

DOWN SYNDROME

A REVIEW FOR DENTAL PROFESSIONALS

Purpose of this Module

The information presented in this module is intended to provide the institutional dental staff with an in depth review of the medical and dental implications of Down Syndrome and suggestions for future treatment and preventive regimens based upon this detailed knowledge.

Learning Objectives

After reviewing the enclosed information the participant shall be able to:

1. Describe the three genotypes of Down Syndrome.
2. Discuss the incidence of Down Syndrome, particularly related to maternal age.
3. Discuss the relationship of Down Syndrome to Alzheimer Disorder and the problem of atlanto-axial instability.
4. Describe the characteristics of mid-face dysplasia common to individuals with Down Syndrome.
5. Discuss the issues and impact of congenital cardiopathies common to persons with Down Syndrome.
6. List and discuss three additional medical conditions common to persons with Down Syndrome.
7. Discuss in detail, the periodontal and oral hygiene issues involved with Down Syndrome.
8. Describe the malocclusion problems common to Down Syndrome.
9. Describe the effects of partial glossectomies, adenotonsillectomies and cosmetic surgery on persons with Down Syndrome.
10. Describe six treatment approaches to the periodontal problems associated with Down Syndrome.
11. Describe the impact of cardiopathies and upper respiratory infections on the treatment of persons with Down Syndrome.
12. Discuss the issues impacting prosthetic services for individuals with Down Syndrome.

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INTRODUCTION

The institutional dental staff will encounter patients with Down Syndrome on a daily basis. Therefore, appropriate that the staff be familiar with the medical and dental implications of this condition. Although generalizations are helpful to better understand the impact of Down Syndrome on the provision of dental care, it is important to also be aware of the wide variance in expression of the various aspects of this genetic syndrome within individuals.

DEFINITION AND ETIOLOGY

Down Syndrome, Trisomy 21 or Mongolism, was first described by Dr. Langdon Down in 1865. At that time, the diagnosis of the syndrome was based solely on physical findings. Surprisingly, it was only in 1956 that the normal complement of 46 human chromosomes was determined and it was not until 1959 that it was demonstrated that Down Syndrome was associated with an extra chromosome of the twenty first group, for a total of 47 chromosomes. Note, the number and structure of chromosomes within an individual cell is referred to as the karyotype of a cell. The term mongolism, mongoloid or mongoloid idiot are outdated and stigmatizing terms and should not be used to describe persons with this condition.

The etiology of Down Syndrome relates to the problem of nondisjunction of a 21 chromosome during oogenesis, thus an extra 21 chromosome is provided to the offspring by the mother. Recent studies also implicate paternal etiology through nondisjunction during spermatogenesis.

There are three types of Down Syndrome, although it is generally thought that there is no clinical difference in the three genotypes.

(1) **Trisomy 21** (94%): The extra 21 chromosome (three instead of the usual two) produces a complement of 47 chromosomes. Trisomy 21 may also be referred to as Trisomy G.

(2) **Translocation** (5%): A segment of a 21 chromosome is found attached to other pairs of

chromosomes (usually #14, thus referred to as a 14/21 translocation). These individuals have the normal complement of 46 chromosomes.

(3) **Mosaicism** (1%): Nondisjunction occurs at a later stage of cell division, therefore, some cells have the normal complement of 46 chromosomes and other cells 47 chromosomes (with an extra 21 chromosome).

INCIDENCE AND ASSOCIATION WITH MENTAL RETARDATION

The incidence of Down Syndrome for all ages is one in 700 live births and increases with the age of the mother: age 20 (1 in 2300) age 34-39 (1 in 280) age 40-44 (1 in 130) age 46 (1 in 65) and age 54 (1 in 54). Although there are some persons with Down Syndrome with an IQ above 69, nearly all persons with this condition are mentally retarded. All ranges of mental retardation (MR) can occur but most typical is moderate/severe retardation (IQ 20-50).

