Communications

Clinical aspects of oculo-auriculo-vertebral dysplasia

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Oculo-auriculo-vertebral dysplasia is a condition characterized by epibulbar dermoids or dermolipomata, auricular deformities, and vertebral anomalies. Many other abnormalities have been described, but these three signs are the most constant and give the syndrome its identity. In this communication we are presenting four further cases each of which demonstrates additional clinical and radiological features.

Case reports

Case 1, a 16-month-old boy was admitted to hospital for the removal of a dermolipoma from his left eye. This had been noted soon after birth, but surgery had been deferred.

He had a right indirect inguinal hernia repaired at the age of 3 months and he had had apparent difficulty in swallowing throughout his first year, but a barium swallow had been normal.

EXAMINATION

A dermolipoma was situated on the lower temporal quadrant of the left globe encroaching onto the cornea. This appeared to have increased in size since it was first observed 12 days after birth. A similar but smaller tumour was noted in the lower temporal region of the right eye several millimetres posterior to the limbus adjacent to the conjunctival fornix.

GENERAL EXAMINATION

- (1) Hypoplasia of the left side of the face.
- (2) Deformed left pinna.
- (3) Blind left external auditory meatus which was represented by a small pit only.
- (4) Abnormal left thumb, with the phalanges apparently connected to the rest of the hand by soft tissue only (Fig. 1, overleaf).

RADIOLOGICAL EXAMINATION

(1) *Multiple vertebral and costal anomalies* Spina bifida occulta of cervical and upper thoracic vertebrae; fusion of a cervical rib with right first rib and fusion of a hypoplastic left first rib with the second; fusion of body of T4 with left half of T5; hemivertebrae at T5 and at the thoraco-lumbar junction.

(2) *Anomalies of facial bones* Hypertelorism; hypoplasia of left mandibular ramus.

(3) *Temporal bone anomalies* Atresia of left external auditory canal; elevation of left cochlea producing asymmetry of the petrous temporal bones; underdevelopment of mastoids.

(4) *Abnormal left thumb* The digit arose distally and had three hyperplastic bones with the configuration of phalanges; the metacarpal bone was absent.

FIG. 1 Case 1.
Abnormal left thumb

COURSE

The left dermolipoma was removed under general anaesthesia. It was closely adherent to the underlying sclera and cornea and after dissection the exposed corneal stroma was left bare. The excised specimen consisted of a soft mass with a number of hairs projecting from its surface and histological examination showed that it was composed of adipose tissue. After the operation a purulent discharge developed and pressure on the lacrimal sac resulted in regurgitation of pus, from which *D. pneumoniae* were cultured. The left nasolacrimal duct was probed and the discharge cleared within a few days. A month later, however, the eye had become re-infected and a central corneal ulcer had developed. Corneal sensation at this time was shown to be absent. A further probing again reduced the amount of discharge, but the ulcer persisted and for 6 weeks remained unchanged. Finally it was decided that the diseased cornea should be replaced by a graft, and, in order to prevent further re-infection from the lacrimal sac, a dacryocystectomy was done as a preliminary procedure. After a further 4 weeks, when the child was aged 19 months, a 5.1 mm. central penetrating corneal graft was performed. The post-operative course was uneventful, there was no further infection, and the graft has remained clear (Fig. 2).

FIG. 2 Case 1. *Appearance of graft in left eye 10 months after surgery*

COMMENT

This is a typical case of oculo-auriculo-vertebral dysplasia, showing as it does the characteristic triad of signs. The complications arising after the excision of the left dermolipoma can be directly attributed to corneal anaesthesia and obstruction of the nasolacrimal duct. Sugar (1967) has reported the absence of corneal sensation in this syndrome but we have found no references to nasolacrimal duct obstruction. We believe also that this is the first report of a penetrating corneal graft done in a patient with this syndrome, although lamellar grafts have been described (Sugar, 1966; Heimann, 1968).

Case 2, a girl aged 6 years, was seen in the Eye Clinic in January, 1970, for advice about her right eye which periodically became red and painful. The trouble had been present since the age of 11 months at which time a tarsorrhaphy had been done; this had broken almost immediately.

EXAMINATION

The right eye showed a central corneal scar with some vascularization (Fig. 3) and the visual acuity was hand movements only. There was total corneal anaesthesia and a dermolipoma was present at the limbus in the lower temporal quadrant (Fig. 3). The left eye was normal with a visual acuity of 6/4. The nasolacrimal ducts were freely patent to syringing.

