

Association of Goldenhar with Duane's Retraction Syndrome

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Abstract

A six-year girl presented with history of deviation of eye and facial asymmetry. Diagnosis of Goldenhar syndrome (GHS) with Duane retraction syndrome (DRS) was made. This case highlights DRS as an unusual but known association of Goldenhar Syndrome. Patient with GHS presenting to an ophthalmologist should be screened for vertebral, cardiac, renal and central nervous system defects.

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A six-year girl presented with history of deviation of eye and facial asymmetry (A). On examination, there was a notch in the upper lid (B) and preauricular skin tags (C). Ocular motility showed (D) deficient adduction (black arrow) and abduction (white arrow). Diagnosis of Goldenhar syndrome (GHS) with Type III Exotropic Duane retraction syndrome (DRS) was made.

GHS is known by various names depending upon the components of the disease complex. This includes craniofacial microsomia, oculo-auriculo-vertebral syndrome, hemifacial microsomia and 1st and 2nd branchial arch syndrome. The pathognomic triad includes presence of epibulbar dermolipoma or bulbar dermoid, low set ears and vertebral skeletal anomalies.^{1,2}

GHS affects derivatives of the first and second brachial arch³ Abnormalities are unilateral in 85% and bilateral in 10–33% of the cases and the right side is more frequently affected.⁴ Ophthalmic features include microphthalmia, dermoids, coloboma, cataract and strabismus. Ocular anomalies mainly bilateral dermoids are seen in 60%, vertebral anomalies in

40%, and ear anomalies in 40% of the cases.

DRS is an unusual but known association of GHS. Incidence of Duane Retraction Syndrome (DRS) with GHS is around 5% in sporadic and 10% in familial cases. The frequent association of DRS with other congenital anomalies might be suggestive of teratogenic event occurring between the fourth to eighth week of gestation as an etiological factor.⁶

Patient with GHS presenting to an ophthalmologist should be screened for vertebral, cardiac, renal and central nervous system defects

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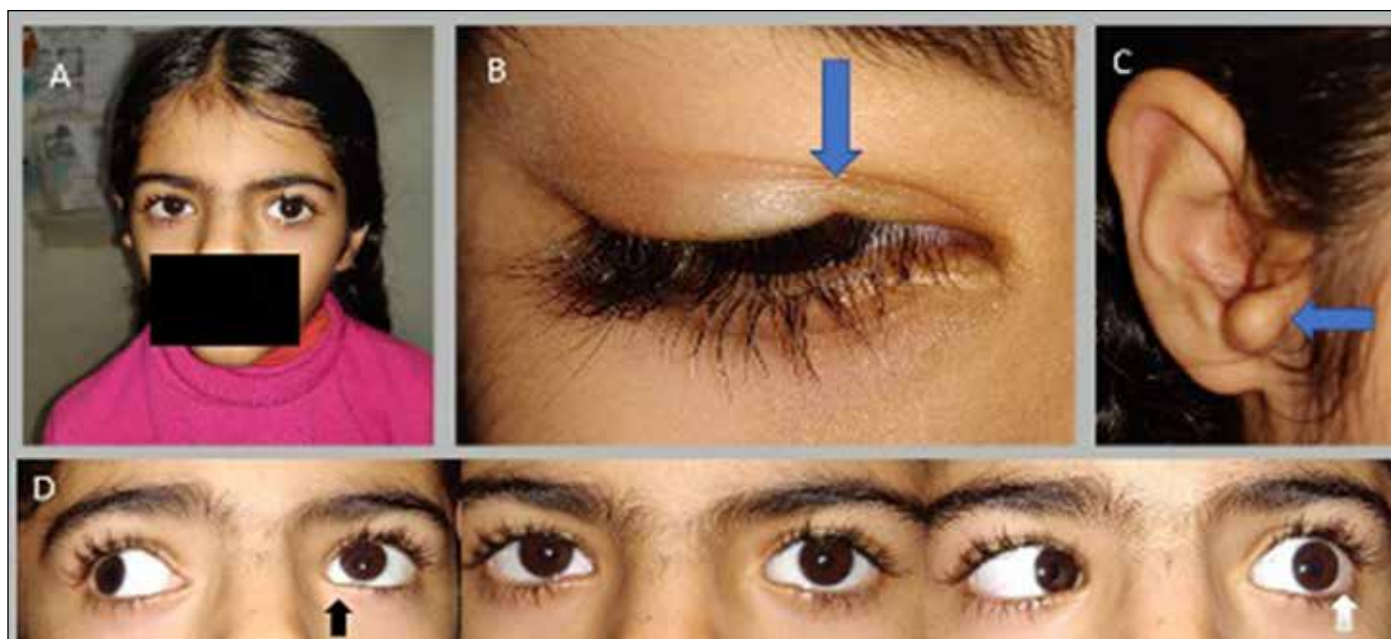


Figure 1: Composite photograph of a 6-year girl highlighting the association of Goldenhar and Duane's Retraction Syndrome. A: She presented with a facial asymmetry and low set ears. B: There was a notch in the upper lid and preauricular tags (C). Ocular motility revealed deficient adduction and abduction (D).

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