

Orofacial Anomalies and Treatments in People with Down Syndrome

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Introduction

Down syndrome (DS) is a genetic disorder resulting from overexpression of genes on the twenty-first chromosome. In 95% of the cases, this is caused by an extra chromosome on the 21st pair. The rest of the cases are due to a translocation of part of chromosome 21 to another one, usually chromosome 14, or post-fertilization non-separation of chromosomes resulting in mosaicism (the 21st chromosome is not found in all cells). DS is one of the most common genetic abnormalities, affecting approximately 1 in 700 to 800 live births. Despite the development of prenatal diagnosis, the incidence of DS births is predicted to remain stable or even to increase.¹

Care for DS can be complicated and involves a myriad of acute and chronic medical problems and psychosocial issues. Some of the medical problems include congenital heart defects, gastrointestinal defects, global muscle hypotonia (a decrease in muscle tone), loose joints/ligaments, upper respiratory infections, various orthopedic problems, and endocrine disorders (particularly hypothyroidism). Many of the medical and physiological characteristics of DS have direct consequences on the oral health of people affected and indirect consequences on the quality of life of persons with DS and their caregivers. This article aims to give an overview of the current literature concerning the orofacial, or mouth and face, motor anomalies in persons with DS and their implications, and to explain the available treatment options.

Orofacial Structural Anomalies

The different anatomical aspects of DS have been well described in the literature and form the basis of the orofacial problems experienced by this population. The primary skeletal abnormality affecting the orofacial structures is an incomplete development of the midfacial region.^{2,3} The bridge of the nose and bones of the midface and upper jaw are relatively smaller in size.³ This underdevelopment of the midface results in reduction of the width and depth of the palate.⁴ The muscles of mastication and facial expression are hypotonic, and there may be laxity of the temporomandibular joint ligaments.⁵ The reduced muscle tone in the lips and cheeks contributes to an imbalance of forces on the teeth, with the force of the tongue being of greater influence.³

Soft-tissue features include a fissured and protrusive tongue that often rests between the dental arches and high against the palate.^{2,5} The tongue appears macroglossic, or enlarged, due to the relatively small size of the oral cavity.^{2,3,5} Tongue protrusion and thrusting during drinking, eating, and speaking is reported in the presence of a hypotonic tongue.² Several occlusion elements are noticed: higher frequency of malalignment², malocclusion, and anterior/posterior cross bite.^{2,4} There is often a severe, Class-III malocclusion^{3,4} which contributes to an anterior open bite due to abnormal tongue position. These classically described features vary significantly between individuals despite the typical faces of DS.

Functional Implications

The functional manifestations of these abnormalities are directly related to the underlying structural defects. Abnormal oral structure and physiology compromise the development of feeding, chewing, swallowing, and speech capabilities.

Feeding

Studies have shown that children with DS take longer to develop the motor coordination necessary for normal feeding.⁵ A retrospective chart review of 49 DS children showed that 80% had problems related to food or feeding.⁶ The literature suggests that the development of oral-motor function in children with DS not only lags behind intellectually but also follows an irregular pathway.⁷ Since numerous feeding problems occur in infants, studies addressing the variety of emotions experienced by caregivers indicated that feeding therapy and counseling were required. Because of the different conditions underlying the feeding problems of infants, a comprehensive approach, including therapeutic and medical intervention, is necessary.⁸ Interdisciplinary intervention programs have proven successful, as most of the nutritional, behavioral, and environmental problems surrounding food previously encountered in children with DS were prevented or remedied.⁶

Chewing

Several studies have found that persons with DS are not able to produce normal chewing movements. Specific aspects of tongue and jaw function were impaired along with problems in initiating and maintaining a smooth sequence of feeding actions.⁷ In particular, Hennequin, et

al. investigated differences in chewing indicators (masticatory time, number of masticatory cycles, number of open masticatory cycles, chewing frequency, and number of food refusals) in a group of 11 adults with DS and compared with a control group. With the exception of puree, DS persons had significantly lower mean chewing frequency than the reference group and were unable to eat all foods presented.⁸ Another study investigated nutritional status and age of introduction of solid food. It concluded that DS delays the age at which solid foods are introduced, which can be deleterious to oral motor development, and recommended pre-speech therapy.¹⁰ Video recordings of 4- to 5-year-old children with DS eating a standard sized meal show that DS children were characterized by a forward placement of tongue in the mouth and the absence of normal maturational changes in the oral cavity.¹¹

Swallowing

Swallow function in children with DS has been assessed using video fluoroscopy in conjunction with foods of different texture. Frasier, et al. reviewed swallowing behavior of 19 children with DS and identified abnormal pharynx movements during swallowing, with aspiration occurring in 10 of the 19 children studied. Silent aspiration was shown to be a problem among this population with liquid or semi liquid food. Recurrent aspiration contributes to high incidence of pulmonary infection.¹²

