental age of 7-8 months. Consequently, all children with DS need an objective measure when tested in the first 12 months. Subsequently, behavioral audiology may be appropriate. Most children with DS have very small ear canals, making it difficult to examine them properly with the instruments found in the pediatrician's office. Consequently, it may be necessary to refer the child to

an Ear, Nose, and Throat physician to visualize the tympanic membranes using the microscopic otoscope. An ENT physician should evaluate all children with an abnormal hearing evaluation and/or tympanogram in order to aggressively manage treatable causes of hearing loss (using antibiotics and/or tympanostomy tubes as indicated). Fluid can accumulate as early as the neonatal period and aggressive otologic care can minimize the effect of any hearing loss on language development.

Individuals with Down syndrome may begin to develop hearing loss in their second decade, which, if undetected may lead to behavioral symptoms which could be misinterpreted as a psychiatric disorder.

ENT: The midfacial hypoplasia (underdevelopment) that is characteristic of individuals with DS leads to increased difficulty with narrow airways. The narrow nares (nostrils) manifest themselves as noisy breathing in the infant; narrow openings to paranasal sinuses predisposes children to frequent sinusitis/nasopharyngitis. These infections, manifested by purulent nasal discharge, should be aggressively treated. The narrow trachea can result in recurrent croup. In addition, infants with DS have an increased likelihood of tracheomalacia (partial collapse of the trachea).

Obstructive airway disease has been recognized as a significant problem for children and adults with DS. Symptoms include snoring, unusual sleeping positions (sitting up or bending forward at the waist with head on knees), fatigability during the day, reappearance of napping in older children or behavior change. Individuals with these symptoms should be evaluated completely via detailed history (looking specifically for evidence of sleep apnea), physical examination with regard to tonsillar size, and prompt referral to an ENT physician for further evaluation (eg. assessment of adenoidal size). In a number of children, hypotonicity and collapse of the airway leads to similar symptoms in the absence of obstruction caused by lymphoid tissue. Surgical intervention may be necessary to avoid hypoxemia and possible cor pulmonale (right heart failure). In the absence of surgically correctable problems, supplemental O₂ therapy under pressure (such as bi-pap) may be indicated.

A recent study from Israel notes that children with DS have significant sleep fragmentation with frequent awakenings and arousals that are not related to obstructive sleep apnea syndrome. [See References, Section K, Levanon et al.]

Two studies point to the importance of keeping children with DS in the hospital overnight following tonsillectomy and adenoidectomy, because of higher rate of postoperative respiratory complications. [See References, Section K, Bower et al, and Goldstein et al.]

Endocrine: The incidence of thyroid disease is significantly increased among individuals with DS of all ages. Normal thyroid hormone levels are necessary for growth and cognitive functioning. The signs of hypothyroidism may be subtle in individuals with DS and may be attributed to the DS itself. Therefore, screening is recommended on a yearly basis by monitoring TSH and T₄ levels. Since autoimmune conditions are common in individuals with DS, evaluation of suspected hypothyroidism in the school age child should include thyroid antibodies to look for thyroiditis. Some infants and young children have a condition known as idiopathic hyperthyrotropinemia, with borderline abnormal TSH with normal T₄. This may reflect a neuroregulatory defect of TSH, which, when studied by 24 hour sampling, varies between normal levels and very high levels. Therefore, some centers recommend repeating the TSH and T₄ every six months, withholding treatment unless the T₄ is low. Immune-mediated hyperthyroidism also occurs in children and adults with DS. High sensitivity TSH levels will be abnormally low in these cases. In addition, weight loss, GI symptoms and heat intolerance are often seen.

Diabetes mellitus, recognized to be an autoimmune condition, also occurs more frequently in individuals with DS, with a prevalence rate between 1.4 and 10.6%. [References, Section M, Anwar et al.]

