

TITLE: Pediatric Syndromes of the Head and Neck

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More than 3,000 known syndromes have been classified in humans. The evaluation and management of children with these syndromes requires the appropriate knowledge and support of multiple specialists. Otolaryngologists are often consulted in the diagnosis and management of the syndromal child. The pattern of disease in these children may often be first recognized by the astute otolaryngologist. The focus of this presentation is to identify several common syndromes affecting children, and the otolaryngologic significance of these syndromes.

Definitions

Prior to the discussion of various head and neck anomalies found in the syndromal child, a thorough understanding of various terms must be appreciated. The following terms are used in the description of anatomic variations to the norm:

- **Deformation:** An abnormality form, shape, or position of a part of the body resulting from mechanical forces, usually occurring late in gestation. The flattening of facial features is an example resulting from oligohydramnios.
- **Malformation:** A morphologic defect of an organ, part of an organ, or a larger region of the body caused by an intrinsically abnormal developmental process. Anencephaly, as an example, results from abnormal development of the brain.
- **Disruption:** A morphologic defect of an organ, part of an organ, or larger region of the body caused by extrinsic breakdown of, or an interference of, an originally normal developmental process. Facial clefting is a disruption resulting from amniotic bands.
- **Association:** A non-random occurrence in two or more individuals of multiple anomalies not known to originate from a syndrome, sequence, or disturbance of single developmental field. For example, the CHARGE association has multiple anatomic abnormalities that are not known to be related in their origin.

- **Syndrome:** A pattern of multiple anomalies pathogenetically related but not representing a single sequence or developmental field. Branchio-otorenal syndrome is an example that consists of branchial cleft, ear, and renal anomalies resulting from disturbances in the development of multiple organ-systems.
- **Sequence:** A pattern of multiple anomalies derived from a single known or presumed prior anomaly or mechanical factor. The primary abnormality of the Pierre Robin sequence is micrognathia, which results in glossoptosis. Glossoptosis contributes to cleft palate in this syndrome.

Diagnosis of the Syndromal Child

The first and usually the most critical step in the evaluation of a child with a known or suspected syndrome is a thorough history. This includes the medical history of the child as well as the family members of the child. A pedigree should be constructed to develop an understanding of the genetic attributes of the family. Information about the age and consanguinity of the parents is necessary. Exposure to known teratogens during pregnancy should be elicited.

The next step is to perform a careful physical examination, noting all minor and major abnormal features. Another important step is to compare various features of the child to the other family members. The otolaryngologic manifestations of various syndromes include anomalies of the ears, cranium, facial features, oropharynx, and neck. Audiologic evaluation is also obtained when appropriate. It is important for an otolaryngologist to acknowledge anomalous features of the entire body, and not be limited to the head and neck. This includes the skeletal structure and features of the hands and feet.

When a child is suspected to be afflicted by a syndrome, a consultation with medical genetics experts is recommended for identification of the genetic anomaly and testing for other abnormalities. Communication among the parents and all medical specialists involved is essential for the appropriate care of the syndromal child.

Otolaryngologic Concerns in the Syndromal Child

The Airway

Understanding the development of the airway and anatomic variations from normal are critical to the otolaryngologist. Newborns are obligate nasal breathers until about 4-6 weeks. Mouth breathing is a learned response. Congenital nasal obstruction or stenosis presents a life-threatening problem. Choanal atresia and midface hypoplasia may present in the neonate as respiratory distress with cyanosis. This is usually effectively initially treated with an oral airway until further surgical management is planned. Micrognathia, retrognathia, glossoptosis, and macroglossia may also significantly obstruct the airway which may be first treated by a nasal airway and prone feeding, and further with adenotonsillectomy when appropriate. A tracheotomy is indicated when other measures have not been successful. In some instance, support and growth of the child is sufficient. Mandibular osteotomies and distraction may be required for malocclusion, when growth is insufficient.

The management of the airway provides a challenge to medical personnel, and it is the responsibility of the otolaryngologist to attain and maintain an appropriate airway for a child with craniofacial anomalies. Communication with the anesthesiologist is critical when operating on a syndromal child. Evaluation of the airway includes examination of the entire airway. This may be achieved by flexible laryngoscopy, direct laryngoscopy, and tracheobronchoscopy. Stertor and the type of stridor provide clues to the level of airway obstruction.

