Goldenhar syndrome: A case report and literature review

* C. O. Bekibele, S. A. Ademola[1], S. D. Amanor-Boadu[2],
E. E. Akang[3] and K. O. Ojemakinde[3]
*Department of Ophthalmology, [1]Plastic and Reconstructive Surgery Unit,
Department of Surgery, [2]Department of Anaesthesia,
[3]Department of Pathology, College of Medicine, University of Ibadan,
University College Hospital, Ibadan.

Summary

The case of a 24-year-old female Nigerian with features of Goldenhar syndrome is presented and the challenges of management especially with reference to reconstructive facial surgery and general anaesthesia are discussed.

Keywords: ***Oculo-auriculo-vertebral dysplesia, Limbal dermoid, First branchial arch, Cleft anomaly, Congenital anomaly.***

Résumé

L'objet de cette étude est le cas d'une femme nigeriane âgée de 24 ans avec des traits du syndrome de Goldenhar et les défis de la prise en charge tout particulièrement en ce qui concerne la chirurgie réparatrice de la face et l'anesthésie générale.

Introduction

Oculo-auriculo-vertebral dysplasia or Goldenhar syndrome is a rare congenital anomaly. The syndrome which is characterised by a triad of anomalies comprising epibulbar dermoid, accessory auricular appendages and aural fistula was first characterised by Goldenhar in 1952 [1]. Other anomalies have since been observed in the patients leading to the adoption of the name oculo-auriculo-vertebral dysplasia as suggested by Gorlin who also included vertebral anomalies in 1963 [2]. The aetiology in most cases is often difficult to ascertain. Some cases appear to be genetic[3], while others occurring in a sporadic manner, are probably due to environmental factors[4]. However, whatever the influence, it must be operational on the derivatives of the first and second branchial arches and clefts before the end of the organogenetic period (7th or 8th week of embryonic life)[5]. The prevalence of Goldenhar syndrome has been estimated to be in the range of 1:45,000 neonates[6]. We present the case of a 24-year-old lady with features of Goldenhar syndrome, seen and is being managed at the University College Hospital Ibadan. It is noted that presentation in adulthood is not common and the challenges of management in relation to facial reconstructive surgery and anaesthetic problems are discussed.

Case presentation

O.E, a 24-year-old female tailoring apprentice was seen at the Eye Clinic of the University College, Hospital, Ibadan with the complaints of swelling in both eyes, total loss of right eye vision, poor left eye vision and deformed facial appearance, all since birth. She also gave a complaint of defective hearing with the right ear, and abnormal gait. None of the parents was available for questioning on the antenatal, birth, and early childhood history since the mother had absconded from the matrimonial home soon after giving birth to her. She was the second sibling in a polygamous setting and the only child of the mother. The father, a commercial driver by profession and about fifty-years old had five other wives, and there was no history of similar problem in any of her siblings or in the extended family.

