

DOWN SYNDROME

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ABSTRACT

The purpose of this report is to share background information about the complex nature of abnormalities associated with Down syndrome, and to offer clinical guidelines that may help orofacial myologists determine whether or not to treat individuals with Down syndrome.

INTRODUCTION: With good intentions, it is tempting to try to apply the procedures of orofacial myology to a variety of patient samples where considerable specialized training and experience is a necessary prerequisite. Such is the case with children with Down syndrome. While the obvious signs of a tongue forward rest posture, with mouth open, and lips apart, serve to characterize individuals with Down syndrome, the myriad of other characteristics and problems associated with this genetically-based abnormality merit careful evaluation prior to designing and implementing a treatment plan to correct orofacial myofunctional disorders (OMDs). To provide orofacial myology services for children with Down syndrome without appropriate training and without the inclusion of language stimulation in therapy, is a disservice to those patients. Children and adolescents with Down syndrome who are candidates for orofacial myofunctional therapy should be thoroughly evaluated in a team setting, and therapy for OMDs should be dispensed by highly qualified personnel who have considerable information about the syndrome and also have a clear view of the language capabilities and delays and cortical processing competencies and deficiencies involved with individuals with Down syndrome.

Judging from the number of orofacial myologist websites that claim expertise with individuals with Down syndrome, there is apparently considerable interest within the field regarding the role that an orofacial myologist can play with such patients. Some clinicians who are speechlanguage pathologists and have training and previous experience with language delay and language stimulation, are qualified to provide services for young patients with Down syndrome, all of whom need language stimulation in addition to any OMD treatment that may be recommended. For teenage patients whose language issues have been resolved, it may be appropriate for some dental hygienist orofacial myologists to provide selected treatment for OMDs.

PURPOSE: The purpose of this report is to share background information about the complex nature of abnormalities associated with Down syndrome, and to offer clinical guidelines that may help orofacial myologists determine whether or not to treat individual individuals with Down syndrome.

BACKGROUND: Down syndrome, also referred to as Trisomy 21 Syndrome, is one of many craniofacial conditions characterized by midfacial retrusion. In 1866, John Langdon Down provided the clinical description of the syndrome. While many continue to refer incorrectly to this syndrome as "Down's" syndrome, the use of a possessive form of the eponym is discouraged since Down did not have the disorder, nor did he claim ownership of it. With an incidence of 1 in 660 births, Down syndrome ranks as the most common malformation pattern in man, and due to the OMD symptoms involved, is of interest to orofacial myologists. The etiology of Down syndrome is trisomy for all or a large part of chromosome 21 (Jones, 2006). A trisomy is a genetic abnormality in which there are three copies instead of the normal two for a specific chromosome; in this case, chromosome 21. There is, as expected, considerable variation seen in the expression of the disorder, except for the general features of mental deficiency and short stature.

A diagnosis of Down syndrome can be made based on the total pattern of anomalous findings. It would be highly unusual to find a given anomaly in 100% of all patients with the syndrome. Full 21 trisomy (an entire extra 21 chromosome copy) occurs in 94% of patients, while 21 trisomy/mosaicism, or translocation cases are found to occur only in about 3 to 4% (Jones, 2006). Such individuals have the features of Down syndrome but have a milder phenotype of trisomy. The recurrence risk for Down syndrome is about 1% (Jones, 2006). As is well known, a faulty chromosomal distribution leading to Down syndrome is more likely to occur at an older maternal age.

SOME SPECIFICS OF DOWN SYNDROME: The genetic condition of Trisomy 21 Syndrome has many characteristics that have been detailed in texts of human malformations. The reference text by Jones, 2006, serves as the primary resource for the genetics and characteristics of the syndrome discussed in this report.