Appropriate management of the pediatric airway requires familiarity with the instruments needed to evaluate and care for the airway. The appropriate size, length, and type of laryngoscope and bronchoscope for each case should be evaluated prior to performing direct laryngoscopy. As a general rule, all instruments should be connected and checked prior to the start of an airway case.

The otolaryngologist may be involved in the management of obstructive sleep apnea in a child with craniofacial anomalies. It is important to understand that functional anomalies contribute to the problem of airway obstruction when developing treatment strategies. Polysomnography is essential in the diagnosis of obstructive sleep apnea. Continuous positive airway pressure may be beneficial in some cases. The management of OSA may require adenotonsillectomy, uvulopalatopharyngoplasty, partial tongue base resection, resection of redundant laryngeal structures, or tracheotomy.

Hearing Loss

Children with craniofacial anomalies are at risk of having or developing hearing loss. Hearing is essential for learning language and verbal communication, as well as education. Therefore, the careful evaluation of the syndromal child includes an appropriate otologic and audiologic examination. Delay in the detection of hearing loss may be very deleterious in the development of the child.

Congenital hearing loss is predominantly conductive in nature, but sensorineural hearing loss may also be involved. Congenital conductive hearing loss may be secondary to microtia, external auditory canal atresia, and ossicular deformity or fixation. In these instances, an appropriate audiologic evaluation with radiography is required prior to surgical correction. If these problems are unilateral, appropriate language development is possible if the hearing in the unaffected ear is optimized. Early detection of otitis media and administration of antibiotics or placement of ventilation tubes, when appropriate, are necessary in the normal ear. Eustachian tube dysfunction (ETD) is common in children with craniofacial anomalies, especially cleft lip/palate, and usually is effectively treated with placement of ventilation tubes. PE tubes may be required into adolescence to prevent hearing loss and complications from ETD.

Speech Disorders

Speech disorders are common in children with craniofacial anomalies, especially in children with nasal obstruction and cleft palate. Hypernasal speech is a common finding in velopharyngeal insufficiency (VPI) following cleft palate repair. Speech therapy is usually sufficient; however, palatopharyngoplasty may be required for children with persistent VPI over the age of 5 years. Hyponasal speech is less common and results from nasal obstruction, which

is usually surgically correctable. Hoarseness is another speech abnormality usually due to the development of vocal cord nodules in compensatory laryngeal activity. This may also result from intubation trauma.

Down Syndrome

Down syndrome is the most common syndrome known in humans. It was first described in 1866 by John Landon Down. The prevalence has been estimated as high as 1 in 700 humans. Approximately 95% of cases are due to nondisjunction of chromosome 21 in gametogenesis. The remaining cases are from unbalanced translocations. There is a high association with increased maternal age. Maternal age of 33-35 carries a 2.8 in 1,000 risk, and maternal age of greater than 44 years carries a 38 in 1,000 risk. There is a 1% risk of having a child with Down syndrome if a sibling has this syndrome. Screening methods have been developed and include ultrasonography, alpha-fetoprotein level, human chorionic gonadotropin and unconjugated estriol levels. The average life expectancy of individuals with Down syndrome is 35 years, with highest mortality early in infancy due to congenital heart defects, leukemia, and respiratory distress.

Common signs in a newborn with Down syndrome include hypotonia, poor Moro reflex, hyperextensible joints, loose skin on nape, flattened facial profile, upward slanting palpebral fissures, single palmar crease, flat occiput, and epicanthal folds. Prenatal and postnatal growth deficiency is present in almost all cases. Interestingly, in older patients, Alzheimer disease is common. This is believed to be due to an abnormality of amyloid beta-A4 precursor mapped to chromosome 21.

The craniofacial manifestations of Down syndrome include the absence of the frontal and sphenoid sinuses with maxillary sinus hypoplasia in 90% of cases, a flattened nasal bridge with relative mandibular prognathism (midface hypoplasia), small ears with overlapping helix, epicanthal folds and upward slanting palpebral fissures. Atlantoaxial instability is a common finding that must be acknowledged when manipulating the head and neck. Macroglossia with a fissured or geographic tongue is also common. Periodontal disease is found in 90% of cases with a relatively low incidence of dental caries.