FIG. 3 Case 2. *Appearance of right eye, showing corneal scarring and dermolipoma*

GENERAL EXAMINATION

- (1) Hypoplasia of the right side of the face (Fig. 4, overleaf).
- (2) Deformed right pinna (Fig. 5, overleaf).
- (3) The remains of preauricular skin tags which had been removed in the past.
- (4) Blind right external auditory meatus.
- (5) Abnormally mobile interphalangeal joint of the right thumb.
- (6) Bilateral pes planus.
- (7) Bifid uvula.
- (8) Peculiar gait. This was the result of hip abnormalities for which she had had plates inserted at the age of 4 years. These have just recently been removed.

RADIOLOGICAL EXAMINATION

- (1) *Multiple vertebral and costal anomalies* Hemivertebra at T5 and cuneiform vertebrae at T2 and T3; atresia of right first rib and fusion of right second and third ribs; spina bifida occulta of cervical vertebrae.
- (2) *Anomalies of skull and facial bones* Biparietal foramina; abnormally small pituitary fossa; hypoplasia of right maxilla including the antrum; hypoplasia of right side of mandible.
- (3) *Right temporal bone anomalies* Atresia of external auditory canal; absence of ossicles; malformed elevated cochlea; short hypoplastic external auditory canal with acute upward inclination; underdeveloped diploic mastoid.

FIG. 4 Case 2. Full face, showing right hypoplasia

FIG. 5 Case 2. Side face, showing deformed right pinna

COMMENT

This patient also presents a typical case of oculo-auriculo-vertebral dysplasia. The bifid uvula is an unusual feature but has been previously reported (Neimann, Cordier, Manciaux, and Barrucand, 1960). The extensive corneal scarring was almost certainly related to the absence of corneal sensation. As the eye was likely to be amblyopic and there was no active ulceration, keratoplasty was not advised.

Case 3, an infant girl aged 1 month, was seen in 1963 because she apparently had only one eye. She was examined under general anaesthesia and indeed there was no evidence of an eye on the left side (Fig. 6); the socket was empty and the bony orbit was only half the normal size. The right eye was normal apart from a limbal dermoid (Fig. 6). When we reviewed her recently at the age of 8 years, we found no change in her ocular state but we noted in addition that her corneal sensation was much reduced. Her mother told us that she had a recurring discharge from her right eye but that this gave her little trouble. A sac wash-out was not done.

GENERAL EXAMINATION

- (1) Hypoplasia of the right side of mandible (Fig. 6, opposite).
- (2) Preauricular skin appendages.
- (3) Cleft palate. This had been treated surgically when she was 2 years old.
- (4) Scoliosis (Fig. 7, opposite).
- (5) Paralytic equino-varus of left foot. This has been under treatment since the age of one month.
- (6) Short atrophic left lower limb below knee level (Fig. 8, opposite).
- (7) Imperforate anus with recto-vaginal fistula.

FIG. 6 Case 3. Full face, showing absence of left eye, right limbal dermoid, and facial asymmetry

FIG. 7 Case 3. Back, showing scoliosis

FIG. 8 Case 3. Short atrophic left lower limb

RADIOLOGICAL EXAMINATION

- (1) *Multiple vertebral anomalies* Failure of segmentation resulting in block vertebrae throughout the thoracic and lumbar spines; fusion of the right lower eight ribs; cuneiform and hemivertebrae.
- (2) *Anomalies of skull and facial bones* Biparietal foramina; an abnormally small pituitary fossa; hypoplasia of the left orbit and left side of mandible; aplasia of the right mandibular ramus.
- (3) *Temporal bone anomalies* Hypoplasia of the bony external auditory canals; absence of the ossicles on the left; hypoplastic internal auditory canals and a malformed elevated cochlea on the right underdeveloped mastoids.
- (4) *Right-sided hydronephrosis and hydroureter* (IVP)

COMMENT

This girl has all the features of oculo-auriculo-vertebral dysplasia as well as a large number of additional anomalies. The feature of corneal anaesthesia is again noted here but in this case there has never been any corneal ulceration.

Case 4, a woman aged 29 years, was referred to the Eye Clinic for advice about her ptosis. She was deaf and dumb but her sister, who used sign language, was able to communicate with her without difficulty. She had had a series of operations on her ears at the age of 13 but there had been no improvement in her hearing. Her eyes had been abnormal since birth and a ptosis operation was performed on the left side at the age of 5.

