Original Research Article

Congenital anomalies in childhood encountered during clinical practice by an otorhinolaryngologist

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ABSTRACT

Background: The otorhinolaryngological developmental anomalies are predisposed by a number of factors like genetic factors, intrauterine infection and maternal nutritional deficiencies. The otorhinolaryngological anomalies may be associated with other systemic maldevelopments.

Methods: A total of 134 cases of congenital otorhinolaryngological childhood anomalies, fulfilling the inclusion criteria, who presented to the out-patient department were examined for a period of one year from January 2019, and were included in the study. They were thoroughly evaluated by undertaking a detailed history and clinical examination. Whenever required, additional investigations were performed. After carrying out the necessary investigations, the cases were either managed surgically or conservatively. Data was evaluated using proper statistical tools.

Results: Out of the 134 cases, 64.9% were male and 35.1% were female. In our study, 116 cases presented in the first decade of life and 18 cases presented in the second decade of life, up to 18 years of age. The most common childhood congenital anomaly was congenital deafness, which accounted for 44% of all cases.

Conclusions: Encountering cases of congenital anomalies in childhood is quite common during practice of otorhinolaryngology. Parents of the suspected children should be counselled properly to investigate other systemic anomalies. Medical and surgical management of the condition should be undertaken hand in hand with adequate counselling.

Keywords: Congenital childhood anomalies, Otorhinolaryngology, Branchial apparatus

INTRODUCTION

Children are a gift of God. Every child is a different kind of flower that altogether makes this world a beautiful garden. All children are special, but children with congenital anomalies are extra-special, as they need special medical and surgical modalities for the management of their condition. Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies that occur during intrauterine life. It can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects. It is estimated that approximately 6% of babies worldwide have an underlying congenital abnormality and is one of the main causes of global burden of disease. The complex developmental pattern of the branchial arches is responsible for the wide range of congenital anomalies encountered during practice of otorhinolaryngology. Otorhinolaryngological malformations may often be associated with gastrointestinal, nephrological and cardiovascular malformations. A number of etiological factors are involved in the complex development of the branchial apparatus and in the
formation of cochlea. Some of these factors are - genetic factors, viral infections (maternal intra-uterine infections), maternal nutritional deficiencies etc. Whenever we encounter any case of congenital anomaly, we should undertake a detailed examination of the other organ systems.

Background knowledge of the various factors involved in the development of the branchial apparatus and the cochlea indirectly acts as a tool to understanding the various congenital anomalies in otorhinolaryngology. Therefore, properly evaluated statistical data regarding the occurrence and presentations of congenital otorhinolaryngological anomalies is needed to help us in determining the type of intervention needed for these cases. This study was carried out to study the occurrence and the clinical manifestations of various congenital anomalies in childhood encountered during the course of clinical practice by an otorhinolaryngologist, to study the association between otorhinolaryngologic anomalies and other systemic involvement, and to study the available devices and surgical modalities of treatment in the management of congenital anomalies in otorhinolaryngologic practice.

METHODS

After approval by the institutional Board of Ethics, this clinical study was carried out under the aegis of the department of Otorhinolaryngology, at a tertiary care hospital-cum-referral centre in North-East India. A total of 134 cases of congenital otorhinolaryngological childhood anomalies presenting to the out-patient department were examined for a period of one year from January 2019, and were included in the study.

Inclusion criteria

Patients of both sexes between ages 0 to 18 years were included.

Exclusion criteria

Patients coming with cleft lip and palate were not included in the study as they are usually examined by the department of oral and maxillo-facial surgery.

Patients fulfilling the inclusion criteria were thoroughly evaluated by undertaking a detailed history and clinical examination. Whenever required, additional investigations were performed. After carrying out the necessary investigations, the cases were either managed surgically or conservatively. Data was evaluated using proper statistical tools.

RESULTS

Out of the 134 cases, 64.9% were male and 35.1% were female. In our study, 106 cases presented in the first decade of life and 28 cases presented in the second decade of life, up to 18 years of age (Table 1 and 2).