There has been some discussion about the use of human growth hormone in children with DS in response to a report that suggested that children with DS have an abnormality of growth hormone secretion. This issue has been addressed by members of the Down Syndrome Medical Interest Group, and published in *Down Syndrome Quarterly*, Vol 1, Number 1 (March, 1996), page 8: "On the basis of the available evidence, and until the recommended scientific studies are completed, the uncontrolled use of hormonal treatments such as growth hormone in children with Down syndrome is not supported by the Down Syndrome Medical Interest Group." A recent study from Sweden and Australia reveals that treatment with human growth hormone did result in "normal height velocity but did not affect head circumference or mental or gross motor development." [See References, Section M, Anneren G, Tuvemo T, Sava VR et al.]

Feeding/Nutrition: Infants with DS may initially experience difficulty with coordination of suck and swallow. They may need assistance of a feeding specialist (such as nurse specialists, occupational therapists, speech-language pathologists) or lactation specialists, if the mother is nursing. Later on, toddlers may have difficulty with texture progression. Significant disruption of family activities, involvement of more than two professionals indicates that a consultation with a multi-disciplinary feeding team may be warranted. [See References Section N, Medlen.] Remember that there is up to a 10% difference in basal metabolic rate for individuals with DS. Follow % Ideal Body Weight, calculated as follows: plot child's height on growth chart for typically developing children and determine the age for which this height is at the 50th percentile. Determine the weight at the 50th percentile for this height. Divide the actual weight of the child by this determined weight and multiple by 100. Goal is 90% - 100% IBW. Chart this sequentially.

Hematology: Leukemia is more common in children with DS than in the general population. Nevertheless, this is rare. Most leukemia in children less than 3 years of age is non-lymphocytic leukemia (usually acute myelogenous leukemia). Children with DS usually respond very favorably to standard treatment, going into remission easily. In the newborn period, there is a 10% incidence of myeloproliferative disorder ("leukemoid reaction") which in some instances develops into acute megakaryoblastic leukemia. Polycythemia has been frequently observed in the newborn period; in one series, as high as 64% of children studied were affected

Infectious Disease/Immunology: Persons with DS who have serious recurrent respiratory and systemic infections are often evaluated for immune function. Consider measuring the IgG subclasses in such individuals. Total IgG level may not disclose any abnormality, although there may be a deficiency of IgG subclasses 2 and 4 and an increase of IgG subclasses 1 and 3. There is a significant correlation between the decreased IgG subclass 4 levels and bacterial infections. The mechanism is not known but theories include the possibility that this subclass plays a role in pulmonary host defense or possibly a deficiency of selenium. Intravenous gamma globulin replacement therapy should be a consideration in a person with DS who presents with serious recurrent bacterial infections and documented IgG subclass 4 deficiency.

The cellular immunity deficits described in individuals with DS have the greatest documented clinical impact on gingivitis and periodontal disease.

Children with chronic cardiac and respiratory disease are candidates for use of pneumococcal, respiratory syncytial virus, and influenza vaccines.

Eye/Vision: Congenital cataracts are a serious problem for infants with DS, leading to vision loss if not detected and treated. The absence of a red reflex is sufficient cause for immediate referral to a pediatric ophthalmologist, as are strabismus and nystagmus. Routine evaluations should begin at 6 months of age, and be performed annually thereafter. Refractive errors are common and will be detected during these evaluations, as would serious, but rarer, conditions, such as keratoconus. Stenotic nasolacrimal ducts may lead to tearing in infancy. Blepharitis and conjunctivitis occur frequently. Keratoconus occurs more frequently in adolescents with DS than in the typical child.

