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GOLDENHAR'S SYNDROME: CASE REPORT

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SUMMARY

A case of Goldenhar's Syndrome (oculoauriculovertebral dysplasia) in a 3-day-old Nigerian neonate with right anophthalmos and lipodermoid, left limbal dermoid, bilateral preauricular appendages and mandibular hypoplasia is presented. The lipodermoid was attached to the tarsal conjunctiva of the lower lid of the anophthalmic side. The causative factor was presumably maternal drug (traditional medicine) ingestion at three months gestation. No similar case has been reported previously. The literature on Goldenhar's Syndrome is briefly reviewed.

INTRODUCTION

Goldenhar's Syndrome (GS), otherwise known as oculoauriculovertebral dysplasia is a rare congenital complex of abnormalities with various modes of expressivity. It was first described by von Arlt in 1845 but was not recognised as a syndrome until 1952 when Goldenhar described the classical triad of epibulbar dermoids, preauricular tags/fistulas and vertebral anomalies(1). The prevalence rate was estimated to be 1:45,000 neonates(2). Most cases are sporadic and unilateral, but familial instances have been reported in first degree relatives(3,4). Other common associated anomalies include coloboma of the upper eyelid, hypoplasia of the mandible or maxilla, ocular colobomata, microphthalmos and rarely, Duane's retraction syndrome(3,5,6). Anophthalmos when it occurs, is often associated with cerebral malformations, hydrocephalus and mental retardation(7). Systemic abnormalities reported in association with GS include cardiovascular, pulmonary, labyrinthine, tracheo-esophageal, renal and genitourinary(2,5). The typical anomalies develop mainly from the first branchial arch during blastogenesis. Aetiology has been traced to teratogenic agents like maternal drug ingestion, maternal infections and irradiation(6,8).

CASE REPORT

A 3-day-old male Nigerian child presented with a history of a mass in front of the right eye since delivery. His mother was 24 years old and father, 28 years old at the time of his birth. He was the product of his mother's second pregnancy; the first terminated by spontaneous abortion at about three months gestation. Prenatal history revealed that the mother obtained ante-natal care in a private hospital and had spontaneous unassisted vaginal delivery at full term. The mother was generally healthy during pregnancy. There was no skin eruption or exposure to radiation. The drugs used were mainly haematinics. She however ingested some quantity of traditional medicine concoction at three months gestational period as a

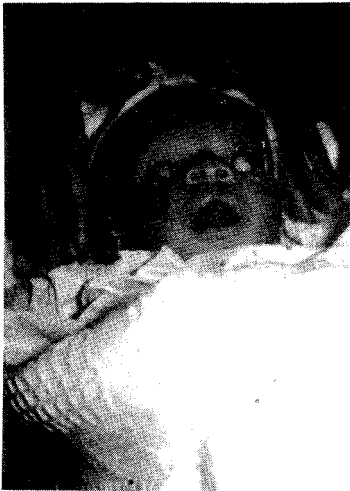
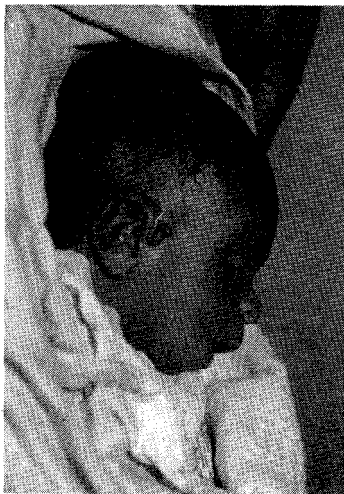
prophylaxis against recurrent abortion.

Ocular examination showed right anophthalmos and a soft solid mass covered with skin, protruding from the right socket (Figure 1). The mass, measuring 5cm x 5cm, was attached to the tarsal conjunctiva of the lower lid by a 5mm base located in the middle one third of the lid, approximately 4mm posterior to the lid margin. The upper lid was normal. The socket was shallow and completely lined by conjunctiva. There was no demonstrable eyeball. The left eyeball was of normal size (Figure 2), with a 3mm x 3mm limbal dermoid temporally. He had bilateral preauricular appendages; the right being bilobed (Figure 2). On general physical examination, there was no clinically detectable vertebral or cardiac anomaly but he had a tense anterior fontanel and mandibular hypoplasia (Figure 3). Radiological examinations and excision biopsy of the lipodermoid were planned but the patient was lost to follow-up.

Figure 1

Right lipodermoid and preauricular appendages



Figure 2*Normal sized left eyeball***Figure 3***Mandibular hypoplasia*

DISCUSSION

Goldenhar's Syndrome or Oculoauriculovertebral dysplasia has been known since 1952 as a rare congenital group of anomalies resulting from maldevelopment of the first and second branchial arches and the first branchial cleft(5). A review of the literature indicates that GS commonly occurs sporadically and there is no sex predilection. Baum and Feingold reported that 90% of cases had no family history(3). Stoll *et al* however reported the case of a mother and

two of her children with GS, showing that it may be inherited as an autosomal or X-linked dominant condition(4).

Clinical features of GS commonly include dermoids, which may be epibulbar or orbital. Dermoids occurred as frequently as 78% in one series(3). They are unilateral in 30-53% of cases(3). Irregular astigmatism may result from large and extensive dermoids encroaching on the cornea. Secondary strabismus due to anisometropic amblyopia may occur(3,5). Histologically, they are choristomas composed of dermis-like collagenous connective tissue, which may contain sebaceous glands, hair follicles, nerve bundles and covered by squamous epithelium(9).