Table 1: Distribution of different cases.

| Anomaly                  | Number of cases (%) | Male cases | Female cases |
|--------------------------|---------------------|------------|--------------|
| Congenital deafness      | 59 (44.0)           | 38         | 21           |
| Pinna anomalies          | 22 (16.4)           | 14         | 8            |
| Pre-auricular sinus      | 18 (13.4)           | 13         | 5            |
| Tongue tie               | 10 (7.5)            | 7          | 3            |
| Thyroglossal duct cyst   | 7 (5.2)             | 5          | 2            |
| Cystic hygroma of neck   | 3 (2.25)            | 3          | 0            |
| Ectopic thyroid          | 1 (0.75)            | 1          | 0            |
| Choanal atresia          | 4 (3.0)             | 2          | 2            |
| External nasal deformity | 2 (1.5)             | 2          | 0            |
| Laryngomalacia           | 3 (2.25)            | 0          | 3            |
| Lymphangioma of neck     | 1 (0.75)            | 0          | 1            |
| Hemangioma of neck       | 1 (0.75)            | 0          | 1            |
| Binder’s syndrome        | 1 (0.75)            | 1          | 0            |
| Goldenhar syndrome       | 1 (0.75)            | 1          | 0            |
| Waardenburg syndrome type I | 1 (0.75)       | 0          | 1            |
| Total                    | 134                 | 87 (64.9)  | 47 (35.1)    |

Table 2: Patient demographics.

| Age group (in years) | Number of patients | Percentage (%) |
|----------------------|--------------------|----------------|
| 0-6                  | 72                 | 53.8           |
| 6-12                 | 44                 | 32.8           |
| 12-18                | 18                 | 13.4           |

Congenital deafness

It was the commonest anomaly detected. Congenital anomalies related to the inner ear presenting as congenital sensorineural hearing loss were seen in 59 (44.0%) cases, which account for nearly half of all the congenital anomalies encountered during clinical practice by an otorhinolaryngologist. Out of those, 38 were male and 21 were female, with a male to female ratio of 1.8:1. In our study, average age of diagnosis is 2.65 years. All were affected bilaterally. All the cases had an otherwise normal otorhinolaryngological examination. Otoacoustic emission and Brainstem evoked response audiometry testing revealed severe sensorineural hearing loss. All the cases had associated delayed speech development.
Causative factors for this might be associated neonatal hyperbilirubinemia in 23 cases and perinatal asphyxia in 19 cases. Two of the cases were diagnosed as auditory neuropathy, 1 was a case of vanishing white matter disease, which was identified on magnetic resonance imaging and 1 suffered from hypoplasia of the Facial and the vestibulocochlear nerves, which was identified on the patient’s computerized tomographic picture and magnetic resonance image (Figure 1). Cochlear implantation was carried out in pre-lingual patients i.e., 26 patients, and the rest of the patients are on hearing aid with regular follow-up at the Department of ENT, GMCH.

**Figure 1:** Magnetic resonance image of a 4-year old patient with absent cochlea on the right, but present on the left (red arrow).

**Pinna anomalies**

External ear abnormalities were noted in 22 patients (16.4%), with a male to female ratio of 1.75:1. Of these 22, 3 (13.6%) were associated with hearing loss. It was the second most commonly detected anomaly. Microtia was the most common pinna anomaly and accounted for 14 cases (Figure 2). Macrotia was found in 3 cases. Two cases of lop ear were assessed. 1 case of accessory tragus, 1 case of a bifid tragus and 1 case of pre-auricular tags was noted (Figure 3). These cases were referred to the Department of Plastic Surgery, GMCH for reconstruction.

**Figure 2:** (A) A 7-month female patient with macrotia and (B) a 14-year female patient with microtia.

**Pre-auricular sinus**

A total of 18 cases of pre-auricular sinus were encountered and all 18 cases were operated upon at our department and the post-operative period was uneventful (Figure 4).

**Ankyloglossia/tongue tie**

A total of 10 (7.5%) cases of ankyloglossia were recorded in the study, of which 7 were male and 3 were female. In the study, 5 cases of grade I, 3 case of grade II and 2 cases of grade III ankyloglossia were encountered (Figure 5). Necessary investigations were carried out and all the cases underwent release of tongue tie with repair using absorbable polythread suture material.

