

Medical Concerns Associated with Down Syndrome

Although many newborns with Down syndrome do not have accompanying medical issues, it is true that DS presents a significantly increased risk for a number of associated medical concerns. Some of these issues are briefly summarized below. For more specific descriptions, physicians should go to www.ndss.org, click onto "Health" in the left-hand column, and find more extensive information on specific medical conditions of interest.

Cardiac conditions

Forty-six to sixty-two percent of persons with DS are born with some form of cardiac defect. Significant defects are typically discovered and repaired during the first year of life. Despite this early intervention, some children with this type of history may require continuing cardiac care throughout their lives.

Dermatological disorders

Skin problems are common in young people with DS. These may be related, in part, to immunologic factors (as in alopecia). Skin texture may be dry and rough. Up to 70% of individuals with DS are affected by xerosis (abnormally dry skin) by the teenage years. Healthcare counseling should address preventative measures such as good hygiene, weight control, use of antibiotic ointments and emollients.

Immunological disorders

Persons with DS are at an increased risk for several immune / infectious disorders, including juvenile rheumatoid arthritis, alopecia areata, celiac disease, deficiencies of cellular and humoral immunity, immunoglobulin deficiencies (such as low IgG Subclasses 2 & 4, with elevated 1 & 3, found in individuals with serious pyogenic infections), and frequent ENT infections such as nasopharyngitis/sinusitis/otitis media. Thyroid dysfunction in DS may be related to autoimmune factors as well.

Musculoskeletal conditions

Persons with DS are at high risk for musculoskeletal abnormalities, including ligamentous laxity and atlantoaxial instability. Fifteen percent of individuals with DS develop the latter. Of these, 1-2% will develop subluxation and cord compression in this area. Clinicians should note that there is a higher incidence of individuals developing atlantoaxial instability during the rapid growth of adolescence.

Ophthalmologic Disorders

Ophthalmologic disorders are very prevalent among individuals with DS. Of these the most common is strabismus, affecting 23 – 44 percent of individuals with DS. Cataracts may also occur frequently in individuals with DS. Due to the increased incidence of

these disorders in persons with DS, a thorough ophthalmologic exam is recommended with each check up.

Thyroid dysfunction

Thyroid disorders are prevalent in persons with DS, with hypothyroidism occurring much more frequently than hyperthyroidism. Autoimmune factors may play a role in the etiology of hypothyroidism in DS, although the exact relationship is not completely understood. Studies have demonstrated that thyroid hormone levels may gradually decrease during the adolescent years. Symptoms of thyroid dysfunction may thus become more evident during this time. Thyroid dysfunction may have implications for multiple systems; cardiovascular, immune, and central nervous system effects are frequently seen. Thyroid hormone levels should thus be monitored at least annually in affected individuals.

Young Adulthood: Behavioral/Emotional Concerns

As children with DS grow, it becomes apparent that they all have their own unique personalities. Historically, persons with DS have been stereotyped as having placid, amiable, dispositions. Recent studies reflect much more heterogeneity in individual temperaments, and, as one would expect with unique personalities in any population, some individuals with DS develop behavioral or emotional issues. No area of problem behavior or emotional instability seems to be specific to DS, however. Disruptive behavior disorders are the most frequently reported, with an incidence of between 11 and 12 percent. Depression may also occur, especially within adolescence and young adulthood. Both difficulties may be precipitated and/or exacerbated by communication deficits. To optimize the effectiveness of therapeutic treatment for behavioral or emotional disorders, it is best to consult a professional/therapist knowledgeable about these issues in patients with developmental disabilities.

Protective Genes On The 21ST Chromosome

Trisomy 21 is protective in two ways; first, a gene present related to homocystine protects against atherosclerotic heart disease. Therefore, it is rare for a person with DS to have a heart attack or stroke unless heart defects have not been corrected. Second, the 21st chromosome houses a tumor suppression gene. Except for testicular cancer, people with DS do not develop solid tumors.

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