FACIAL CHARACTERISTICS

Midface dysplasia is a cardinal characteristic of persons with Down Syndrome (DS). Nose malformations including a flat broad bridge of the nose has been reported in 59-78% of these individuals. Ear malformations, including "lop" ears, low-set ears and ears with a flat or absent helix have been reported in 54%. Eye malformations are common. Epicanthal folds with slanting almond-shaped eyes (narrow palpebral tissue slanting toward the midline), which was responsible for the term mongoloid, are reported in 78%. Strabismus (cross eyes) is reported in 14-54% and nystagmus (constant involuntary cyclical movement of the eyeballs) and refractive errors are also common. The majority of persons with Down Syndrome exhibit brachycephaly (broad, short head) and lack of supraorbital ridges and hypotelorism (secondary to hypoplasia of the central face) are common findings. Absence of frontal sinuses and absent or reduced maxillary sinuses have

been reported. Nasal septum or nasal conchal deviations are often observed which can produce a partially obstructed or narrow air passage and can contribute to the problem of mouth breathing.

MEDICAL CONDITIONS

O Congenital Cardiopathies

A 40% incidence of mitral valve prolapse (MVP) has been reported in persons with Down Syndrome. The diagnosis of MVP and aortic regurgitation (AR) is much higher when echocardiograms are used. In one study, a medical history showed 6% of subjects with DS (versus 2% of the controls) had aortic regurgitation and 14% of the DS subjects (versus 4% of the controls) had mitral valve prolapse. When echocardiograms were obtained on these subjects with Down Syndrome the incidence of AR rose to 11% and that of MVP rose to 57%. A connective tissue abnormality (collagen defect) may be a possible explanation of the high incidence of MVP in these clients. The need for antibiotic prophylaxis in these clients is obvious. The incidence of AR, MVP, hypertension, or arteriosclerosis does not appear to rise with increasing chronological age in these individuals.

O Upper Respiratory Tract

Infections-hepatitis-leukemia

The higher incidence of upper respiratory tract infections (URTI) seen in persons with Down Syndrome is thought to be due to their impaired immunologic response to infectious or inflammatory diseases. Prior to the age of antibiotics, most of these individuals died at an early age from pneumonia. The increase in URTI contributes to mouth breathing and speech problems and along with nasal septal deviations, may complicate the use of general anesthesia.

This immunologic impairment increases the incidence of a carrier state and lowers the incidence of immunity in individuals with Down Syndrome exposed to the Hepatitis virus. A recent study in Georgia showed 27% of institutionalized subjects with Down Syndrome and 7% non Down Syndrome, mentally retarded subjects were positive for HBsAg. However, the incidence in 37 non-institutionalized subjects with Down Syndrome were similar to the general population. The incidence of leukemia (unrestrained growth of leukocytes) has been reported as

15-50 times more prevalent in the population with Down Syndrome compared with the general population.

O Alzheimer Dementia

Many studies show that virtually 100% of persons with Down Syndrome over 35 years old develop neurological signs of Alzheimer disorder, as evidenced by post-mortem findings. Yet many of these individuals live long lives with no clinical signs of dementia. Some studies show no signs of intellectual deterioration occurring prior to 50 years in 50% of Down Syndrome subjects and no relationship between chronological age and presence of disturbed behavior. The discrepancies between neurological and clinical findings may be explained by the evidence that although the neurological changes seen in persons with Down Syndrome and Alzheimer dementia are similar, the regional distribution within the brain is different.

Incidentally, the life expectancy of persons with DS, although shorter than the general population, has increased markedly over the past fifty years (1929: 9 years versus 1980: 30 years).

O Atlanto-axial Instability

The incidence of atlanto-axial instability in persons with Down Syndrome has been reported as ten to twenty percent. This condition refers to an abnormal increase in mobility of the upper two cervical vertebra (C1/C2) due to congenital ligamentous laxity. What percentage of instances of instability leading to actual dislocation, which can lead to severe spinal cord injury, is unknown. Although some sources recommend early (5-6 years) clinical and radiographic evaluation in these individuals, others claim that instability leading to dislocation is not well founded and that radiographs are not predictive in this regard. This problem of ligamentous laxity apparently decreases with age compared with an age related increase in degenerative cervical arthritis which is also commonly seen in these individuals. Ligamentous laxity can affect other parts of the body producing a hyperflexibility of all joints. This problem together with general muscle hypotonia can produce the shuffling gait often seen in persons with Down Syndrome.