Orthopedic Disorders & Atlanto-axial Instability (AAI): Ligamentous laxity is responsible for a number of orthopedic difficulties in individuals with DS. Interestingly, congenital hip dislocation is not commonly encountered. Hip dislocation is more often seen in the older child and the adolescent. Chronic patellar dislocation can lead to gait disturbances in the adolescent. Atlantoaxial instability is a term used to describe increased mobility of the cervical spine at the level of the first and second vertebrae. This condition is found in approximately 14% of individuals with Down syndrome. The majority of individuals with atlantoaxial instability are asymptomatic, but approximately 10% of these individuals with AAI (representing 1% of individuals with Down syndrome) have symptoms, which occur when the spinal cord is compressed by the excessive mobility of the two vertebrae which form the atlantoaxial joint. Symptoms of spinal cord compression may include neck pain, unusual posturing of the head and neck (torticollis), change in gait, loss of upper body strength, abnormal neurological reflexes, and change in bowel/bladder functioning.

Routine radiographic screening for atlantoaxial instability of individuals with Down syndrome is controversial. In a recent review, the American Academy of Pediatrics Committee on Sports Medicine and Fitness concluded that screening radiographs are of "potential but unproven value" in detecting individuals at risk from sports injury. Close clinical scrutiny and further study of this issue was recommended. However, these studies continue to be required for participation in Special Olympics and community programs in horseback riding, gymnastics, etc.

Currently, DSMIG recommends screening individuals between 3 and 5 years of age with lateral cervical radiographs in the neutral, flexed, and extended positions. The space between the posterior segment of the anterior arch of C1 and the anterior segment of the odontoid process of C2 should be measured. Measurements of less than 5 mm are normal; 5 to 7 mm indicates instability, and greater than 7 mm is grossly abnormal. The cervical canal width should also be measured. The interpretation of these studies should be performed by a radiologist experienced in this area. Individuals with Down syndrome who have not been screened may need to be evaluated prior to surgical procedures, especially those involving manipulation of the neck. These children should be managed cautiously by anesthesiology staff. The studies should be repeated, as needed, for participation in Special Olympics.

Children with borderline findings or abnormal films should be evaluated with a careful neurological examination to rule out spinal cord compression. Neuro-imaging (CT Scan or MRI) is probably indicated. Significant changes in a child's neurological status would necessitate evaluation and possible treatment (i.e., spinal fusion). Asymptomatic children with instability (5 to 7 mm) should be managed conservatively, with restriction only in those activities which pose a risk for cervical spine injury. Contact sports, such as football, wrestling, rugby, boxing, and recreational activities such as trampolining, gymnastics (tumbling), and diving, which require significant flexion of the neck, would best be avoided. It is unnecessary to restrict all activities.

We are no longer recommending repeat screenings at fixed intervals, inasmuch as the value of this procedure has not yet been confirmed in preventing injury. We strongly recommend careful neurological examination of the individual with Down syndrome, immediate attention to symptoms indicating neck or spinal cord problems (see above), and vigilance by ENT physicians and anesthesiologists during surgical procedures which may hyperextend the neck.

The editor understands that the Special Olympics Medical Advisory Committee is involved in clarifying the problematic issue of detection and prevention of spinal cord injuries. [For a recent review of the subject, see References, Section W, Pueschel (1998) & Cohen (1998).]

Physical/Occupational Therapy: Since infants with DS may have difficulty with feeding from birth, keep in mind that many centers have professionals (such as occupational therapists, speech pathologists, feeding nurse specialists, etc) who can provide expertise in this area. Some centers involve the occupational therapist or feeding specialist on a routine basis, while others assess the child's oral-motor function and refer as needed. In general, physical and occupational therapy services are included in most early intervention programs for infants, where positioning, feeding, and motor strengthening exercises are some of the services available.

Gastrointestinal disorders: In addition to congenital abnormalities, such as duodenal atresia and imperforate anus, which are readily identifiable, babies with Down syndrome are more likely to have partial upper GI obstruction (duodenal web), tracheo-esophageal fistula, and pyloric stenosis. Chronic constipation occurs frequently, and the serious conditions in the differential diagnosis includes hypothyroidism, and Hirschsprung disease. Failure to pass meconium in the first 24 hours suggests the possibility of Hirschsprung disease. Significant constipation which is refractory to dietary management warrants further investigation, such as referral to a pediatric gastroenterologist for further studies (barium enema, rectal biopsy).