**Figure 3:** (A and B) A 6-year old male patient with bilateral lop ears; (C) A 4-month old male patient with bifid tragus.

**Figure 4:** Two cases of pre-auricular sinus: (A) a 6-year old male patient with an uninfected pre-auricular sinus and (B) that on the right shows a 7-year old female with infected pre-auricular sinus.

**Cystic swellings in the neck**

In the neck region, 7 cases of thyroglossal duct cyst, 3 cases of cystic hygroma (Figure 6) and 1 case each of ectopic thyroid, lymphangioma of neck and hemangioma of neck were reported. This amounted to 9.7% of total anomalies encountered during the course of this study. Anomalies related to thyroglossal duct were encountered in seven patients. All the 7 cases were managed surgically by Sistrunk operation. Three cases of cystic hygroma of neck were reported and all were surgically excised out.
Figure 5: Two cases of ankyloglossia: (A) a 5-year old male patient with grade I ankyloglossia and a 14-month old male patient with grade III ankyloglossia.

Figure 6: A 4.5 year old male with cystic hygroma of neck.

Choanal atresia

Choanal atresia can be perfectly described as “Birth without breath”. In our study, two patients with choanal atresia were encountered, with a male to female ratio of 1:1. Both the patients presented with unilateral choanal atresia and were managed accordingly.

Laryngomalacia

We examined and assessed three cases of noisy breathing, and in all the cases, the classical omega shaped epiglottis was examined by 70° endoscope. The noisy breathing sounds were reduced with prone position. In our study, age of presentation was between 1st to 3rd months of age. The parents of the patients were adequately counselled and reassured.

Goldenhar syndrome

Goldenhar syndrome is condition characterized by anomalies of the derivatives of the first and second branchial arches, vertebral defects and ocular abnormalities. It is also referred to as oculo-auriculo-vertebral syndrome (OAVS), hemifacial microsomia, or first or second brachial arch syndrome. In this study, we report one case of Goldenhar syndrome who is currently undergoing treatment for his ocular abnormalities at the Regional Institute of Ophthalmology (Figure 7).

Binder's syndrome (maxillonasal dysplasia)

Binder's syndrome or maxillonasal dysplasia is a rare clinical entity, but the exact birth prevalence remains unknown. It is important to note that Binder's syndrome has a variable presentation, and various treatment options are available for management of such cases. In this study, we encountered one case of Binder’s syndrome which was managed conservatively.

Figure 7: A 3-year old male with Goldenhar syndrome.

Waardenburg syndrome type I

A patient of Waardenburg syndrome type I with the presence of bilateral severe hearing loss, brilliant blue iris, synophrys, wide-set eyes with broad nasal root and dystopia canthorum was noted (Figure 8).

Figure 8: Clinical photograph of an 18-month old female patient with brilliant blue iris, synophrys, wide-set eyes with broad nasal root and dystopia canthorum.

DISCUSSION

In our study, maximum patients with congenital malformations were male, and 53.8% presented in the first 6 years of life. This is similar to the findings of Sinha et al, where they tabulated 100 patients of otorhinolaryngological developmental anomalies. According to that study 64% were male and 36% were...
female and maximum patients presented in the first decade of life.³