O Other Medical Problems

- **Speech:** The speech problems commonly noted are related to the central motor deficit and degree of mental retardation rather than a peripheral articulation problem. Delayed speech and a husky quality of voice are common findings in these individuals.
- **Hearing:** Studies show that persons with DS have a significantly higher level of hearing impairments with an incidence of 77% reported. This hearing impairment, usually mild, is related to smaller ear canals and consequent impacted cerumen.
- **Eye problems:** There is an apparent increased risk for cataracts in these individuals.
- **Epilepsy/Cerebral Palsy:** There is no increased incidence in these conditions for persons with Down Syndrome.
- **Other Problems:** Persons with Down Syndrome usually are below average in height and often have a stooping posture. Obesity and sexual underdevelopment are common. Dry rough scaly skin is also common, and a single palmar crease (simian crease) is seen in 45% of these individuals.

DENTAL CONDITIONS

Although many of the earlier and classical studies reporting dental conditions in persons with Down Syndrome were institutional based and often poorly controlled, these findings have been consistently supported in more recent investigations.

O Periodontal Disease/oral Hygiene

Inadequate oral hygiene has been a universal finding in the institutional based studies. There is no evidence that institutional or community based persons with Down Syndrome experience a different level of oral hygiene than other persons with mental retardation. Also there has been reported no difference in the presence of calculus in these persons. However, a severe, early onset, often dramatic fulminating periodontal disease is a universal finding with an incidence of 90-96% in these individuals. It is common to see alveolar bone loss in persons with DS age 6-16 years. This increased incidence of periodontal disease is, therefore, not related to an increased amount of plaque or more virulent plaque;

but thought to be directly related to the reduced immunologic response to infections and inflammatory disease reported in these individuals. The issue of ligamentous laxity/ degeneration may also play a part in the destruction of the periodontal ligament. This defect in host response has been reported in many controlled studies showing greater inflammation and cellular response occurring with equal plaque levels. The clinical course in individuals with DS is similar to juvenile periodontitis, except it is not isolated to a few teeth. Other evidence supports this issue of deficits in host response being responsible for the periodontal problems of persons with Down Syndrome. Several reports have indicated a high incidence of acute necrotizing ulcerative gingivitis (ANUG) in these individuals. One study showed 45% of institutional DS subjects versus 4% of the non DS/MR controls with evidence of ANUG. This condition historically has been associated with alterations in host response, often stress related. Oral conditions associated with an increase in ANUG in these individuals were: crowded dentition (vs spacing), traumatic occlusion, peg shaped anterior teeth, lack of root resorption in primary teeth and incidence of high frenum attachment. The clinical picture of ANUG in persons with Down Syndrome differs from the usual symptoms in that fetid breath and exquisite pain are rarely reported.

O Caries

Older studies reported a dramatically lower decay rate in the population with Down Syndrome compared with controls (53% caries free vs. 0.5% in controls). More recent studies continue to support the lower decay rate in persons with Down Syndrome, but the difference is shown to be far less than previously reported. Overall reduction in dental decay and controlling for eruption times may explain the differences in the earlier and more recent studies. Incidentally, the delay in eruption of permanent teeth has been reported as only 0.7 years.

O Malocclusion

There is an increased incidence of malocclusion in individuals with DS, particularly Class III malocclusions. The higher incidence of Class III malocclusions is due to the underdevelopment of the midface (nasal, premaxillary and maxillary bones) not to prognathism. The increased incidence of all malocclusions has been reported as: Class III, 32-70%;

Class II, 3-32%; posterior unilateral and bilateral cross bites, 71%; and open bites, 5%. Contrary to many medical reports, a high arched palate is not a characteristic of this population; the palatal height and shape is normal but the palate is relatively small in all dimensions including a decreased arch length and arch circumference.

Microdontia, especially in the mesio-distal dimension is common. The incidence of missing permanent laterals has been reported as high as 35-43% compared with 2% in the general population. Rotated teeth (especially centrals), spaced teeth (especially lower bicuspid), peg shaped teeth (especially laterals) and more congenitally missing permanent teeth are common findings.

Additional findings indicate more over retained primary teeth (especially primary cuspids and second molars), delayed eruption of permanent teeth, more impacted teeth (especially cuspids and bicuspid) and more variable tooth morphology (especially nipples cuspids and shovel shaped maxillary incisors).