Gastroesophageal reflux (GER) occurs in infants with DS, as it does in the typical population. In addition to spitting up and vomiting, some children have respiratory symptoms, such as cough, stridor, wheezing and pneumonia. GER must be part of the differential diagnosis for these conditions, and appropriate treatment given.

Celiac disease occurs in from 7 to 16% of children with DS, though many of these studies are from European sources. Individuals with DS are felt to be predisposed to this condition because of the known increased incidence of autoimmune disorders. Screening is best accomplished using IgA antiendomysium antibodies, following up positive results with a villous biopsy. Symptoms usually resolve following institution of a gluten-free diet.

Genetics: A medical genetics consultation should be encouraged, in order to explain the genetic basis and risk of recurrence of DS. Such consultation may be considered optional for children with Trisomy 21. However, in cases of translocation, the parents should be evaluated to determine whether one of them is a balanced carrier of the translocation, thereby increasing the likelihood that subsequent children may have Down syndrome. This service should also be made available to individuals with DS, when appropriate.

Prenatal screening & testing technologies continue to evolve. Proposed methods include separating fetal cells from the maternal circulation, and use of multiple serum markers and nuchal thickness as measured by ultrasonography.

Developmental, including Speech and Language: Early intervention programs (for infants 0-3 years old) are designed to comprehensively monitor and enrich development, focusing on feeding, gross and fine motor development, language, and personal/social development. Individuals with DS frequently understand spoken language better than they can express themselves verbally. Consequently, infants and children may be taught language using a total communication approach, which includes signing as well as spoken language. Signing permits these children to communicate more effectively at a time when their expressive language abilities may preclude the development of intelligible speech. Speech and language services should be considered throughout life, to maximize intelligibility. Additionally, some individuals may benefit from the use of augmentive (computer-based) communication devices. Children with DS often continue to develop verbal expressive language into their adolescent and young adult years. The strong visual skills of individuals with DS have led to the development of reading programs to improve speech and language development. [See References, Section N, Laws et al.] Professor Sue Buckley has researched this area extensively. Many reports are published in Down Syndrome Research and Practice. [See References, Section Z, Internet Resources.] Gynecology: Sexually active women should have a cytologic screening (Pap smear) every 1-3 years, starting at the age of first intercourse. Those women who are not sexually active should have a single-finger "bimanual" examination with a finger directed cytologic screening every 1-3 years. Screening transabdominal pelvic ultrasound every 2 to 3 years for women who have a baseline bimanual examination but refuse to have or have inadequate follow-up bimanual examinations of adnexa and uterus. Yearly mammograms for women over 50 years of age. Begin yearly mammograms at age 40 for women with a first-degree relative with breast cancer. [Adapted from Heaton CJ, "Providing reproductive health services to persons with Down syndrome and other mental retardation." See References, Section Q, for full reference.)

Neurodevelopmental Issues: The frequency of seizure disorders in persons with Down syndrome is greater than that seen in the general population, but lower than in persons with mental retardation due to other etiologies. Recent studies report an incidence of 5-10%. There appears to be a relationship between age and seizure prevalence in Down syndrome, with the peaks occurring in infancy and again in the fourth or fifth decade. There also appears to be a smaller peak in adolescence. Infantile spasms are the most common type of seizures seen in infancy and usually are well controlled with either steroids or other anticonvulsants. They generally have a favorable cognitive outcome, compared with the general population. Tonic-clonic seizures are most commonly seen in older persons with Down syndrome, and they respond well to anticonvulsant therapy in most cases. The increased incidence of seizures is not thought to be solely the result of abnormal brain development, but can be related to cardiac defects, infections, and irregularities of one or more neurotransmitters.

