Abstract: CHARGE syndrome is a rare, recently well recognized entity with non-random pattern of congenital anomalies. The syndrome associations consist of C-coloboma of the eyes, H-heart disease, A-atresia of the choanae, R-retarded growth and development, G - genitourinary anomalies and E-ear anomalies and hearing loss. All anomalies are not seen in every case and a varied spectrum of associations is seen in most of the cases. The exact incidence is not known. However, the reported prevalence is approximately 110,000 births. We report one such case.

Keyword: CHARGE SYNDROME, CHOANAL ATRESIA, COLOBOMA, CONGENITAL DEAFNESS

INTRODUCTION:
The CHARGE association was first described in 1979 by Hall et al., in 17 children with multiple congenital anomalies who were ascertained by choanal atresia [1,2]. In the same year, Hittner reported this syndrome in 10 children with ocular colobomas and multiple congenital anomalies [3], hence the syndrome is also called Hall-Hittner syndrome [4]. Pagon et al., in 1981 first coined the acronym CHARGE association [5], (Coloboma, Heart defect, Atresia choanae, Retarded growth and development, Genital hypoplasia, Ear anomalies/deafness) as a non-random association of anomalies occurring together more frequently than one would expect on the basis of chance. The original diagnostic criteria required the presence of four out of six of the CHARGE characteristics. Over the past 15 years the specificity of this pattern of malformations has reached the level that many clinicians now consider it to be a recognizable CHARGE syndrome. [2]

CASE REPORT:
A female child aged 14, was brought to the OPD by her grand mother, with the complaints of Discharge from left side of nose since birth. There was history of hard of hearing both ears, Difficult in speech, Defective vision on left eye. Otherwise, the perinatal and developmental history was normal. On examination, pt had unilateral choanal atresia, sensorineural hearing loss on both side. Eye examination revealed Left eye micro cornea with typical coloboma choroid involving optic disc and iris, squint, spontaneous nystagmus, loss of left eye vision. General examination of the patient showed facial asymmetry, polydactyly, poor breast development on left side, systemic examination revealed, wide fixed split on auscultation, abdominal and respiratory systems are normal.

FIG 1 - SHOWS FACIAL ASYMMETRY, MICRO CORNEA, SQUINT

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DISCUSSION

CLINICAL DESCRIPTION:

FIG 3 - SHOWS LEFT EYE COLOBOMA

COLOBOMA:
This feature may be unilateral or bilateral and may affect only the iris or extend to involve the retina, or only the retina. Vision may be normal or impaired. The eye abnormalities range from iris coloboma without visual impairment to microphthalmos and anophthalmos. Retinal coloboma is more prevalent than iris coloboma and can affect the optic nerve. Eye malformations have been reported in as many as 80% of patients with CHARGE syndrome, with retinal involvement being the most common [6]. External inspection is not sufficient and testing for functional vision is important but challenging especially in CHARGE individuals with extensive bilateral chorioretinal coloboma involving the optic nerve [7].

HEART DEFECT:
Congenital heart defects occur in 75–80% of patients with CHARGE syndrome. The most common major heart defect is tetralogy of Fallot (33%). Other frequent anomalies are patent ductus arteriosus, double outlet right ventricle with atrophicventricular canal, ventricular septal defect and atrial septal defect with or without cleft mitral valve. Vascular rings and more complex heart defects need to be anticipated [8-11].

FIG 4 - CT PNS SHOWING LEFT SIDE CHOANAL ATRESIA

CHOANAL ATRESIA:
Choanal atresia is a narrowing or a blockage of the passages between the nasal cavity and the naso-pharynx. It represents a primary feature with a high index of suspicion for CHARGE syndrome and it should focus attention on other organ systems such as the eye and heart. Choanal atresia may be membranous or bony; bilateral or unilateral. Bilateral posterior choanal atresia (BPCA) was shown to be associated with increased neonatal mortality, especially if associated with major cardiac malformations and/or tracheoesophageal atresia [8]. However, the Canadian epidemiological study data suggests that an individual from this population with a more severe clinical presentation of CHARGE features generally survive [9]. Polyhydramnios in pregnancy is seen commonly in individuals with bilateral posterior choanal atresia, and may also be present without BPCA, probably due to an insufficient swallowing mechanism. Chronic middle ear infections and deafness can be associated[12].