The specific abnormalities associated with Down syndrome include a tendency to keep the mouth open and to protrude the tongue. There is generalized muscle hypotonia, hyperflexibility of joints; a relatively small stature with awkward gait; increased weight in adolescence; mental deficiency; and several craniofacial abnormalities including brachycephaly, mild microcephaly with upslanted palpebral fissures, either hypoplasia or aplasia of frontal sinuses, short hard palate, a small nose with a low nasal bridge, and the tendency to have inner epicanthal folds (Jones, 2006). The nasal abnormalities severely limit the ability to achieve a successful nasal pattern of breathing. Other abnormalities (Jones, 2006) involve the eyes (there are many abnormalities including peripheral hypoplasia of the iris); ears (conductive, mixed, or sensorineural hearing loss [66%]); otitis media (60% to 80%); dentition (hypoplasia of teeth, Class III skeletal malocclusion with underbite, irregular placement of teeth, and periodontal disease); neck (short); hands (a variety of shortness and variations in the fifth fingers); feet (wide gap between first and second toes); pelvis (hypoplasia); cardiac (an anomaly in approximately 40%); skin (loose folds in neck, hyperkeratotic skin); hair (fine, soft and sparse); and genitalia (primary gonadal deficiency and small genitalia). The primary features of interest to orofacial myologists are maxillary retrusion, a small nasopharynx, a normal size tongue in a small oral cavity, a deficiency in cognitive ability, and difficulty maintaining a nasal breathing pattern.

NATURAL HISTORY: Although muscle tone in Down syndrome individuals tends to improve with age, the rate of developmental progress tends to slow with age. Although the cognitive ability range is said to be from 25 to 50, with an average IQ around 50, an occasional few have an IQ well above 50. Fortunately, the social performance of Down syndrome children usually exceeds their mental age (Jones, 2006).

Early developmental enrichment programs for Down syndrome children have resulted in an improved rate of progress during the first 4 to 5 years of life (Jones, 2006). This finding reinforces the view that any treatment for OMDs with young children with Down syndrome should also include language stimulation and thus, be provided only by those orofacial myologists with specialized training in speech-language pathology. Whether such therapy will appreciably improve speech and language performance remains to be seen.

Another part of the natural history with Down syndrome pertains to sleep-related issues. Upper airway obstruction occurs in approximately one third of cases (Jones, 2006). Overall craniofacial and body growth is relatively slow, and secondary centers of skull ossification are often late in development during the first 8 years. Final overall body height is achieved around age 15 (Jones, 2006). In 1997, the median age at death for patients with Down syndrome was 49, and the major cause was congenital heart defects (Jones, 2006). Mortality is also much higher than in the general population from respiratory diseases such as pneumonia, and other infectious diseases.

MAXILLA: Maxillary retrusion indicates that the upper jaw is small, mostly in the horizontal plane. A small maxilla and hard palate contribute to a reduction of the area of the nasal cavity for establishing and maintaining a nasal pattern of breathing.

NASOPHARYNX: The nasopharynx is also reduced from a normal size due to the retruded maxilla, but as well, the angle of the cranial base is somewhat acute in many patients. The result is that the posterior wall of the pharynx is positioned closer to the posterior entrance into the nose (posterior choanae), so the presence of any adenoid tissue can interfere with the flow of air in or out of the nose. The consequence of this, of course, is that children with Down syndrome will move the tongue forward as a means of maintaining the airway to make up for the reduction in size of the nasopharynx and nasal cavity. The reduced size of the posterior choanae and the nasopharynx itself combine to encourage an oral pattern of breathing. In addition, large (faucial) tonsils, when present, compete with the space for the posterior tongue and further encourage a tongue forward rest posture. Mouth breathing is obligatory for most children with Down syndrome.

TONGUE: Children with Down syndrome typically have an open mouth posture and a protruding tongue that appears to be most prominent in early childhood. The tongue protrusion is related to a combination of several factors: a smaller than normal oral cavity due to the retruded upper jaw (maxilla), enlargement of the posterior portion of the tongue near the tonsils, and a lack of normal muscle tone in the tongue (Leshin, 2000). Subtelny, an orthodontist, described the tongue in Down patients as being a "relative macroglossia"; that is, a tongue of normal size housed in a small oral cavity (Subtelny, 1970). The tongue appears macroglossic, or enlarged beyond normal proportions, because it protrudes, but the mandible to which it is attached is normal in size and yet, the maxilla to which it also relates is small. The oral cavity is therefore small because of the retruded, small maxilla and hard palate.