The otolaryngologic concerns include airway obstruction and hearing loss. Due to midface hypoplasia, the nasopharynx and oropharynx dimensions are smaller and slight adenoid hypertrophy may result in upper airway obstruction. Obstructive sleep apnea is a very common finding ranging from 54-100% of cases and is due to a combination of anatomic and physiologic abnormalities. Hypotonia with macroglossia and midface hypoplasia contribute to the development of OSA. Polysomnography is diagnostic of OSA, and there are several management options available. Medical management including CPAP and weight loss when indicated is effective in some cases. However, surgical management is required when medical management is not effective. Adenotonsillectomy alone is a controversial approach due to hypotonia in addition to possible lymphoid hypertrophy. Uvulopalatopharyngoplasty and partial tongue resection are other surgical options. A tracheotomy is the most effective manner to bypass upper airway obstruction when other options have failed. It is also important to note that there is evidence of congenital mild to moderate subglottic narrowing in patients with Down syndrome. Therefore, post-extubation stridor is not an uncommon finding.

The otologic issues include a small pinna and stenotic EAC which contribute to cerumen impaction in Down syndrome patients. Conductive hearing loss may be secondary to chronic otitis media with effusion and Eustachian tube dysfunction. This may be addressed with placement of ventilation tubes. Ossicular fixation is also not uncommon, and may be surgically managed. Sensorineural hearing loss is present in relatively few cases, and is attributed to progressive ossification along outflow pathway of the basal spiral tract.

Cardiovascular anomalies in Down syndrome is present in 40% of cases and may range from ventricular septal defects, atrial septal defects, tetralogy of Fallot, and patent ductus arteriosus. Gastrointestinal involvement in 10-18% includes pyloric stenosis, duodenal atresia, and tracheoesophageal fistula. Of importance is the 20 fold higher incidence of acute lymphocytic leukemia in patients with Down syndrome compared to patients without Down syndrome. It is important to have all systemic issues addressed in the team approach to the management of Down syndrome.

Velocardiofacial Syndrome

Velocardiofacial syndrome (VCFS) is one of the most common syndromes involving the head and neck. The prevalence may be as high as 1 in every 4,000 births. Although patients may appear normal, they have characteristic facial structures. For these reasons, it is essential for otolaryngologists to be familiar with the facial anomalies and physiologic disturbances these patients may display. This syndrome typically has a manifestation of congenital heart disease, hypernasal speech, cleft palate, learning disabilities, and a characteristic facial appearance. An estimated 8% of cleft palate clinic patients have VCFS. The inheritance pattern is autosomal dominant with variable expressivity. In 85% of cases there is hemizygous microdeletion shared with the DiGeorge sequence at the 22q11.2 locus.

The oropharyngeal findings include structural malformations of the neck in 75% of cases, but may vary from readily apparent cleft palate (10-35%), submucous cleft (33%), occult submucous cleft and velar paresis (33%), and a hypernasal speech pattern. Malocclusion is a common finding. The tonsils and adenoids are small or aplastic in 50% and 85% of cases, respectively. Airway obstruction is not an uncommon finding and up to 50% of neonates are diagnosed with obstructive sleep apnea. However, it is very important to avoid tonsillectomy and adenoidectomy in these patients as the obstruction does not improve after this surgery. An oropharyngeal or a nasopharyngeal airway is useful in the urgent setting. Ultimately, repair of the cleft palate or surgical management of velopharyngeal apparatus may be required.

The facial characteristics in VCFS include microcephaly, a long face with vertical maxillary excess, malar flatness, and retrusion of the mandible. The nose is usually prominent with a squared nasal root, hypoplasia of the alae, and narrow nasal passages. The philtrum is usually long with a thin upper lip. Facial asymmetry is not uncommon. In addition, 15% of cases exhibit Pierre Robin sequence and 15% with Pierre Robin have VCFS. In 35-50% of cases, the palpebral fissures may appear narrow with allergic shiners. Ophthalmologic findings small optic disks, bilateral cataracts, tortuous retinal vessels, and rarely colobomas may be present. Anomalies of the ears are common and include small auricles with thickened helical rims. In 75% of cases, CHL is present and likely is due to serous otitis media and cleft palate. SNHL may be present in 8-15% of cases.

Cardiovascular anomalies are present in 75-85% cases of VCFS. Ventricular septal defects, right-sided aortic arch, aberrant subclavian artery, and Tetralogy of Fallot are common cardiac problems. Approximately 10% of infants with VCFS die as a result of congenital cardiac defects. The internal carotid arteries are medially displaced and tortuous in 25% of cases, but generally straighten with age. This should always be acknowledged prior to cleft repair.

Mild mental retardation and poor social interaction with a flat affect may be present in some individuals. Renal anomalies may be present in 35% of cases. Skeletal growth is also retarded in a large proportion of cases. Hypocalcemia and immunologic findings of T-cell dysfunction, allude to a relationship to the DiGeorge sequence in 15% of patients. Both, failure to thrive and frequent infections are present in this population.