EXAMINATION

There was a gross divergent squint, bilateral ophthalmoplegia, and marked bilateral ptosis with scarring of the left upper lid (Fig. 9). In the left eye abduction was full and some depression was possible, but elevation and adduction were absent. In the right eye partial abduction was the only movement elicited. She adopted an abnormal head posture to compensate for her ptosis and

FIG. 10 Case 4. Fingers, showing hypoplasia of terminal phalanges

FIG. 9 Case 4. Full face, showing ptosis and squint

ophthalmoplegia. She had no diplopia. The corrected visual acuity was 6/9 in the right eye and 6/12 in the left. The fundi were normal. There was a lobulated epibulbar dermolipoma a few millimetres to the lateral side of the limbus on the right eye. Corneal sensation was normal on both sides and the naso-lacrimal ducts were patent on syringing.

GENERAL EXAMINATION:

- (1) Deformed right pinna.
- (2) Blind external auditory meati on both sides.
- (3) Hypoplasia of terminal phalanges on several fingers and toes (Fig. 10).

RADIOLOGICAL EXAMINATION

- (1) *Vertebral anomalies* Abnormally high vertebral bodies with narrow disc spaces and marked osteophyte formation; narrow spinal canal.
- (2) *Skull anomalies* Patchy hyperostosis of frontal bones; abnormally small pituitary fossa.
- (3) *Temporal bone anomalies* Bilateral atresia of external auditory canals; bilateral absence of ossicles; underdeveloped mastoids.
- (4) *Hand abnormalities* Hypoplasia of third, fourth, and fifth terminal phalanges of both hands.

COURSE

Under general anaesthesia the dermolipoma was removed and the right lateral rectus recessed 8 mm. Histological examination of the excised specimen showed that it consisted largely of adipose tissue. Post-operatively a purulent conjunctivitis developed with a marginal ulcer adjacent to the site of excision of the dermolipoma. The condition resolved in a few days with the administration of topical antibiotics. The effect of the squint surgery was disappointing and the cosmetic improvement slight. The patient is soon to be fitted with a shelved contact lens to assist the ptosis.

COMMENT

Although the ophthalmoplegia and ptosis are the most striking features of this case, we believe it may be classified as oculo-auriculo-vertebral dysplasia since it shows the association of an epibulbar dermolipoma, auricular abnormalities, and spinal changes. As in Case 1 a corneal ulcer developed after the excision of the dermolipoma. It resolved rapidly but if corneal anaesthesia or naso-lacrimal duct obstruction had been present the course may well have been more protracted. The absence of discrete middle ear ossicles was demonstrated by tomography of the petrous temporal bones. Unfortunately we have been unable to obtain any records relating to the operations performed on the patient's ears so we cannot say with any certainty whether this anomaly is congenital or the result of surgery.

Discussion

Goldenhar (1952) drew attention to the association of epibulbar dermoids, auricular appendages, and blind fistulae in the preauricular area, and his name has since become associated with this particular syndrome. The vertebral anomalies often found in Goldenhar's syndrome were not reported until later (Gorlin, Jue, Jacobsen, and Goldschmidt, 1963; Sugar, 1967; Berkman and Feingold, 1968) and the condition has now become known as oculo-auriculo-vertebral dysplasia. Two other conditions which may cause confusion are:

- (1) Oculo-vertebral dysplasia (Weyers and Thier, 1958), which is characterized by unilateral mandibulo-facial dysostosis, malformation of the globe, and developmental anomalies in the vertebral column;

(2) Cervico-oculo-acoustic dysplasia (Wildervanck, 1960), which has as its main features the Klippel-Feil syndrome, congenital ocular muscle anomalies, and deaf-mutism. These conditions are separate entities and can be differentiated from the syndrome in question.

The typical ocular findings in oculo-auriculo-vertebral dysplasia are epibulbar dermoids or dermolipomata. These are milky white or yellow in colour and soft in consistency and are usually situated at the limbus where they may encroach onto the cornea. Histological examination of these lesions in two cases showed a preponderance of adipose tissue. The characteristic auricular abnormalities are hypoplasia and deformity of the