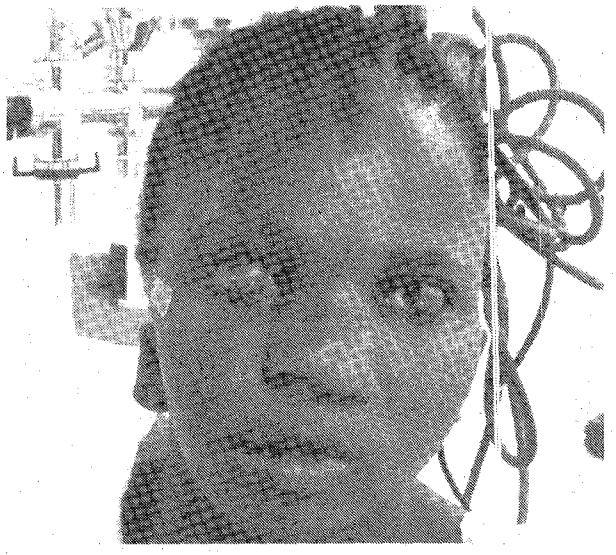


Fig. 1 *Front view of patient with Goldenhar syndrome*

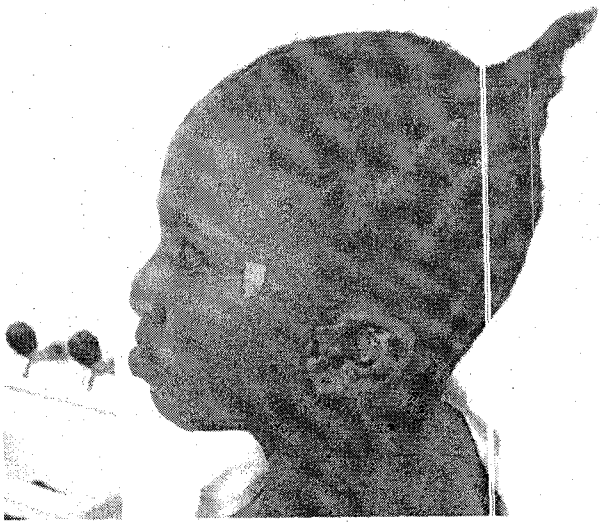


Fig. 2 *Left ocular and auricular anomalies of patient with Goldenhar syndrome*

Examination revealed a young lady with small stature (weight, 43kg; height, 1.35meters) and dysmorphic facial appearance. She had facial asymmetry with low set ears, bilateral pre auricular tags, and low set, deformed pinnae, and microtia. She also had right mandibular and maxillary hypoplasia, and hypoplastic right upper premolar and molar

* *Correspondence*

Fig. 3 *Right ocular, facial and auricular anomalies of patient with Goldenhar syndrome*

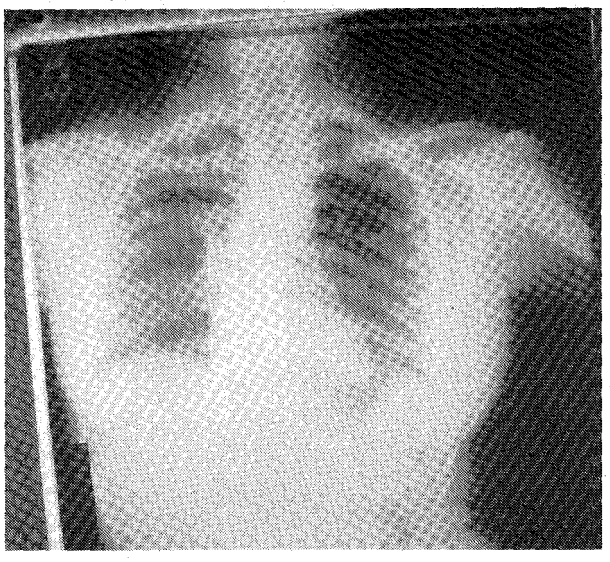
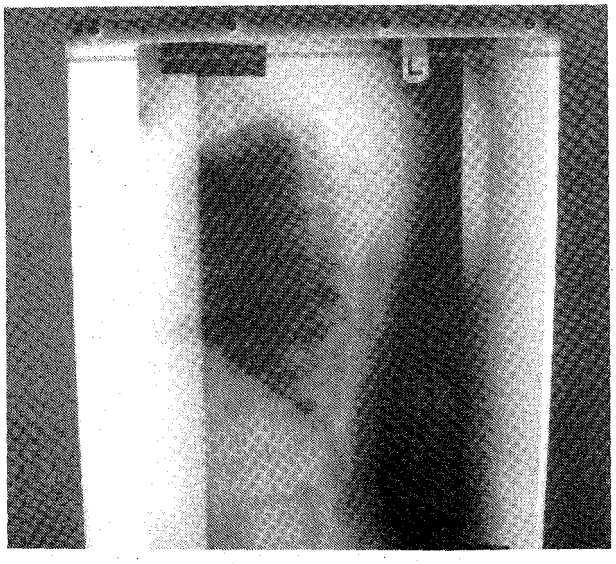


Fig. 4 *Chest X-ray showing bony fusion of posterior ribs of patient with Goldenhar syndrome*



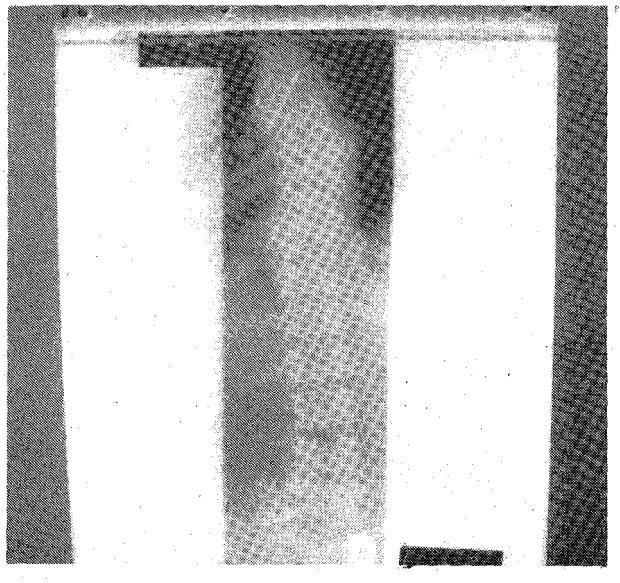


Fig. 5a & b *X-ray dorso-lumbar spine showing multiple vertebral abnormalities in patient with Goldenhar syndrome.*

teeth. The nasal bridge was flattened and the nasal septum was deviated to the right side with engorged inferior turbinates on both sides. Figures 1, 2 and 3 show the ocular, facial and auricular anomalies.