In our study, congenital hearing loss accounted for 44.0% of total cases. The way human beings perceive the world is mediated via sensory experiences. Out of all the senses, it is the sense of hearing which intrinsically facilitates communication and fosters social interaction, allowing people to construct relationships, participate in daily activities, be alerted to danger, and experience life events. Around 360 million people, 5% of the world’s population live with hearing loss which is considered disabling; of these, nearly 32 million are children.² The vast majority live in the world’s low-income and middle-income countries. For children hearing is the key to learning spoken language, performing academically, and thus interacting with people socially. Hearing loss poses as a giant barrier to education and social integration. While the most obvious repercussion of childhood hearing loss is on language acquisition, the condition also has consequences for overall literacy, the development of social skills and attitudes, including self-esteem. Untreated hearing loss is oftentimes associated with academic underachievement which can lead to lower job performance and fewer employment opportunities later in life. For a child, difficulties in communication may result in feelings of anger, stress, loneliness and emotional or psychological consequences which may have a profound effect on the family as a whole. In low-resource settings in which a child would already be at higher risk of injury, hearing loss can place a child in dangerous and difficult situations due to decreased alertness. In a broader context, hearing loss, if left untreated, affects the social and economic development of communities and countries. The children with hearing loss can benefit greatly from being identified early in life and offered appropriate interventions. Some of the factors causing congenital hearing loss are genetic (e.g. children born of consanguineous marriage), conditions at the time of birth (e.g. prematurity, birth asphyxia, neonatal jaundice) and maternal infections during pregnancy (e.g. rubella, cytomegalovirus infection). The World Health Organization (WHO) estimates that around 60% of childhood hearing loss could be avoided through prevention measures.² When unavoidable, interventions like hearing aids and cochlear implants are needed to ensure that children reach their full potential through education, rehabilitation and empowerment.⁴

In the ENT region, 50% of the malformations affect the ear.⁵ In our study, malformations of the pinna accounted for 16.4% cases. Pre-auricular sinus amounted to 13.4% of the total cases, with mean age of presentation 3.7 years, which was similar to the study carried out by Tang et al.⁶ Malformations of the outer and middle ear are predominantly unilateral (70-90%) and mostly involve the right ear. Inner ear malformations can be unilateral or bilateral.⁵ The incidence of ear malformations is approximately 1 in 3800 newborns.⁷ The incidence of outer ear malformations has been reported at 1:6000 newborns to 1:6830 newborns.⁸,⁹ The prevalence of microtia is higher, i.e 3:10,000 according to Schloss.¹⁰ Severe malformations can be expected in 1:10,000 to 1:20,000 newborns, gross malformation or aplasia in 1:17,500 newborns.⁵ Often there are middle ear problems associated with outer ear disorders. Usually the inner ear remains unaffected as it has a separate development in the uterus and forms early in utero (1⁴ trimester). Ear malformations may be genetic - associated with syndromes (30% of all cases), or acquired in nature. Tewfik et al in 1997 published a detailed list of syndromes and conditions associated with congenital ear malformations.¹¹ The acquired ear malformations originate from exogenic injury during pregnancy. The noxae comprise of mostly viral infections like cytomegalovirus, rubella and herpes simplex virus; chemical agents, malnutrition (maternal diet low in carbohydrate and folic acid), Rh-incompatibility, hypoxia, irradiation, atmospheric pressure changes and noise exposure.¹ Bleeding occurring in the first part of pregnancy and disturbances of metabolism such as diabetes are some additional factors that increase risk of giving birth to a child with ear malformations.¹² Among the chemical teratogens, medicinal drugs play a principle role, a prominent example being thalidomide, which led to a considerably increased frequency of malformations in the early 1960s. Quinine and aminoglycoside antibiotics can also give rise to congenital malformations. Cytostatics and medications used in the treatment of epilepsy (e.g. trimethadione, diphenylhydantoin and valproic acid) may also be responsible. Even excessively high doses of retinoic acid (retinoic acid embryopathy) and vitamin A deficiency (VAD syndrome) during pregnancy can produce ear malformations.¹² Environmental agents such as mercury-containing fungicides, herbicides and lead can exert teratogenic effects. Deficiency of certain hormones (e.g. thyroid hormone) can be associated with ear malformations (e.g. dysfunction of thyroid hormone causes sensorineural hearing disorder and inner ear malformations in Pendred syndrome). Many congenital ear malformations can be easily corrected if they are diagnosed very early, i.e within a few weeks of birth. Diagnosis is usually based on a thorough physical examination and associated audiological tests to detect any hearing loss. Diagnostic imaging such as computerised tomographs are usually done to see the parts of the ear as well as to look for related facial or jaw deformities. Children who have minor ear malformations may not need treatment if their hearing is normal. For more severe cases, the treatment goals focus on correcting the shape of the ear and restoring any hearing loss.