The appropriate diagnosis and management of patients with VCFS requires a coordinated team effort of multiple specialties. The most critical factors include appropriate management of cardiac defects early in life and maintenance of good health through early childhood. Hearing and speech impediments must be appropriately managed, as well any disturbances to vision to allow for the greatest academic development. Communication between otolaryngologists, craniofacial surgeons, cardiologists, nephrologists, ophthalmologists, pediatricians, speech pathologists, teachers, and parents is essential in caring for patients with such diverse medical problems.

Branchio-Otorenal Syndrome

Branchio-otorenal syndrome (BORS) was first termed by Melnick et al in 1975. BORS has a prevalence of 1 in every 40,000 newborns and has an autosomal dominant inheritance pattern with high penetrance typically isolated to the gene at the 8q13.3 locus. The characteristics of this syndrome are most commonly branchial cleft cysts or fistulas, preauricular pits, malformed auricles, hearing loss and renal anomalies.

Branchial cleft cysts, sinuses, or fistulas are present in 50-60% of cases, predominantly found in the lower third of the neck, and are usually bilateral. Fistulas, when present, may open into the tonsillar fossa. Facial nerve paralysis and aplasia/stenosis of the lacrimal duct are not uncommon (10% and 25% of cases, respectively).

The otologic manifestations of BORS range from structural anomalies of the external ear to the inner ear. Auricular malformations are found in 30-60% of cases and vary from severe microtia to minor anomalies of the pinna. Helical or preauricular pits are present in 70-80% of cases, and rarely communicate with the tympanic cavity. Hearing loss is present in 75-95% of affected individuals, composed of conductive (30%), sensorineural (20%), and mixed hearing loss (50%). The onset of hearing loss may vary from early childhood to young adulthood. The middle ear anomalies include malformation and/or fixation of ossicles and abnormal size or structure of the tympanic cavity. In the inner ear, anomalies, although rare, include dilated vestibule and/or endolymphatic duct/sac, bulbous internal auditory canal, small semicircular canals, and hypoplastic cochlea.

The structural anomalies of the urinary system are present in 12-20% of cases. This is likely underreported as only 6% of those with renal involvement have severe symptoms. The

anomalies range from renal agenesis to mild hypoplasia or abnormalities of the renal pelvis or ureters.

Appropriate diagnosis of BORS is dependent upon a thorough history and physical examination. One must keep a high index of suspicion when ear anomalies, hearing loss, neck masses/sinuses, and renal problems are encountered. Initial management includes antibiotics for infected branchial cleft sinuses/cysts and an audiogram. Profound hearing loss has been found in 1 of 200 individuals with preauricular pits. Further management includes neck CT and possibly temporal bone CT if hearing loss is present. When clinically appropriate a renal ultrasound or intravenous pyelogram may be beneficial. Genetic counseling is beneficial for families.

The treatment for branchial cleft cyst/sinus/fistula is surgical excision to prevent repeated infection and possibility of airway obstruction or dysphagia. The external ear may also be addressed surgically, from microtia repair to simple excision of preauricular pits. Ossicular chain reconstruction may be performed, when indicated, to improve hearing. Hearing aid devices are used frequently when hearing is impaired. Consultation with urologic specialists is appropriate when there is renal involvement.

Treacher Collins Syndrome

Mandibulofacial dysostosis was first described by Thomson and Toynbee in 1846-1847. The essential elements of this syndrome were later described by Treacher Collins in 1960. It is a relatively uncommon syndrome with an incidence of 1 in 50,000 births, and is an autosomal dominant disorder with variable expressivity. The gene called Treacle or *TCOF1*, has been mapped to the 5q32-33.1 locus. Approximately 60% of cases are from new mutations, and have an association with increased paternal age. The pathogenesis is likely derived from abnormal migration of neural crest cells into the first and second branchial arch structures. The features of this syndrome are mostly bilateral and symmetric. The characteristics include supraorbital and malar hypoplasia resulting in relatively large appearance of the nose, a narrow face with downward sloping palpebral fissures, malformed pinna, receding chin, and relatively large downturned mouth.

In addition to malar hypoplasia and non-fused zygomatic arches, the paranasal sinuses are often hypoplastic. The mandibular components are also often hypoplastic, with a concave shape to the undersurface of the mandibular body. The angle of the mandible is also more obtuse than normal. Colobomas of the outer third of the lower eyelid and absence of the lower eyelid cilia may also be present. Cleft palate is found in 35% of cases with an additional 30-40% with palatopharyngeal incompetence.