Lipodermoids are less frequent than epibulbar dermoids, occurring in 47% of cases. Twenty-eight percent are unilateral and 19% bilateral(3). They are usually located subconjunctivally and rarely seen attached to the skin at the lateral canthus(3,9). In the case reported, the lipodermoid was attached to the inferior conjunctiva close to the lid margin. Histologically, they are solid masses consisting of fatty and fibrous tissue, covered by keratinized or non-keratinized epithelium with hairs on the surface of the lesion(9). Upper eyelid coloboma is present in 24-60% of GS cases(3,5,6). It commonly occurs on the more affected side and is bilateral in 3%.

Anophthalmos is a rare ocular presentation of GS(7). Clinically, it is often difficult to differentiate between complete absence of the eye and extreme microphthalmos as only serial histological sections of the orbit can confirm this(9). Children with GS and anophthalmos or microphthalmos tend to be at increased risk for cerebral malformations and mental retardation(7). The child reported had a full anterior fontanel and may well manifest cerebral signs if followed up. Other rare ocular manifestations of GS include iris abnormalities, Duane's retraction syndrome(3,5) and reduced corneal sensation with neuroparalytic keratitis, due to a defect in the ophthalmic division of the trigeminal nerve, probably at the nuclear level(1,3).

Ear abnormalities vary but as a rule, preauricular appendages are a must in the diagnosis of GS(5). All of Baum's series had preauricular appendages and only 30% were bilateral. They were bilateral in the child reported, with one side bilobed. Malformation of the external ear may be present and external auditory canals may be slightly narrowed or atretic(5).

The skull and face may be underdeveloped with facial asymmetry, which is usually unilateral, but may be bilateral, with one side more severe than the other. Cleft palate, cleft lip and tongue do occur(5,9). Only mandibular hypoplasia was demonstrable in the case reported. Vertebral anomalies occur in 35-60% of patients(3,5). Rarely, phocomelia has been reported in association with GS(9).

Concomitant systemic abnormalities that may occur

include cardiovascular, pulmonary, renal, neural and gastrointestinal(2,5,9). The aetiological factor in the case reported was presumably traditional medicine (a concoction of herbal extracts) ingested by the mother within the first trimester of pregnancy. No similar case has been reported in literature. Teratogenicity of the concoction could not be ascertained, as the chemical composition is unknown. The mother admitted that besides the concoction, she only ingested haematinics. It has been reported that maternal drug ingestion, exposure to hazardous chemicals in the environment, maternal viral infection and irradiation within the first trimester of pregnancy can predispose to GS(6,8). Lessick *et al*(8) reported a cocaine-exposed infant who developed severe manifestations of GS.

Generally, the management of patients with GS is plastic surgery; excision of lipodermoid and dermoid cyst, with lamellar keratoplasty in cysts that encroach on the cornea with significant astigmatism. Early prenatal diagnosis of GS is possible, with the option of pregnancy termination. De Catte *et al* reported sonographic detection of GS in a fetus at 15 weeks gestation, by observing a maxillary cleft in association with unilateral microphthalmia(10).

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REFERENCES

1. Mohandessan, M.M, and Roman, P.E. Neuroparalytic keratitis in Goldenhar Gorlin syndrome. *Amer. J. Ophthalmol.* 1978; **85**:111-113.
2. Morrison, P.J., Mulholland, H.C., Craig, B.G. and Nevin, N.C. Cardiovascular abnormalities in the oculo-auriculo-vertebral spectrum (Goldenhar syndrome). *Amer. J. Med. Genet.* 1992; **44**: 425-428.
3. Baum, J.L. and Feingold, M. Ocular aspects of Goldenhar syndrome. *Amer. J. Ophthalmol.* 1973; **75**: 250-257.
4. Stoll, C., Viville, B., Treisser, A. and Gasser, B. A family with dominant oculoauriculovertbral spectrum. *Amer. J. Med. Genet.* 1998; **78**: 345-349.
5. Duane, T.D. and Jaeger, E.A. *Clinical Ophthalmology*. Revised edition, Philadelphia. Harper & Row Publishers. 1987; **2(30)**: 1.
6. Warburg, M. Update of sporadic microphthalmos and coloboma. Non-inherited anomalies. *Ophth. Paed. Genetics.* 1992; **13**: 111-122.
7. Schrandt-Stumpel, C.T., de Die-Smulders, C.E., Hennekam, R.C., Fryns, J.P., Bouckaert, P.X and Brouwer O.F. *et al*. Oculoauriculovertbral spectrum and cerebral anomalies. *J. Med. Genet.* 1992; **29**: 326-331.
8. Lessick, M., Vasa, R. and Israel, J. Severe manifestation of oculoauriculovertbral spectrum in a cocaine exposed infant. *J. Med. Genet.* 1991; **28**: 803-804.
9. Yanoff, M. and Fine, B.S. *Ocular Pathology. A text and atlas*. 2nd edition, Philadelphia. Harper & Row Publishers. 1982; 316-319.
10. De Catte, L., Laubach, M., Legein, J. and Goossens, A. Early prenatal diagnosis of oculoauriculovertbral dysplasia or the Goldenhar syndrome. *Ultrasound in Obstetrics and Gynecology.* 1996; **8**: 422-424.