Speech

Kumin collected data from 937 parent questionnaires regarding intelligibility of speech in children with DS and found it to be a widespread problem.¹³ Parents reported evidence of difficulties classified as oral motor skills, motor programming skills, and specific speech skills. Children experienced greater difficulty with reciting sentences and engaging in conversation than with reciting single words. Intelligibility problems were more frequent when the child was conversing with unfamiliar adults.¹³ Another study found that one of the factors that affected speech intelligibility of children with DS was difficulty with voluntarily programming, combining, organizing, and sequencing the movements necessary for speech. Historically, this difficulty, childhood verbal apraxia, has not been identified or treated in children with DS but recent research has documented that symptoms of childhood verbal apraxia can be found in children with DS. Results indicated that children with DS who have clinical symptoms of childhood verbal apraxia have more difficulty with speech intelligibility, i.e. there was a significant correlation between childhood verbal apraxia and parental assessment of intelligibility ratings. Children with apraxia often do not begin to speak until after age five.¹⁴

Prevention and Therapeutic Options

Different therapies to correct orofacial problems can be grouped into four categories: neuromuscular stimula-

tion, behavior modification, orthodontic intervention, and surgical intervention. Orofacial therapy of neuromuscular stimulation has been studied extensively. It includes physiotherapy of the oral structures and an appliance to stimulate the lips and tongue. The appliance is a palatal plate designed according to Castillo-Morales. The main goal is to increase muscle tone around the mouth and enhance the development of oral function.⁵ It is accomplished by establishing a resting position for the tongue behind the front teeth, leading to improvement in swallowing, chewing, and articulation. Multiple studies concluded that early intervention methods employing a combination of Castillo-Morales therapy and his palatal plate could significantly improve orofacial function, facial appearance and speech, as well as prevent dental diseases, malocclusion, and open mouth. Early therapy is recommended, starting at age 6-8 months.^{18,19,20} The effects of the Castillo-Morales stimulating plates were followed by a longitudinal study up to 53 months after the end of treatment, and long-term results showed improvement in orofacial appearance- even when the plate was not in place, the result remained stable in 65.8% of the patients.¹⁵ Hohoff, et al. studied the effects of the palatal plates on speech development, and demonstrated an enhancement of DS children's oral motor capacity and initial speech development.¹⁶ Methods of behavior modification include parental reinforcement of the desired behavior and the use of tactile or audio cueing aids.⁵ Functional orthodontic therapy is also useful but highly dependent on the full cooperation of the child. The use of surgical modalities, including glossectomy (removal of part of tongue), tonsillectomy (removal of tonsils), and plastic surgery, are controversial. Few studies have evaluated the esthetic appearance and intelligibility of speech after partial glossectomy and found no significant difference in acoustic speech intelligibility; in some patients, there was an esthetic improvement during speech.¹⁷

Conclusion

Children affected with DS display a variety of orofacial anomalies that influence their feeding, chewing, swallowing, and speech. Children diagnosed with DS should be exposed to early interdisciplinary intervention therapies, which include behavior/speech therapy and neuromuscular/orthodontic treatment in an effort to minimize the effects of the anomalies. A team of medical specialists should regularly work with the caregivers and the child to identify and prevent functional problems.

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References

1. B. Goodwin, C. Huether, *Prenatal Diagnosis*. **7**, 261 (1987).
2. S. Desai, *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology, and Endodontics*. **84**, 279 (1997).
3. E. Pilcher, *Down Syndrome Research and Practice*. **5**, 111 (1998)
4. G. Jensen, J. Cleall, A. Yip, *American Journal of Orthodontics*. **64**, 607 (1973).
5. M. Hennequin, D. Faulks, J. Veyrone, P. Bourdiol, *Developmental Medicine and Child Neurology*. **41**, 275 (1999).
6. P. Pipes, V. Holm, *Journal of the American Dietetic Association*. **77**, 277 (1980).
7. Q. Spender, A. Stein, J. Dennis, S. Reilly, E. Percy, D. Cave, *Developmental Medicine and Child Neurology*. **38**, 681 (1996).
8. E. Lewis, A. Kritzinger, *Down Syndrome Research and Practice*. **9**, 45 (2004).
9. M. Hennequin, P. Allison, D. Faulks, T. Orliaguet, J. Feine, *Journal of Dental Research*. **84**, 1057 (2005).
10. E. Hopman, C. Csizmadia, W. Bastiani, et al., *Journal of the American Dietetic Association*. **98**, 790 (1998).
11. E. Gisel, L. Lange, C. Niman, *American Journal of Occupational Therapy*. **38**, 666 (1984).
12. J. Frazier, B. Friedman, *Developmental Medicine and Child Neurology*. **38**, 695 (1996).
13. L. Kumin, *Perceptual and Motor Skills*. **78**, 307 (1994).
14. L. Kumin, *Down Syndrome Research and Practice*. **10**, 10 (2006).
15. A. Hohoff, U. Ehmer, *Journal of Orofacial Orthopedics*. **60**, 2 (1999).
16. A. Hohoff, U. Ehmer, *Journal of Orofacial Orthopedics*. **58**, 330 (1997).
17. P. Klaiman, M. Witzel, F. Margar-Bacal, I. Munro, *Plastic and Reconstructive Surgery*. **82**, 403 (1988).
18. B. Backman, A. Grever-Sjolander, et al., *International Journal of Paediatric Dentistry*. **17**, 19 (2007).
19. G. Schuster, R. Giese, *Journal of Orofacial Orthopedics*. **62**, 255 (2001).
20. G. Limbrock, R. Castillo-Morales, H. Hoyer, et al., *International Journal of Orofacial Myology*. **19**, 30 (1993).

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