The central nervous system examination revealed an intelligent, conscious, alert and well orientated lady, with a muffled nasal speech. Her cutaneous sensations, long tracts, muscle tone, bulk and power were normal. There was kyphoscoliosis of her thoracolumbar spine and hypoplasia of her left lower leg (tibia and fibular). Other systems were essentially normal.

The right ocular examination revealed a visual acuity of no perception of light, extensive upper lid coloboma and the eye was covered by dermoid tissue with very little view of keratinised conjunctiva. There was no view of the cornea and other ocular contents. The left eye had a visual acuity of 6/24, the eyelids were intact but the temporal limbus and temporal half of the cornea were covered by epibulbar dermoid tissue. The rest of the left eye appeared normal. Ocular ultra

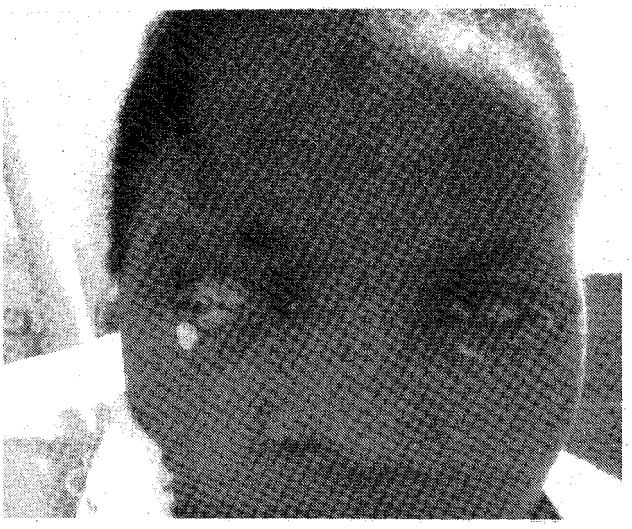


Fig. 6 *Post op front view of patient with Goldenhar syndrome.*

sound scan revealed normal size right globe, with cataractous lens, normal aqueous, vitreous, retina and retro bulbar areas. The left eye was normal. The chest X-ray (Figure 4) showed double scoliosis of the dorsal spine. There was bony fusion of multiple posterior ribs at their costovertebral ends (6 and 7, 8 and 9 on the right; 9 and 10 on the left. No soft tissue abnormality was seen. X-ray of the dorso-lumbar spine (Figure 5) showed multiple vertebral abnormalities, including hemivertebrae and block vertebrae involving the dorsal spine, with resultant scoliosis. Other abnormalities include spina bifida oculta at D11. The disc spaces appeared relatively spared.

An attempt to carry out an estimation of the respiratory vital capacity prior to anaesthetic exposure for surgery was abandoned because patient could not pout her mouth to exhale maximally. However, an attempt at Peak Expiratory Flow Rate (PEFR) done was 200 litres /minute. The Mallampati[24] score to assess difficult intubation was 3, hence difficult intubation was predicted.

Anaesthesia was induced with nitrous oxide, oxygen and halothane. After test ventilation, suxamethonium was administered but she was difficult to intubate, the laryngeal structures could not be visualized. Intubation was abandoned after several attempts and anaesthesia was administered via laryngeal mask airway. The left eye epibulbar dermoid was successfully excised without sequel but the right eye with more extensive lesion was a more difficult challenge. The extensive dermoid tissue was excised and the bare sclera and part of the tarsal conjunctiva was covered by mucosal membrane harvested from the buccal mucosa and a temporary tasorrhaphy done. The histology of excised epibulbar tissue was in keeping with dermoid tissue.

Post operatively (Figure 6), the patient had improved vision of hand movement right eye, and 6/18 left eye. The left eye was free of dermoid tissue and only a minimal cornea scar. The right eye on the other hand had a dense corneal opacity, cataract, residual dermoid tissue in the inferonasal fornix with associated symblepharon, upper lid coloboma with the associated risk of cornea exposure.

The outstanding problems requiring further intervention and specialised care include, right cornea graft and cataract extraction, excision of auricular tags, reconstruction of deformed pinnae, and nasal bridge as well as provision of hearing aid for the defective hearing. Kyphoscoliosis and compressed ribs may predispose the patient to restrictive airway disease in the future.