Attention Deficit Hyperactivity Disorder (ADHD) occurs in individuals with Down syndrome in the same frequency as it does in the general population of individuals with mental retardation. In both cases, this is more frequent than in the general population. In general, children with Down syndrome respond well to stimulant therapy. There is no research to indicate that children with Down syndrome respond any differently to stimulant medication than children with other etiologies of mental retardation, who respond, in general, very well.

Autistic disorders appear to be more prevalent in children and adults with Down syndrome. Whereas the incidence of autism in the general population is reported at 13 per 10,000 population, current evidence suggests that the prevalence in Down syndrome is approximately 5 to 10%.. [See References, Section X, Cohen & Patterson.]

Neuropsychiatric Disorders, including Alzheimer Disease: Changes in behavior and decline in intellectual and functional capabilities usually leads the caregivers of persons with Down syndrome to consider the possibility of a psychiatric disorder. After excluding any medical reason(s) for the behavior, the individual should be evaluated by a clinician who is skilled in assessing individuals with mental retardation and psychiatric disorders. There are potential limitations in diagnosing psychiatric disorders in persons with Down syndrome. Individuals with moderate or severe mental retardation generally are unable to accurately describe their thoughts and perceptions. Persons with mild mental retardation, however, may be able to accurately respond to questions about feelings, perceptions, and thoughts.

This section will focus on affective disorders, adjustment disorders, dementia (including Alzheimer disease), anxiety disorders, compulsive behavior. Attention Deficit Hyperactivity Disorder (ADHD) and autistic disorders are discussed in the preceding section.

The presenting symptoms may include one or more of the following: "decreased self-care, loss of skills in activities of daily living, loss of verbal skills, loss of social skills, loss of job skills, withdrawal, slow down in activity level, paranoid features, increase in talking to themselves, aggressive behavior, self-abuse, change in sleep patterns, weight change, and/or persistent forgetfulness." [See References, Section B, Chicoine B, et al. p. 103]

The major differential diagnosis is between depressive disorder and Alzheimer disease (dementia). Dementia is a neuro-psychiatric syndrome of memory loss that prevents new information from being learned and is characterized by a decline of intellectual skills which impairs social and/or occupational functioning. Alzheimer disease is a neurological disorder which is a progressive form of dementia which has certain characteristic changes in the structure of the brain. It results in a total inability to care for oneself, and, eventually, in death. A careful history must be elicited from caregivers to look for evidence of potentially reversible conditions, such as depression.

There has been great interest in the association of Alzheimer disease with DS for two main reasons. First of all, pathological study of the brains of individuals with DS reveal the characteristic neuropathological findings of plaques and neurofibrillary tangles. These changes can be found in relatively young individuals without signs and symptoms of AD. Secondly, Chromosome 21 contains the genes for amyloid precursor protein, and amyloid contributes to these pathological changes in the brains of individuals with Alzheimer disease (AD). DS is associated with other signs of early aging, and consequently these factors, in conjunction with functional decline in individuals with DS, suggested this was a very common problem. Current observers have noted that the original observations were cross-sectional studies conducted on institutionalized populations, and that this may be playing a role in the high incidence described. For example, Zigman et al. cogently note that "the neuropathological criteria for diagnosis of Alzheimer disease are not strongly correlated to clinical expression of symptoms." Prasher's group, involved in a longitudinal study, noted that "age-related decline in adaptive skills does not equate to Alzheimer's disease." If individuals are in good physical health, there is usually no decline. This certainly validates the experience of Chicoine and McGuire. Out of 443 adults seen at their Clinic, 148 presented with a decline in function. And only 11/148 met the criteria for progressive and nonreversible decline and deterioration and would therefore merit the diagnosis of AD. This translates to 2.5% of the total 443 patients. [See References, Section X.]