Table *Additional anomalies in oculo-auriculo-vertebral dysplasia*

<i>Ocular</i>	<i>Facial</i>	<i>Oral</i>
Lid colobomata	Unilateral facial hypoplasia	Macrostomia
Ptoxis	Receding chin and mild malar hypoplasia giving a parrot-like appearance	Enlarged philtrum
Nasolacrimal obstruction	Nostril atresia	Micrognathia
Ophthalmoplegia	Displaced nasal septum	Aplasia of ramus and condyle of mandible
Squint	Fibrous band from angle of mouth to tragus	High-arched palate
Duane's retraction syndrome	Abnormal eyebrow	Bifid tongue
Exophthalmos	Mandibulofacial dysostosis	Bifid uvula
Corneal anaesthesia	Oculo-vertebral syndrome	Double lingual frenum
Corneal ulceration		Cleft palate
Persistent pupillary membrane		Malocclusion of teeth
Iris atrophy		Dental malformations
Polar cataract		
Uveal colobomata		
Microphthalmia		
Anophthalmos		
<i>Cranial</i>	<i>Musculo-Skeletal</i>	<i>Miscellaneous</i>
Biparietal foramina	Occipitalization of the atlas	Deaf-mutism
Open anterior fontanelle	Hemivertebrae in any position	<i>Cri-du-chat</i> syndrome
Accentuated frontal suture	Cuneiform vertebrae	Tetralogy of Fallot
Frontal bossing of skull	High vertebral bodies	Bundle branch block
Hypertelorism	Narrow disc spaces	Inguinal hernia
Hypoplasia of orbit	Complete or partial synostosis of two or more vertebrae	Mild mental defect
Low hair line	Supernumerary thoracic or lumbar vertebrae	Epilepsy
Petrous temporal anomalies	Narrow spinal canal	Umbilical hernia
Small pituitary fossa	Anomalous ribs	Hydronephrosis and hydroureter
	Hip abnormalities	Imperforate anus
	Spina bifida	Rectovaginal fistula
	Scoliosis	
	Lumbarization of S1	
	Sacral vertebrae aplasia	
	Sacral fovea	
	Limb and digit anomalies	
	Equino varus	
	Pes planus	

pinnae of the ears, blind or absent external auditory meati, bilateral preauricular skin appendages, and blind fistulae in the preauricular area. The commonest vertebral anomalies are cuneiform vertebrae, hemivertebrae, complete or partial synostosis of two or more vertebrae, and spina bifida.

The many associated anomalies, including our new findings, are summarized in the Table (Goldenhar, 1952; Paufigue, Etienne, and Moreau, 1952; Kühnel and Dominik, 1954; Vannas, 1955; Cordier, Dureux, and Barrucand, 1961; Wille-Jørgensen, 1962; Gorlin and Pindborg, 1964; Joannidis and Protonotarios, 1964; Manfredini and Manuelli, 1964; Smithells, 1964; Brancato and Artifoni, 1965; Sugar, 1966, 1967; Slem, 1967; Ladekarl, 1968; Mortada, 1969; Pieroni, 1969). The four cases described in this paper illustrate well the variety of signs which can appear in these patients. The abnormal radiological signs are particularly abundant and include features that have not been described previously. For instance, asymmetry of the petrous temporal bones has been noted by Darling, Feingold, and Berkman (1968), but by the employment of a polytomographic apparatus we have been able to add to this observation by demonstrating the underlying anomalies of the structures of the internal ear. These radiological features will be described in greater detail in another communication (Rees, Collum, and Bowen, 1971).

The particular ocular feature to which we wish to draw attention is corneal anaesthesia. In Cases 1 and 2 corneal sensation was completely absent and in Case 3 it was grossly reduced. In Case 1 there was an associated obstruction of the naso-lacrimal duct on one side and a corneal ulcer developed after the excision of a dermolipoma. This ulcer became so resistant to conservative treatment that a therapeutic penetrating corneal graft was necessary. Case 2 had extensive corneal scarring in one eye and a history of recurrent ulceration. Case 3 had reduced corneal sensation but no ulceration. Normally the excision of a limbal dermoid would be a straightforward procedure, although it might require a lamellar graft (Sugar, 1966; Heimann, 1968). The post-operative complications arising in Case 1, however, show that surgery in these patients should be undertaken with some caution. In Case 3, for instance, the limbal dermoid involves the patient's only eye and, although it is a cosmetic blemish, the risk involved contra-indicates surgery.

Case 4 presented with an ophthalmoplegia and ptosis, which we do not believe have been previously reported in association with oculo-auriculo-vertebral dysplasia. Both signs were said to have been present at birth and to have since remained static. This lack of progression and the absence of any other muscle weakness probably excludes a diagnosis of chronic progressive external ophthalmoplegia.

Summary

Four cases of oculo-auriculo-vertebral dysplasia are presented. New clinical and radiological features are described and the management of corneal complications is discussed.

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