There is evidence reported that both crowns and roots, but especially roots, of permanent teeth in this population are shorter than normal, thus creating an unfavorable crown-root ratio. Increased taurodontism has also been reported in persons with Down Syndrome. Taurodontism together with abnormally short roots would reduce the extent of periodontal ligament attachment and would be expected to contribute to tooth mobility so commonly seen in these persons.

O Other Dental Problems

The incidence of macroglossia has been reported as 11-60% in persons with Down Syndrome although the presence of true macroglossia has been questioned by some investigators. There is agreement, however, on the presence of a relative macroglossia due to the small palatal space and hypotonic tongue. Fissured tongue and protruding tongue due to forward position of the mandible and open mouth is a common finding. Also, increases in bifid uvula and submucous clefts and cleft palates have been reported in this population.

Reports on the differences in parotid gland secretions are conflicting. Earlier studies showed parotid gland secretions with an elevated PH (alkaline saliva) and elevated sodium, calcium and bicarbonate levels which may contribute to the documented

lower decay rate. More recent studies, however, have found no PH differences in the saliva of DS subjects compared with other MR subjects or the general population.

GENERAL TREATMENT CONSIDERATIONS

Down Syndrome is a permanent genetic disability. The general management of persons with this condition including medical and educational management will not be included in this review. Also, management of maladaptive dental behaviors for those individuals with DS and behavioral problems is covered in Modules 2, 5 and 6. There are, however, some medical issues that are germane to the dental care of these individuals.

O Immune Response and Vitamin Therapy

Although it may seem unusual to dentists and physicians, there has been considerable interest on the part of parents and other professionals in pursuing mega-vitamin therapy in addressing the lowered immune response to infection seen in persons with Down Syndrome. All studies, however, conclude that short or long term administration of various vitamin therapies have no effect on any aspects of Down Syndrome.

O Partial Glossectomies or Adenotonsillectomies

Many recent studies support the positive outcomes of partial glossectomies in persons with Down Syndrome with relative macroglossia. These positive outcomes include: less tongue protrusion, better speech and pronunciation (due to higher pitch with mouth closed)(45%), less upper respiratory tract infection (83%), increased ability to maintain a closed mouth (60%), better mastication and drinking, better self confidence and pleased parents (95%). The dentist may well be involved as a consultant on this issue. Similarly, there are reports on the positive outcomes of adenotonsillectomies in persons with Down Syndrome. These include: improvement in sleep apnea (83%), reduced mouth breathing (91%), reduced snoring and nasal drainage (75%). In absence of nasal obstruction, no improvement in tongue protrusion was noted.

O Cosmetic Facial Surgery

There is considerable controversy regarding the advantages and disadvantages of cosmetic surgery to reduce the stigma of Down Syndrome and to aid in social development. These procedures would include augmentation of the nasal, cheek and chin areas, glossectomies and lateral canthoplasty. Proponents, usually parents, support the surgery as a normalizing procedure. Opponents contend that the accompanying mental retardation is the main disability and cosmetic surgery is too radical and serious a procedure for the minor gains expected.

DENTAL TREATMENT CONSIDERATIONS

As with cerebral palsy, there is a paucity of information in the literature regarding treatment options for persons with Down Syndrome. The clinician must employ an innovative approach to dental care for these individuals based upon the data provided in the previous section entitled "Dental Conditions." The following information is derived from the literature and suggestions presented by the authors.

O Periodontal Disease and oral Hygiene Problems

- **Documentation:** Early documentation of developing periodontal disease, especially bone loss and pocket formation, is important. It is quite unusual for most dentists to contemplate documentation of periodontal disease onset and advancement at such an early age (6-16). Yet when confronted by a parent or advocate at the time when extraction of a permanent tooth has been recommended for an 18 year old patient with Down Syndrome due to advanced bone loss, proper documentation of the disease progression and therapies attempted assumes greater importance for the dentist.
- **Communication:** Early communication with parents/guardians regarding the limitations of dental care in preventing tooth loss and other dental sequela including prosthetic limitations is important. It is at this time that the responsibility for meticulous oral hygiene and the support and development of acceptable dental behaviors by the parents/caretakers can be

emphasized. Early diet counseling can also be of benefit in preventing the obesity and other diet problems commonly encountered in this population.