The signs of depression in typical individuals usually consist of a sad, irritable mood, along with disturbances of appetite, sleep, and energy, and loss of interest in previously enjoyable activities. Persons with Down syndrome are more likely to present with skill and memory losses, significant activity slowdowns, and hallucinatory-like self-talk and more extreme withdrawal (psychotic features). Persons with Down syndrome often develop depressive disorders in reaction to loss: death of a family member, change in a roommate, retirement of a caregiver from a group home, etc.

In general, the presentation of most psychiatric disorders tends to be more extreme, making the diagnosis more difficult. For example, an anxiety disorder may be manifested by self-injurious behavior or hyperactivity. Adjustment disorders to stressors may likewise include more severe or dramatic symptoms, such as self-injury, reversal of sleep patterns, and anorexia.

Schizophrenia and psychotic disorders occur very infrequently in persons with Down syndrome in spite of the widespread use of anti-psychotic medication.

Self-talk is common and usually developmentally appropriate, given the cognitive levels of these individuals. Although obsessions are rare, compulsive behaviors occur quite commonly.

Treatment is available for most of these disorders. This treatment may consist of pharmacologic agents, psychotherapy, and/or behavior therapies. It is important to stress that treatment should be under the direction of an individual who is skilled in addressing psychiatric disorders in individuals with mental retardation.

Kishnani and her colleagues at Duke University report that the use of the acetylcholinesterase inhibitor donepezil (Aricept) improved "communication, expressive language, attention and mood stability" in four adults, ages 24, 27, 38, and 64. The two older patients met diagnostic criteria for dementia. This pilot study suggests that this treatment may be great value to adults with DS in general and as well as those with AD. The drug had been given for 6 months with no significant side effects observed. A large-scale double-blind placebo controlled study is being planned.

Unconventional and Controversial ("Alternative") Therapies: Over the years, a number of controversial treatments of therapies have been proposed for persons with Down syndrome. Sometimes, such modalities are referred to as "alternative" therapies, meaning that they are outside of the mainstream of traditional medicine. Often the claims made in support of such treatments are similar: that the treatment will result in improved intellectual function, alter physical or facial appearance, decrease infections and generally improve the well-being of the child with Down syndrome. Nutritional supplements including vitamins, minerals, amino acids, enzymes and hormones in various combinations represent one form of therapy. There are a number of well-controlled scientific studies that have failed to show any benefit from megadoses of vitamins and minerals. Supplemental zinc and/or selenium may have an effect on immune function or susceptibility to infection, but studies thus far have been inconclusive. Sicca cell treatment (also called cell therapy) consists of injections of freeze-dried fetal animal cells, and has not been shown to be of any benefit. It also has potential side effects of allergic reactions and the risk of the transmission of slow virus infections.

There has been much interest generated in 1995 in the use of a Piracetam, a drug that is classified as a cerebral stimulant or nootropic. It has been tried in adults with Alzheimer disease without any benefit. It was shown to improve the reading abilities of typical boys with dyslexia. Piracetam is not approved by the Federal Drug Administration for use in the United States except as an orphan drug for myoclonus. At the time of its initial popularity there had been no scientific studies published reporting its use in children with Down syndrome. DSMIG has expressed concerns about its use in young children in the absence of studies demonstrating its safety. The first double-blind placebo-controlled crossover study of Piracetam, conducted by Lobaugh and her colleagues, was reported at the Pediatric Academic Societies Annual Meeting in San Francisco CA on May 3, 1999. Twenty children with DS were studied. There was no improvement in either cognitive or behavioral measures. There were significant central nervous system side effects noted which led the researchers to conclude that "it is unlikely that larger doses can be tolerated." [See References, Section Y, Lobaugh et al.]

Facilitated communication is a technique whereby a person known as a "facilitator" assists a person by providing support to the hand or arm to enable them to communicate using some type of communication keyboard. Although there are claims of usefulness for persons with many types of disabilities, a number of carefully designed studies have not established this as a valid treatment.