Treatment options include ear molding for newborns with protruding, constricted, or Stahl’s ears, ideally at 1 to 3 weeks of age; surgery (usually after 6 years of age, when the ear has grown to almost adult size) to correct problems with the outer ear or earlobes; hearing aids or cochlear implants to improve hearing; counselling and reassurance plays an important part, as external ear
deformities are gross anomalies and often the patients are subjected to teasing, and ostracized by their friends and family. For a child, this may lead to anger, loneliness and emotional and psychological instability.

In our study, 7.5% of cases presented with ankyloglossia. Ankyloglossia (more commonly called “tongue-tie”) is a congenital anomaly characterized by an abnormally short lingual frenulum, which may restrict tongue tip mobility. Its incidence varies from 0.02% to 5%, depending on the study, its definition of ankyloglossia, and population examined. The incidence among outpatients of a children’s hospital with breast-feeding problems was almost 3 times higher (13%). Two independent studies of oral anomalies in neonates found a significant 3X predilection for ankyloglossia in males. It may occur with increased frequency in various congenital syndromes, including oro-facio-digital syndrome, Beckwith-Wiedemann syndrome, Opitz syndrome, Simpson-Golabi-Behmel syndrome, and X-linked cleft palate. Although there is a lack of scientific evidence proving a true relationship between speech disorders and ankyloglossia, there seems to be a consensus that it may be the cause of specific speech disorders in certain individuals. Ankyloglossia may interfere with articulation, but it does not prevent or delay the onset of speech. Several treatment methods for ankyloglossia have been suggested and the approach depends on the grade of ankyloglossia. The normal range of free tongue movement is more than or equal to 16 mm. Range of tongue movement between 12-16 mm is mild ankyloglossia, between 8-11 mm is moderate, between 3-7 is severe and below 3 mm is complete ankyloglossia. Management approaches range from very early treatment without anesthesia to the other extreme—that ankyloglossia should never be treated. In our study, the patients underwent tongue tie release and repair using polythread sutures. Doctors oftentimes delay recommending treatment of a short lingual attachment, unless there are obvious speech or nursing difficulties. Treatment options such as observation, speech therapy, frenotomy under local anesthesia, and frenectomy under GA have all been propounded in the literature.

In our study, congenital neck masses accounted for 9.7% of total cases. Common congenital developmental masses in the neck include thyroglossal duct cysts, branchial cleft cysts, dermoid cysts, cystic hygromas, vascular malformations, and hemangiomas. A neck mass present since birth or discovered during the neonatal period is commonly benign and developmental. Vascular malformations such as lymphangiomas present at birth and keep growing as the child gets older. On the other hand, hemangiomas develop a few weeks after birth and have a rapid growth phase. Developmental masses may present later in life, either with superimposed infection or with growth over time. The location of the neck mass provides many clues to the diagnosis. The most common cystic midline neck masses are thyroglossal duct cysts and dermoid cysts. Thyroglossal duct cysts are often located over the hyoid bone and elevate with tongue protrusion or swallowing, whereas dermoid cysts typically move with the overlying skin.

Ultrasoundography is the preferred imaging investigation for a developmental or palpable neck mass. Excision is the recommended treatment of choice for congenital neck masses. Sistrunk operation is carried out for removal of a thyroglossal duct cyst, which was performed in all 7 of our cases.

In our study, 3.0% of patients presented with choanal atresia. Choanal atresia, defined as the anatomical closure of the posterior choanae in the nasal cavity, is a relatively uncommon disease entity with an estimated incidence of 1:5000-7000 birth. Historically reported in 1910, the deformity was made up of 90% bony and 10% membranous atresia. Anatomical boundaries of the posterior choanae include the undersurface of the body of the sphenoid bones superiorly, the horizontal portion of the palatal bone inferiorly, the medial pterygoid lamina laterally, and the vomer medially. The actual narrowing could be caused by one of the above-mentioned bony components. The key to surgical success is correctly identifying and addressing the point of obstruction. Interestingly, the characteristics of choanal atresia follow a “211” rule, namely the ratio of unilateral to bilateral choanal atresia, female to male, and the right sided to the left sided choanal atresia. Initial clinical evaluation includes introduction of a six or eight Fr suction catheter via the nostrils, methylene blue dye test, cotton wisp test, and laryngeal mirror test. Flexible nasal endoscopy (gold standard test) in a patient with proper preparation, such as nasal decongestion and mucous suctioning, allows direct visualization of the point of obstruction in the nasal passage and helps confirm the presence of an atretic plate in the choana. Definitive diagnosis of choanal atresia is made by computed tomography scans. CT scans help in delineating the nature and severity of choanal atresia, besides being useful in differentiating other causes of nasal obstruction from choanal atresia. Management of choanal atresia includes initial airway management (in cases of bilateral choanal atresia) by means of feeding tubes or tracheostomy; and surgical management. Surgical modalities of treatment include transnasal puncture, transpalatal repair, transnasal endoscopic repair (currently in prevalence) and laser-assisted surgery.