Since the tongue is attached relative to the mandible and hyoid bone, the tongue remains normal size in Down syndrome patients rather than adapting to the size of the maxilla. In other skeletal Class III conditions where the mandible is large and is surgically retro-positioned, the tongue adapts and appears smaller following surgical retraction of the mandible. Preoperatively, there is the appearance of a large tongue, while postoperatively, the tongue appears smaller as it adapts to the newly created and reduced size of the mandible. With children with Down syndrome, a normal sized tongue and mandible, coupled with a small nasopharynx and maxilla, result in the tongue resting and functioning forward. This protruded Rest position helps to maintain the airway. A major role of the tongue, of course, is to maintain and protect the airway. There is yet another reason why the tongue is forward in individuals with Down syndrome. A feature of Down syndrome is a variable level of mental deficiency, often accompanied by a neuromotor maturation delay for speech and other oral functions. It is well known that the most primitive movement pattern of the tongue is a protrusion and retraction in the horizontal plane. Even children with frank neurological disorders have no difficulty in protruding the tongue; however, such children have difficulty with controlled elevation of the tongue tip for speech and other tasks. Children with Down syndrome exhibit flaccid tongues, and do not perform well on oral diadochokinetic tasks, exhibiting difficulty with rapid vertical tongue movements. Since the starting position for speech is abnormal with a protruded tongue at rest, teaching correct speech sound productions presents a special challenge for the speech-language clinician.

SURGERY FOR CHILDREN WITH DOWN SYNDROME: (Note: this section credits Len Leshin, MD, as the primary source of information). Part of the history involving the treatment of Down syndrome includes attempts to surgically reduce the size of the tongue. A tongue reduction, also called a "partial glossectomy" consists of removing a wedge of tongue to make it smaller and shorter (Mason & Serafin, 1984). Some physicians and surgeons have recommended a tongue reduction for aesthetic reasons, presuming that it would help keep the lips together (Olbrisch, 1985; Wexler et al, 1986; Champion et al, 1992). The problem with such tongue reductions is the noble but misguided decision to modify a symptom of the syndrome (tongue protrusion) rather than modifying the cause - either the reduced size of the airway at the nasal cavity or nasopharynx. In the 1980s and early 1990s, there was a period of enthusiasm among some surgeons for reducing tongue size in children with Down syndrome to achieve either cosmetic or speech intelligibility improvement. Some reports of improved speech were found to be subjectively evaluated by the surgeons who performed the procedure (Olbrisch, 1985; Lemperle & Radu, 1980; Wexler et al, 1986). Two studies that used objective criteria reported no improvement in speech intelligibility following tongue reduction surgery (Parsons, et al, 1987; Margar-Bacal et al, 1987). The assumption that reducing tongue size would retro-position the tongue and that lip closure would naturally follow was not affirmed following surgery. The problem was that the small nasopharynx and nasal cavity

were not addressed, so the tongue soon returned to its presurgical and forward rest position rather than being retracted or adapting otherwise. Since children with Down syndrome have generalized hypotonia, this characteristic may have contributed to the lack of spontaneous lip closure following partial glossectomy.

