



Choanal Atresia with Other Uncommon Abnormality: A Rare Case Report

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Abstract

A 37 year old male patient presenting to department of ENT in a tertiary rural hospital, with right sided nasal obstruction since childhood. On clinical examination found to have Coloboma iris, ptosis and microphthalmia of right side along with a complete Choanal atresia and right lop ear. There were no other clinical abnormalities detected. Keeping possibility of CHARGE syndrome systemic workup was conducted. But no other known component of this syndrome was found.

Introduction

CHARGE association is a rare congenital disorder described by Hall [1] and Hittner [2] separately in the year 1979. In the year 1981 Pagon [3] and colleagues proposed the acronym CHARGE. Recent studies showed the incidence to be 1 in 8500 in Atlantic province of Canada [4]. The syndrome consists of C-coloboma of the eyes, H-heart disease, A-atresia of the choanae, R-retarded growth and development, G – genital hypoplasia/genitourinary anomalies and E-ear anomalies and/or hearing loss. The syndrome has also been described with many other abnormalities. Blake et al. [5] suggested diagnostic criteria where he describes 4 major and 7 minor criteria. Presence of all 4 major criteria, or 3 major and 3 minor will be sufficient for the diagnosis of CHARGE syndrome. Simultaneously, Verloes suggested an update of diagnostic criteria, emphasising the most specific embryological defects while avoiding non-specific or secondary anomalies [6]. Recently gene CHD7 on chromosome 8 (8q12.1) has been found to be the major gene involved in this syndrome [7,8]. We report a special case where ptosis was associated with microphthalmia coloboma of iris, unilateral Choanal atresia and lop ear.

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Case Presentation

A 37 year old male patient from rural background presented to our OPD with complaints of unilateral nasal obstruction since childhood which was undiagnosed to this age. He also had right sided unilateral mucopurulent nasal discharge. Clinical examination revealed deviated nasal septum to the left side with caudal dislocation to the right. Diagnostic nasal endoscopy revealed mucopurulent discharge in right nasal cavity and complete obstruction of right choana which was hard on palpation with probe. Rest of the nasal cavity was normal. Left choana was patent. He also had ptosis of right eye, microphthalmia and Coloboma of iris (Figure 1-4). On further evaluation right eye was blind with perception of light. Fundocopy was done to rule out other abnormality and it revealed the Coloboma of retina also. Patient had lop ear abnormality but hearing was normal. Echocardiogram was done and no cardiac abnormality was detected. No abnormality of genitourinary system was



Figure 1: Showing Coloboma of right iris with ptosis (upper lid retracted up to show iris) notice normal eye on the opposite side.

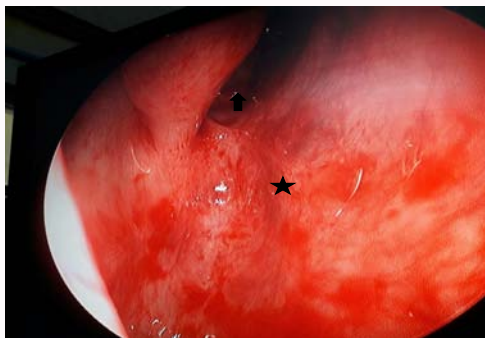


Figure 2: Right choana showing complete atresia (asterisk) and posterior end of inferior turbinate (up arrow).

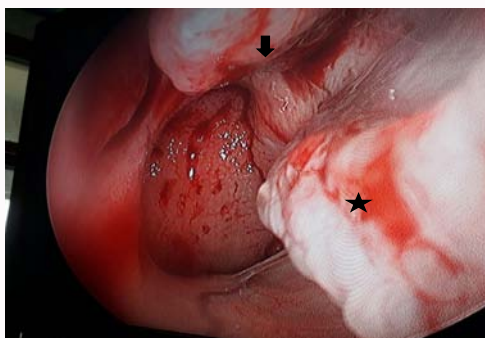


Figure 3: Left nasal choana showing normal anatomy, posterior end of inferior turbinate (asterisk) and posterior end of middle turbinate (down arrow).

found in clinical and ultrasonography imaging. Patient did not have growth retardation or mental retardation. CT scan revealed right side choanal atresia with thin bony plate (0.37 cm).

Endoscopic septoplasty with recanalization of right choana was done and the atretic plate was found to be thin except on inferomedial aspect. Adequate wide opening was secured hence no stenting was done. Now after 4 months choana is patent and patient doesn't have nasal obstruction.



Figure 4: Sagittal section showing right nasal cavity with complete bony Choanal atresia of 37mm thickness with nasal pack *in situ*.