Discussion

Goldenhar syndrome or oculoauriculovertebral dysplasia is a rare congenital anomaly that has no sex predilection and more often presents in childhood than in adulthood[3]. Presentation in adulthood is rare because of the multiple anomalies, which would have necessitated early medical consultation and intervention in childhood in the developed world. In the developing word, babies with gross congenital anomalies are often abandoned[23] as observed in this case and our patient may have survived this long because of the absence of cardiovascular and central nervous system anomalies[7]. Originally characterised by Goldenhar to consist of a triad of epibulbar dermoid cyst, accessory auricular appendages and aural fistula[1]. The aetiology of the syndrome is often difficult to ascertain. Baum and Feingold[3] reported this syndrome to be sporadic in over 90% of cases while positive family history was also reported in the maternal grandmothers and mother of two cases in their series[3]. Maternal diabetes is thought to be an aetiologic factor[8] and a report of Goldenhar's syndrome in a 3 month old infant from Ile-Ife, Nigeria incriminated maternal ingestion of traditional herbal medication during pregnancy.[4] It was not possible to determine the aetiology in this case since the mother and other close relations were not available for interrogation.

The major ocular features of the disease are dermoid or lipodermoid cysts. They are usually in the inferotemporal quadrant and limbal dermoids are reported more frequently than lipodermoids. They are most often unilateral (75%) than bilateral, with the right side more often affected than the left[9]. This was as observed in our case. The dermoid cysts usually impinge on the visual axis but more commonly interfere with vision by causing astigmatism and predisposing to secondary strabismus from anisometropia. Another common finding is an upper eyelid coloboma almost always on the most affected side. Less common ocular anomalies include Duane syndrome[20], dacryocystitis, stenosed lacrimal duct, anophthalmos, cryptophthalmos, microcornea decreased cornea sensation, cataract and iris abnormalities (including poorly reactive elliptical pupil and coloboma of iris)[3,10]. Other ocular abnormalities include, decreased tear production, cornea ulcer[10], caruncle abnormalities, eyelid tags, pendular nystagmus, pseudopapilloedema, canthal coloboma and proptosis [3,11]. Our patient did not have these problems, and with her limited vision was able to read and was apprenticed for a trade that required acute sense of vision.

Various cardiovascular anomalies are associated with the syndrome [3,12]. They include major anomalies like Tetralogy of Fallot, dextrocardia and transposition of the great vessels which are of grave significance as co-existing disease. Other cardiac congenital anomalies include, right bundle branch block, pulmonary stenosis and mitral incompetence, patent ductus arteriosus, atrial septal defect. Our patient did not have any cardiovascular anomaly. Central nervous system anomalies such as hydrocephalus, meningoencephalocoele and mental retardation [13] have been associated with the syndrome but our patient did not exhibit any of these. Other systemic anomalies that may be associated include vertebral anomalies[21], kidney defects, urethral defects, rectal and anal defects, inguinal hernia, hemangiomas, recto vaginal fistula and club-feet [3,14].

Our patient had all three components of the Goldenhar triad of epibulbar dermoid, accessory auricular appendages and aural fistula as well as defective hearing on the right side, defective facial bones and vertebral anomalies principally limited to the dorso-lumbar spine. She also had a defective right lower limb.

The major challenges in the management of this patient had to do with her cosmetic appearance. Her initial management team consisted of an ophthalmologist, a plastic surgeon and an anaesthetist. In view of the extensive external

deformities it was expected that she would require a multistage approach in the management. The initial management consisted of simple excision of her ocular dermoid cysts to provide an improvement in the visual acuity and ocular appearance. General anaesthesia was required for the excision of the epibulbar dermoids and difficulty was encountered with tracheal intubation and ultimately anaesthesia was successfully provided with the aid of a laryngeal mask. Difficult tracheal intubation has been reported to be as high as 39.5% in association with first and second branchial arch syndromes[15]. Difficult tracheal intubation may be overcome by use of tracheostomy [16], modified nasopharyngeal tube[17], and tracheal intubation using suspension laryngoscope[18]. Early surgery (in the first decade of life), has been advocated by Hunt and Hobar[19] who have observed better results when surgery is done before the age of ten. As regards the hearing defect on the right side a cochlear implant[22] may have to be considered.

Conclusion

Goldenhar syndrome is a rare congenital abnormality associated with cosmetically unacceptable defects whose management may pose numerous challenges and requires a multistage and multidisciplinary approach for its optimal management.

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