Abnormalities of the airway include choanal atresia resulting in respiratory distress in the newborn. Obstructive sleep apnea is the most common breathing dysfunction and is frequently caused by mandibular hypoplasia that displaces the tongue posteriorly into the oropharynx. An oral airway may assist ventilation, but a tracheotomy may be performed if needed.

Otologic manifestations include a malformed pinna often misplaced toward the angle of the mandible. One third of patients with anomalous pinna have EAC atresia or ossicular abnormalities. Conductive hearing loss is common and must be addressed for normal

development. It is important to recognize that intelligence is usually normal in patients with Treacher Collins syndrome.

Apert and Crouzon Syndromes

Apert and Crouzon syndromes belong to the family of syndromes with craniosynostoses. Although each is unique, they share some characteristics.

Wheaton first described the features of acrocephalosyndactyly in 1894, but Apert expanded on this syndrome in 1906. Apert syndrome is characterized by craniosynostosis, midfacial malformations, and symmetric syndactyly of the hands and feet. The prevalence of Apert syndrome is 15-16 per million newborns and contributes to 4-5% of all craniosynostoses. It has an autosomal dominant inheritance pattern, but, most cases are sporadic from new mutations associated with increased paternal age.

The coronal sutures are fused in Apert syndrome at birth, with larger than normal head circumference. The cranial base is malformed and often asymmetric with a short anterior cranial fossa. Shallow orbits result in exophthalmos. The midface is retruded and hypoplastic in some cases, resulting in relative prognathism. The nasal bridge may be depressed and the nose is usually beaked. Hypertelorism, downward slanting palpebral fissures, proptosis, strabismus, and cleft palate are frequently associated.

In 1912, Crouzon first described the characteristics of craniofacial dysostosis. These features were craniosynostosis, maxillary hypoplasia, shallow orbits, and ocular proptosis. It is inherited in an autosomal dominant pattern with one third of cases reported to be sporadic. The prevalence of Crouzon syndrome is also 15-16 per million newborns and accounts for 4.5% of all craniosynostoses.

In Crouzon syndrome fusion of cranial sutures begins during the first year of life and usually complete by 2-3 years of age. Shallow orbits with exophthalmos at birth without involvement of the hands and feet are usually diagnostic for Crouzon syndrome. Midface retrusion, relative prognathism, hypertelorism and a beaked nose are also present as in Apert syndrome.

In both syndromes, the reduced nasopharyngeal dimensions along with choanal stenosis may result in respiratory embarrassment, especially in the newborn. Obstructive sleep apnea and cor pulmonale are associated with airway compromise. In these circumstances, adenoidectomy for hypertrophic lymphoid tissue, endotracheal intubation and tracheotomy may be needed. A polysomnogram is a useful tool for determining airway compromise. Additionally, proptosis results in a high frequency of conjunctivitis and keratitis. The ears may be low set with otitis media and conductive hearing loss resulting from ETD is present in most cases. Congenital fixation of stapes footplate may also be encountered in Apert syndrome. Ventilation tube placement and stapedectomy may be performed when indicated. Fronto-orbital advancement surgery may be required to allow for growth of the brain and expansion of the cranial vault. Orthodontic attention may also be required for abnormalities of the maxillary teeth and crossbite. Cervical spine anomalies are more common in Crouzon syndrome, but may also be present in Apert syndrome.

Pierre Robin Sequence

The triad of cleft palate, micrognathia, and airway obstruction was first described by St. Hilaire in 1822, later by Fairbairn in 1846, and by Shukowsky in 1911. Pierre Robin, a French stomatologist, first reported the association of micrognathia with glossoptosis in 1923. He later included cleft palate as part of this sequence. The prevalence of Pierre Robin sequence is reported to be 1 in 8,500 newborns. Children with this disorder are classified into two major categories: nonsyndromic and syndromic. Approximately 80% of cases are nonsyndromic and have the potential for normal patterns of growth and development if airway and feeding concerns are addressed early in infancy. Syndromic cases do not have as good a prognosis for growth and development in spite of early intervention. Velocardiofacial syndrome, Treacher Collins syndrome, and fetal alcohol syndrome are three of many conditions that have an association with Pierre Robin sequence.