Discussion

Choanal atresia is a rare congenital disorder with incidence of 1 in 8500 live births. It is called charge syndrome when associated with other abnormalities as mentioned earlier. Table 1 shows the diagnostic criteria suggested by Blake [5] and Verloes [6]. Other than the listed abnormalities many other associations are being reported including hand and limb defects. To the best of our knowledge there are no cases of charge syndrome associated with ptosis. Ours was an interesting case as patient was an adult with choanal atresia, unilateral visual loss, ptosis, microphthalmia, external ear abnormality (lop ear) without hearing loss. There was no abnormality found.

CHD7 gene is found in many of subjects with the CHARGE syndrome. It is now being investigated for correlation between the spectrum of mutation in this gene and its phenotypical manifestation. In many of the studies conducted ear anomalies and deafness is the constant finding. Heart defects, Coloboma, cranial nerve abnormality and Choanal atresia occur in lesser frequency. The spectrum of eye abnormality usually limited to the presence of iris coloboma. Finding of microphthalmia, posterior segment Coloboma retinal detachment as a result of it is also documented. Presence of ptosis is a rare finding

Table 1: Diagnostic criteria for CHARGE syndrome.

Verloes [6] (typical CHARGE = 3 major criteria or 2 major and 2 minor criteria)	Blake et al [5] (all 4 major criteria, or 3 major and 3 minor criteria)
Major criteria	Major criteria
Coloboma (iris, choroid ± microphthalmia)	Coloboma (iris, retina, choroid, optic disc, or microphthalmia)
Atresia of choanae	Atresia of choanae
Hypoplastic semicircular canals	Cranial nerve dysfunction - I: anosmia, VII: facial palsy, VIII: sensorineural deafness and vestibular problems, IX and/or X: swallowing problems Characteristic external ears (absent or hypoplastic lobes, asymmetry, decreased cartilaginous folds, and triangular concha) and inner ear anomalies (temporal bone findings with cochlear hypoplasia and or absent/hypoplastic semicircular canals)
Minor criteria	Minor criteria
Rhombencephalic dysfunction (brainstem dysfunctions, cranial nerve VII to XII palsies, and neurosensory deafness)	Characteristic facial features - broad, sloping forehead, laterally protruding ears, small mouth, and high nasal bridge
Malformation of mediastinal organs (heart, oesophagus)	Congenital cardiovascular malformations of all types
Hypothalamo-hypophyseal dysfunction (including GH and gonadotrophin deficiencies)	Tracheo-oesophageal fistula
Abnormal middle or external ear	Growth deficiency
Mental retardation	Genital hypoplasia - micropenis and/or cryptorchidism or hypoplastic labia. Delayed, incomplete pubertal development
	Orofacial cleft
	Developmental delay: delayed motor milestones, hypotonia, mental retardation

and our case is one such occurrence.

The ear abnormality is present in almost all of many series and cup shaped and lop ears are mildest and commonest. Rarely there can be hearing loss alone or combined with the external ear abnormality. Recently lateral semi-circular canal abnormality and severe SNHL needing cochlear implants also has been found to be a common in cases with CHD7 mutation.

Learning disability and speech abnormality is common. And growth retardation with stunting is also seen. It is known to improve mid childhood. Hypogonadism and micropenis undescended testis and absent testis also has been found. Genital involvement is most commonly seen in males and girls do not have many of these. Hypoplasia of uterus can be found. Hormone supplement in right time can correct the abnormality. Horseshoe kidney, agenesis of the kidney, renal cyst can be seen in CHARGE syndrome. Choanal atresia and cleft palate and lips are common associations. Tracheo-oesophageal fistula, oesophageal atresia, diaphragmatic hernia and anal atresia have been reported. A minority of patients have central nervous system abnormalities, including corpus callosum agenesis combined with cerebellar hypoplasia, hydrocephaly, and atrophy of the cerebral cortex. Facial nerve palsy was present in minority of patients in many studies. The spectrum of cardiac abnormalities can range from an atrial septal defect to tetralogy of fallot and great vessel abnormalities like coarctaion. The detection of the condition might become difficult when the spectrum of anomalies is narrow and the anomaly not involving major structures like eyes, heart, kidney or cranial nerves. The detection of Choanal atresia might be difficult in such a patient who hails from a rural place. The mild symptoms of unilateral complete atresia with contralateral normal choana, ignorance of the parents on child's symptom and the inaccessibility to speciality care made the diagnosis late.

Conclusion

Finding of choanal atresia should always arise suspicion of associated systemic congenital anomalies and a thorough

examination should be conducted. The genetic workup should be considered to find out genetic basis. The present knowledge of CHD7 gene mutations and its phenotypical expressions should be expanded. However early detection of heart and renal anomaly can definitely improve quality of life in such people. Intrauterine high suspicion should arise in all cases of polyhydramnios. Focused detection of choanal and esophageal atresia should be attempted. The test for CHD7 gene will be expected to be included in all such cases once the association is proven beyond doubt.

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