In our study, 2.25% of cases were found to have laryngomalacia. Laryngomalacia is the collapse of supraglottic structures during inspiration. It is the commonest laryngeal disease of infancy. Laryngomalacia presents as stridor - a high-pitched, musical, vibrating, multiphase inspiratory noise - within the first 10 days of life. Signs of severity are present in 10% of cases. Poor weight gain (probably the most contributive element), dyspnoea with permanent and severe intercostal or xiphoid retraction, episodes of respiratory distress, obstructive sleep apnoea, and/or episodes of suffocation while feeding or feeding difficulties are associated findings in cases of laryngomalacia. The diagnosis is
based on systematic office flexible laryngoscopy to confirm laryngomalacia and to exclude other causes of supraglottic obstruction. Rigid endoscopy under general anaesthesia is only performed in cases where there is absence of laryngomalacia on flexible laryngoscopy, presence of laryngomalacia with signs of severity, to search for any associated lesions prior to surgery, to check for discrepancy between the severity of symptoms and the appearance on flexible laryngoscopy, and/or atypical symptoms (mostly aspirations). The diagnostic work-up is adjusted to suit each child’s symptomatology; however, guidelines recommend objective respiratory investigations in infants presenting with signs of severity. Laryngomalacia is commonly well tolerated and has a favourable disease course in most cases. In 10% of cases, laryngomalacia is poorly tolerated with signs of severity. An assessment is then performed to plan and guide the treatment course, which is usually surgical.

In our study, we encountered one case each of Binder’s syndrome, Goldenhar syndrome and Waardenburg syndrome, which are relatively uncommon. Management of Goldenhar syndrome requires a multidisciplinary approach. Binder’s syndrome has variable presentation and the treatment strategy varies depending on the facial correction needed and patients’ demands. We encountered one patient of Waardenburg syndrome type 1, who had a broad nasal root, dystopia canthorum, brilliant blue irises and sensorineural hearing loss in both ears. All these patients were accordingly managed.

CONCLUSION

An estimated 3,03,000 newborns die within the first four weeks of birth every year, worldwide, due to congenital anomalies. Encountering cases of congenital anomalies in childhood is quite common during practice of otorhinolaryngology. Congenital anomalies often contribute to long-term disability, which in turn may have significant impact on individuals, families, health-care systems, and societies. Although congenital anomalies may be the result of one or more factors (genetic, infectious, nutritional or environmental factors), it is often difficult to identify the exact causes. Some congenital anomalies are preventable. Maternal vaccination, adequate intake of folic acid or iodine through fortification of staple foods, or supplementation during ante-natal period, and adequate antenatal care are just three examples of preventive methods. Whenever the otorhinolaryngologist encounters any case of congenital anomaly, he/she should make it a point to carry out a thorough examination of the patient, including the necessary investigations. This will help in not just highlighting the primary anomaly, but will also help in uncovering associated anomalies of other organs/organ systems. Genetic counselling, special education and adequate support should be given to the family members, especially the parents, so as to allay their apprehensions and fears regarding their child’s condition, and to outline a realistic expectancy regarding the cure. This study is important as it highlights the need for communication between doctors and the patient’s guardians and also the need for inter-departmental cooperation in timely diagnosis of congenital childhood anomalies, especially multi-system anomalies, which will help lower the morbidity.

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