The initiating factor in Pierre Robin sequence appears to be mandibular deficiency during fetal development. The hypoplastic and retruded mandible maintains the tongue high in the nasopharynx early in development. The high position of the tongue prevents the medial growth and fusion of the lateral palatal structures which is usually complete at 11 weeks of fetal life. Further into development, the tongue descends into a more normal position; however, at this point the palatal shelves are unable to join. This results in a U-shaped palatal cleft.

Airway obstruction is a major concern in Pierre Robin sequence. The posterior displacement of the tongue and floor of mouth due to retrognathia results in upper airway obstruction. However, airway obstruction in this disorder has anatomic and neuromuscular components. Impairment of the genioglossus and other parapharyngeal musculature are observed and predispose the airway to collapse.

The management of airway obstruction may be achieved by several methods. Prone positioning displaces the tongue anteriorly, and previously was thought to be definitive treatment for glossoptosis. Due to the inability to observe chest retractions, this method has been replaced with the use of mandibular traction devices. The placement of a nasopharyngeal airway may be the most appropriate initial method of managing the airway in infants with Pierre Robin sequence. A tube with an internal diameter of 3.0mm or 3.5mm is used and advanced 8cm or until appropriate ventilation is achieved. This provides some time to prepare for more definitive treatment. Gavage feeds via a nasogastric tube is usually recommended. In addition, tongue-lip adhesion has also been effective in the initial management of some cases. The mucosal surface of the tongue, along the floor of the mouth, over the alveolus, and onto the lower lip is denuded. The tongue is then sutured into a more anterior position. A tongue release is performed at the time of cleft palate repair. Speech is not affected with this method. In some more severe cases, tracheotomy is required when less invasive temporizing methods are unsuccessful. Mandibular distraction osteogenesis provides a definitive means to correct the bony and soft tissue involved in micrognathia. In some cases, patients exhibit catch-up growth and achieve maxillo-mandibular equilibrium without the need for mandibular corrective surgery.

Otologic concerns in Pierre Robin sequence are primarily due to conductive hearing loss secondary to chronic otitis media with effusion. Approximately 80% of patients have bilateral conductive hearing loss. Patients with abnormalities of the palate generally have anomalous

anchorage of the muscles associated with the eustachian tube (tensor veli palatine and levator veli palatini). Placement of ventilation tubes is usually sufficient for management of eustachian tube dysfunction and middle ear effusion.

CHARGE Association

The acronym of C.H.A.R.G.E. was first described by Pagon et al in 1981 to identify a nonrandom collection of malformations. The true incidence of this association is not known. The acronym stands for colobomas, heart abnormalities, atresia choanae, retardation of growth or mental development, genitourinary anomalies, and ear anomalies. In addition to these abnormalities, the head and neck anomalies manifested in this association include facial nerve palsy, pharyngoesophageal dysmotility, laryngomalacia, vocal cord paralysis, obstructive sleep, and gastroesophageal reflux. Anomalies of the temporal bones include hypoplasia of the semicircular canals and Mondini malformation.

The most urgent otolaryngologic concern in a child with this CHARGE is respiratory distress due to bilateral choanal atresia. Choanal atresia should always be included in the differential diagnosis of a newborn child with respiratory distress and cyanosis at rest, with improvement when crying. Diagnosis of choanal atresia may be established by simple auscultation of each nostril with the bell of a stethoscope, use of a mirror to observe fogging under the nostrils, passage of a 6 French nasogastric feeding tube, and direct visualization with flexible laryngoscopy. A CT is a useful radiologic study used preoperatively to determine the abnormal bony structures involved in choanal atresia. Unilateral choanal atresia usually may be managed without any interventions. Management is initially the placement of an oral airway and feeding with a McGovern nipple may be helpful in symptomatic unilateral and bilateral choanal atresia. Surgical treatment of unilateral choanal atresia may be delayed until school-aged. In bilateral cases, surgical intervention is needed earlier in infancy to prevent respiratory decompensation. A tracheotomy is usually performed prior to definitive surgical repair. There are different techniques including transnasal and transpalatal approaches, the use of a laser, the use of stents, and use of Mitomycin-C topically (0.3mg/cc for 2 minutes).

Otologic abnormalities

Otologic abnormalities include anomalies of the external, middle, and inner ear. Hearing loss is usually of a mixed type with a characteristic wedge-shaped audiogram. The intervention for chronic otitis media with effusion and Eustachian tube dysfunction is usually the placement of ventilation tubes. The use of amplification devices is also useful.

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