The surgical history with individuals with Down syndrome also includes attempts to improve facial aesthetics. Strauss et al (1988) contended that children with Down syndrome are subjected to a decreased level of acceptance by their peers, solely on the basis of facial features. The goal of improving the appearance of the face led to plastic surgical procedures (including a partial glossectomy) that involved placing implants in the zygomatic (cheek) bones; modifying the appearance of the eyes by removing the excess skin at the corner of the eyes (epicanthal folds); reorienting the slanted palpebral fissures to a more horizontal position; elevating a down-turned lower lip; recontouring the flat nasal bridge; and removing fat from the neck (Leshin, 2000). Several post-operative studies demonstrated that the parents involved were pleased with the results of the plastic surgery (Lemperle & Radu, 1980; Olbrisch, 1982; Arndt et al, 1986; Wexler et al, 1986). However, studies have not demonstrated a significant positive impact on the social functioning of the children who underwent surgery (Katz & Kravetz, 1989; Kravetz et al, 1992). Other studies have also disputed the assumption that the facial appearance of children with Down syndrome has a negative effect on how they are perceived (Pueschel et al, 1986; Cunningham et al, 1991). Although surgery has not been found to have a wide, predictable and positive impact on the facial appearance, speech intelligibility or social functioning of children with Down syndrome, it continues to be an area of interest, especially among parents. The risks and benefits of plastic surgery with children with Down syndrome should be clearly and thoroughly discussed with those parents of Down syndrome children who desire surgical management for their child (Leshin, 2000).

CLINICAL APPLICATIONS: Orofacial myologists that embrace the optimistic and admirable view that orofacial myofunctional therapy can retract a protruded tongue, achieve a lip-together rest posture and maintain a nasal pattern of breathing in individuals with Down syndrome, should undertake a thorough clinical evaluation of such patients that includes, at a minimum, lateral and frontal cephalometric x-ray films to evaluate the posterior airway, the size of the adenoids, and the faucial tonsils. A friendly orthodontist would be needed to do the cephalometric study and help the orofacial myologist to evaluate the morphology of interest.

Because the anatomy seen on x-ray films does not adequately predict functions, especially breathing, an assessment should also include, at a minimum, airway functions and interferences and the ability of the patient to breathe intranasally. In addition, an assessment of cognitive abilities should be obtained and a detailed history of other associated abnormalities and problems should be identified and considered in any treatment planning related to OMDs.

It is no secret that children have to be reminded to blow their noses. A key issue for determining a child's candidacy for therapy with has Down syndrome's involves *nasal hygiene*. Poor nasal hygiene combined with abnormalities in the nasal cavity and surrounding anatomy and a reduction in cognitive function can combine to compromise the ability of patients in monitoring and maintaining patency of the nasal cavity. Keeping the nasal chambers open and free of the debris that interferes with nasal respiration presents a special challenge in establishing and maintaining

a nasal breathing pattern in children with Down syndrome. If an airway issue persists, there will most likely be no success achieved with orofacial myofunctional therapy.

In evaluating a child with Down syndrome, an intraoral exam of the posterior airway is mandatory. Can the posterior pharyngeal wall be clearly observed? What is the range of elevation of the soft palate? Does the velum elevate to the plane of the hard palate? Altogether, and with regard for the characteristics discussed here, the role of the orofacial myologist with Down syndrome patients would seem very negligible. Those advocates for such therapy are encouraged to report well-documented studies of long-term stability that may follow from any claims of short-term successes.

CLINICAL TREATMENT PERSPECTIVES: With an appropriate regard for patients served by orofacial myologists, one of the worst things that a clinician can do is underestimate a child's ability. By like token, it should be considered unethical to waste a child's time in therapy when no progress can be logically expected, nor is it good practice to offer false hope to parents with unsupported claims of success with children with Down syndrome. A rule to live by, and hopefully practiced by all orofacial myologists, is: It is better to over-estimate a child's abilities and cognitive capacity than to underestimate and find out later that you were wrong. Such is the diagnostic dilemma with individuals with Down syndrome. A thorough evaluation in a team setting is indicated to properly determine a child's candidacy for therapy.

The development of a treatment strategy for those speech-language pathologists who may work with language stimulation and the OMDs of children with Down syndrome should involve a decision as to whether to work on the deficits that a child possesses, or work on the strengths. In the case of Down syndrome, children's tongues function well in the horizontal plane of space; they can easily protrude the tongue although they have difficulties with controlled vertical movements of the tongue. In light of the ability to protrude, a reasonable target for linguoalveolar sounds is the back of upper teeth rather than the maxillary alveolus. In some cases, even using the lingual surfaces of lower incisors is an appropriate target, since "t,d,n,l" can be made with acoustic acceptability with the tongue tip at the lower incisors.

