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Rare case of Goldenhar syndrome in a 3 years male child

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Goldenhar syndrome is a rare condition presenting with congenital presence of limbal dermoid with congenital. Association of pre-auricular skin tag or pre-auricular appendage or sometimes squint, there is defect in chromosome yet it is not inherited but it is also called occulo auriculo vertebral syndrome or dysplasia there is incomplete development of 1st and 2nd branchial arch which manifests as defects in head and neck involving 1 ear 2 nose 3 soft palate and mandible in very small percentage 5 to 15 percent cases one has hearing defect 2 impairment of memory 3 defects in spine and limbs and may be kidney involvement and congenital heart and dental anomalies one has to do MRI in these cases to exclude underlying orbital dermoid 2 hearing test 3 X-ray limbs and spine and ultra sound abdomen to exclude kidney involvement key words 1 limbal dermoid 2 pre auricular skin tag 3 squint 4 hearing defect 5 impairment of intelligence 6 spine and limb deformities 7 congenital heart introduction Goldenhar syndrome is a rare condition characterised by congenital presence of a limbal dermoid and congenital pre auricular appendage limbal dermoid may sometimes be bilateral they either involve entire cornea or may be confined to conjunctiva only incidence is 1 in 500 to 2500 inferotemporal site of limbal dermoid is the commonest about 70 percent results are satisfactory most of these cases live a normal life and most of them have normal intelligence case report 3 years male child was seen by me in my office 6 months back with parents having noticed an oval palish white inferotemporal limbal lesion left eye with congenital presence of pre auricular skin tag right ear first child born after LSC section breast fed normal stones no other congenital deformities 1 normal intelligence hearing no deformities of teeth limbs and spine discussion limbal dermoids are graded according to involvement of cornea by grade v1 when only epithelium is involved grade 2 des membrane grade 3 entire anterior segment my case was grade 1 limbal dermoid and had the commonest site of inferotemporal vision refraction and fundus was normal so was MRI orbits and ultrasound abdomen conclusion the treatment is 1 visual and 2 cosmetic if limbal dermoid involves pupillary area and the vision one can do 1 lamellar keratoplasty 2 amniotic membrane graft 3 stem cell graft references there are families of Greek who have Goldenhar syndrome in Gulf War children were born with Goldenhar syndrome born in various military hospitals 3 drusen of optic nerve head is reported to be associated with Goldenhar syndrome.

Biography

Gowhar Ahmad has pursued his MBBS from University of J&K and Master of Surgery in Ophthalmology, SN Medical College, University of Agra. He had Fellowship in paediatric ophthalmology, Moorefield's Eye Hospital, London. Also had Fellowship in Occuloplasty and Neuro ophthalmology from KK eye specialist hospital, Riyadh, KSA. Ex Sr consultant and head ophthalmology, Qatif Central hospital, Eastern province, KSA. Presently working as sr consultant ophthalmologist in Florence hospital, Chanapora, Srinagar, Kashmir, India. Special interests are Squint surgery, ROP screening in premature children, Oculoplastic surgery, Ptosis surgery, Lacrimal duct procedure for dacryocystitis, fitting of prosthesis, medical ophthalmology, glaucoma screening, IOL implantation and glaucoma surgery.

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