O Early, Aggressive and Innovative Periodontal Therapy

It may be helpful to the clinician to employ a treatment approach to patients with Down Syndrome with actual or anticipated periodontal problems similar to the treatment approaches advocated for patients with acquired immunodeficiency syndrome (AIDS). That is, early aggressive, innovative, and often untried approaches based upon knowledge of the host response problems may prove fruitful in addressing these unusual conditions.

- Topical antimicrobial agents (e.g., peridex®, Listerine®) may be indicated on a long-term basis for these individuals. The ability of the individual to rinse and expectorate may be a limiting factor. Other delivery methods such as gels or sprays may be helpful.
- Systemic antimicrobial agents, particularly tetracycline may be helpful. Long term tetracycline therapy (10-30 years) is common in the field of dermatology, yet the ADA statement that "long term antibiotic use in dentistry is considered experimental" certainly creates a barrier for the dentist.
- Early preventive periodontal therapy, including vestibular extensions, frenectomies and grafts should be considered. The major problem is the lack of data confirming the success of periodontal therapy in the presence of an altered immune or host response. Long term studies documenting the success or failure of such therapies should not be expected in the near future. Thus the clinician in consultation with the patients' parents/guardians should rely on clinical judgement and reasonable expectations in recommending an early aggressive surgical approach. The dental cooperation of the patient is an important and limiting factor in this regard. Since host response is impaired and post surgical healing may be prolonged, the use of antibiotics following even minor surgical procedures may be indicated. There is some evidence that some persons with Down Syndrome have a peripheral circulation problem with an abnormal capil-

lary or vascular system which could affect the healing and success of autogenous gingival grafts. Early and aggressive therapy, where there is evidence of ANUG or similar dental papilla destroying periodontal disease, is important. Since over retained primary teeth are associated with increased ANUG and periodontal disease in these patients, close attention to extraction of primary teeth showing reduced root resorption rates should be considered. Also, since crowded dentition is associated with increased periodontal problems, selected extraction of primary or permanent teeth or radical enameloplasty to create interdental spacing is another consideration.

- The use of bone grafts or bone replacement products (e.g. hydroxyapatite) may hold promise for future use. Aggressive and radical occlusal equilibration to reduce occlusal interferences and to reduce crown height producing a more favorable crown-root ratio, should be strongly considered. A flat plane occlusion may be helpful in overcoming the periodontal impact of Class III malocclusions and posterior cross bites. Early orthodontic intervention with palate expansion and cross bite correction may reduce subsequent adverse impact of expected malocclusion. Restoration of pegged shaped cuspids with cast or composite restorations may also contribute to improved periodontal health.

O Prosthetic Treatment

Prosthetic treatment choices can be very limited with this population. The existence of periodontal disease and tooth mobility can severely limit the choice of fixed or removable partial dentures. The lack of cooperation seen with many patients with Down Syndrome also renders these choices problematical. Complete dentures are often not a viable treatment choice due to lack of retention (caused by severe periodontal disease), presence of severe class III jaw relationship and lack of cooperation by the patient. Relative lack of maxillary sinuses in some of these patients would theoretically aid in use of implant technology, however the issues of host response and patient cooperation usually preclude this treatment choice. It is imperative that the limitations of prosthetic replacement be fully explained to the parent/guardian early in the treatment planning

stage, to prevent inappropriate expectations in this regard.

O Medical Considerations

- Attempts to refer a patient with Down Syndrome for a complete cardiac evaluation, including an echocardiogram, may prove fruitful if client cooperation allows. If a cardiac evaluation is not available, routine antibiotic coverage of these patients may be a reasonable alternative.
- The effects of frequent upper respiratory tract infections, reduced size of nasal passage due to septal deviation and mouth breathing on the use of sedation in persons with Down Syndrome should be considered. Special attention to patient positioning and sedation monitoring, especially with pulse oximetry, is indicated.
- The hearing loss experienced by many of these individuals should be taken into consideration when communicating with the patient. All of the techniques employed to overcome difficulties presented by a hearing impairment in other individuals would be appropriate for these persons.

SUMMARY

The provision of dental services to the person with Down Syndrome who is severely/profoundly retarded presents unique challenges to the institutional dental staff. A thorough knowledge of the unusual dental implications of this syndrome and an innovative problem solving approach to treatment planning and preventive procedures will do much to alleviate the dental effects of this handicapping condition.

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