In spite of the obligatory breathing need for children with Down syndrome to protrude the tongue in the early years, this may change as the mandible grows. Orofacial growth in the teenage years involves vertical, transverse, and horizontal growth of the jaws and vertical growth of the pharynx. During the teenage years, individuals with Down syndrome may have the ability to retract the tongue at rest by adapting to

the growth increases in the vertical plane. Working to increase muscle tone at the lateral surfaces of the tongue may also aid in repositioning the tongue at rest. Teenage patients may also be able to respond favorably to exercises to achieve lip competence. As a result, overall appearance may improve.

When a clinician is unsure of the value of therapy, a period of *diagnostic therapy* may be appropriate. Such therapy recognizes that there may be delimiting factors that will cause a clinician to cease and desist therapy at some point. It is better to try and fail than not try and be wrong. This should be a caveat with determining whether myofunctional therapy is appropriate for individuals with Down syndrome. In spite of a sincere desire to help, however, the decision whether or not to enroll patients with Down syndrome in therapy should be made only after a thorough evaluation of each patient. Therapy should be dispensed only by those who are well qualified to provide such services. Generally, orofacial myologists cannot claim any expertise to work with children and adolescents with Down syndrome. Speech-language pathologists, however, can provide services, especially if working within a team. Dental hygienists, as part of the practice of dental hygiene, may also provide appropriate dental services with Down syndrome patients such as cleaning teeth, etc. Myofunctional therapy is not a magic pill that can positively impact all conditions.

QUESTIONS AND ANSWERS REGARDING DOWN SYNDROME:

Q: Regarding the small nasopharynx: Is the nasopharynx small at birth? Yes.

Q: Regarding the narrow pharynx: How is it narrow? Is it front to back or side to side or just overall. A: Primarily front to back, but with some narrowing laterally. But each patient is different, so this is only a general reply.

Q: Children with Down syndrome, if I remember correctly, have a broad neck. Is the pharynx wider in children with Down syndrome, with the narrowing only in the front to back wall relationship? If so, how much could the side to side wider dimension compensate for the narrowness for breathing? A: Determining how a child "compensates" presents a host of variables that would be very difficult to identify. The width of the pharynx changes along the pharyngeal tube, and even throughout the nasal cavity, so a constriction at one point may change according to posture, temperature, and other influencing factors.

Q: Regarding the tongue adapting to the environment: I understand the tongue readapting to the environment after reduction surgery. But even without surgery if the lips were closed and the rest posture established, wouldn't the tongue also adapt to that environment with a reduction in size, or become a smaller than normal sized tongue? A: The airway problem in the nasal cavity and nasopharynx requires the tongue to rest forward, so until and unless the source of the airway problem is resolved, you would not expect to see effective and wellmaintained adaptations in tongue size and position.

Q: Would tongue surgery results have been better maintained if the children who underwent surgery were able to maintain a closed lip rest posture and proper tongue rest posture? A:

Yes, but this does not occur because the midfacial retrusion and reduced size of the pharynx were not corrected -- and it is not feasible to surgically correct an anteroposterior narrowing of the nasopharynx that is a consequence of an acute cranial base angle.

Q: Should each child who may have OMD's be evaluated as a candidate for therapy by his/her mental ability and capacity to respond to therapy. A: Yes!

Q: Is it appropriate for an orofacial myologist to advise or otherwise help with feeding problems with infants with Down syndrome? A: No. The specialized training of those clinicians who participate in feeding regimes with clefts, other malformations and conditions, or even normal infants, is neither a part of orofacial myology training, nor is this an area where expertise can be claimed. Being an orofacial myologist neither qualifies a clinician as a feeding expert nor as a specialist who should treat any conditions from infancy to four years of age. However, feeding problems may be addressed by those orofacial myologists with specialized early feeding training in speech-language pathology.

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- KEY WORDS:** Down syndrome; Trisomy 21 syndrome; maxilla; nasopharynx; tongue; relative macroglossia; lingual reduction surgery.