Some parents choose to include chiropractic care in the spectrum of interventions for their children with Down syndrome. The scope of the chiropractic services offered to children includes musculoskeletal manipulations, recommendations for supplemental vitamins, and agents purported to improve immunologic function. The range of conditions claimed to be amenable to chiropractic treatment is broad and includes constipation, gastroesophageal reflux, and ear infections. Individuals with Down syndrome have ligamentous laxity and therefore may be at increased risk of injury from cervical-spinal manipulation. Parents should be very cautious when considering such treatment, especially if it is promoted in lieu of immunizations, antibiotics for infections or hormone replacement for endocrine deficiency.

The treatments mentioned in this section are only a few of the approaches that have been tried or claimed to pose some benefit to children with Down syndrome. So far, there are no alternative medical therapies that have been scientifically documented to result in a significant improvement in the development and health of children with Down syndrome. Recently, members of DSMIG have received many anecdotal reports of significant and satisfying changes in a wide variety of functional areas (eg. muscle tone, sleep, general health, etc.) following the institution of the use of nutritional supplements. We are carefully evaluating these reports in order to be able to formulate a thoughtful plan to address the questions voiced by the parents of children and adults with Down syndrome about the value of these supplements.

Facial plastic surgery has been promoted in a number of countries, especially Israel, as a means of altering some of the physical features of Down syndrome. This is particularly controversial when performed on young children, since the facial features naturally undergo changes into adolescence. It is claimed that children are better accepted by society. This is not a medically indicated procedure, and most health insurance will not reimburse the surgeon or hospital. Tongue reduction surgery has also been promoted to improve esthetic appearance. Often this is recommended under the pretext of improving speech intelligibility. Several studies have demonstrated that this surgery has no effect on speech/language abilities nor the articulation of sounds. [See References, Section Y.]

Selected References

[Note: A selected list of current papers on Down syndrome is published in each issue of *Down Syndrome Quarterly*. This feature is edited by David Smith MD. (DS Center of Wisconsin in Milwaukee WI.)]

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* Z. Internet Resources

1. National Organizations

National Down Syndrome Society: www.ndss.org

National Association for Down Syndrome: www.nads.org

National Down Syndrome Congress: www.ndsccenter.org

2. Journals

Down Syndrome Quarterly

Down Syndrome Research and Practice, information.downsed.org/library/periodicals/dsrp/

Disability Solutions. www.disabilitysolutions.org

3. General Information

Down Syndrome: Health Issues. www.ds-health.com/

Foot Notes

¹The Down Syndrome Medical Interest Group (DSMIG) was founded in early 1994 with the express purpose of serving as a forum for professional addressing aspects of medical care of persons with Down syndrome. Bonnie Patterson, M.D. and William I. Cohen, M.D. serve as co-chairs. *DSMIG* wished to promote the highest quality care for Children and adults with DS 1) by fostering and providing professional and community education; 2) by disseminating tools for clinical care and professional support; such as these Health Guidelines; 3) and by engaging in collaborative clinical research regarding issues related to the care of individuals with Down syndrome. DSMIG schedules its meetings with a variety of national organizations, such as the National Down Syndrome Congress (NDSC), the National Down Syndrome Society (NDSS), the American Association of Mental Retardation (AAMR), etc. For more information on DSMIG, contact William I. Cohen, M.D. at Children's Hospital of Pittsburgh, 3705 Fifth Avenue, Pittsburgh, PA 15213 (412-692-7693 or cohenb@chplink.chp.edu).

²American Academy of Pediatrics, **Guidelines for Health Supervision III**, Elk Grove Village, IL, 1997.

³Recommended Childhood Immunization Schedule, United States, January-December, 1999. Approved by the Advisory Committee on Immunization Practices (ACIP), The American Academy of Pediatrics (AAP), and the American Academy of Family Physicians (AAFP).

⁴Available on the internet at www.growthcharts.com

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