RETARDATION OF GROWTH AND DEVELOPMENT:
Growth and developmental retardation become more obvious as the child matures. At birth, children with CHARGE syndrome usually have normal weights and lengths [13]. When growth deceleration is due to cardiac and respiratory problems, there may be catch up growth, and normal height can be obtained [14]. However, the influence of feeding problems on growth in infancy should not be underestimated. Early and continued intervention for feeding difficulties is vitally important [15]; occasionally there is growth hormone deficiency. The majority of school-aged children with CHARGE syndrome are below the third percentile for physical growth norms [13]; feeding with solids and lumpy foods, and risk of aspiration may still exist. Mental retardation is variable with intelligence quotients (IQ) ranging from near-normal to profound retardation. Behavioral issues and an autism-like spectrum disorder are now being recognized as features of the syndrome [16,17].

GENITOURINARY PROBLEMS:
Under-development of the external genitalia is a common finding in males but it is more difficult to recognize in females. Microphallus, penile agenesis, hypospadias, chordee, cryptorchidism, bifid scrotum, atresia of uterus, cervix and vagina, hypoplastic labia and clitoris are reported genital anomalies in this syndrome. Reported renal anomalies include solitary kidney, hydronephrosis, renal hypoplasia, duplex kidneys and vesicoureteral reflux. Hypogonadotropic hypogonadism has been reported and is associated with delays in puberty or pubertal arrest [18,19].

FIG 5 - PTA SHOWS B/L SENSORINEURALHEARING LOSS turns to the cochlea (Mondini defect), and, in particular, absent semicircular canals. These distinctive radiological findings are classical for CHARGE syndrome and can aid diagnosis in a suspected case [21]. For this reason, a neonatal CT scan to look at the choanae and temporal bones can be extremely useful.

MANAGEMENT
Children with CHARGE syndrome require intensive medical management as well as numerous surgical interventions. The most common neonatal emergencies in CHARGE syndrome involve cyanosis due to bilateral posterior Choanal atresia and/or congenital heart defects,
Photophobia is often a significant problem that can be ameliorated with tinted spectacles or by wearing a cap or visor with a dark brim. In the presence of facial palsy, patients should avoid noxious scarring by using artificial tears. Hearing aids should be used as soon as hearing loss is documented. Frequent re-molding of the earpieces is necessary as the ear canals can be initially very small and ear cartilage may be insufficient to support a hearing aid.

Children with CHARGE syndrome require aggressive Medical management of their feeding difficulties. Often needing gastrostomy and jejunostomy feeding Tubes. Gastrooesophageal fundoplication may be Required for GERD that does not respond to medical management. As intubation can be difficult in children with CHARGE syndrome, a pediatric anesthesiologist or pediatric Otolaryngologist should be present for planned surgical Procedures. Any infant suspected of having CHARGE syndrome should have a complete eye examination by an ophthalmologist, with follow-up every three to six months thereafter, depending on the eye involvement.

Cochlear implants have been successfully performed in CHARGE syndrome patients. Children with CHARGE syndrome who undergo cochlear implantation should be allowed to continue with their sign language in parallel with their expressive speech training [23]. Adapted educational and therapeutic services to deal with dual auditory and visual sensory impairment should be proposed early in the child's life [23-25]. However, this population is unique with respect to their aberrant cranial nerve pathways and problems with expressive language. In terms of endocrine issues, sex steroid therapy has been used for penile growth and descent of testes in males with CHARGE syndrome. The main use for testosterone is for delayed and incomplete male puberty during adolescence. Females often require hormone replacement at puberty [18]. Sex hormone replacement is also indicated for prevention of osteoporosis [19].

REFERENCES


