MANDIBULO-FACIAL DYSOSTOSIS - THE EYE SIGNS OF A CASE STUDY

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ABSTRACT

The mandibulo-facial dysostosis syndrome (Treacher-Collins syndrome) was first described in 1889. It is a syndrome with multiple presentations, the classification for which was devised by Franceschetti and Zwahlen in 1944.

The eye signs are an important part of this syndrome. In addition to the main ocular features of colobomata of the lower eyelids and an anti-mongoloid slant, many other eye signs have been reported.

An 18-year-old Indian male was found to have features not previously described. These are high myopia, dermolipoma, lens subluxation and secondary glaucoma.

Keywords: Mandibulo-facial dysostosis, Treacher-Collins syndrome, high myopia, dermolipoma, lens subluxation, glaucoma.

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INTRODUCTION

The first reference to the comparatively rare congenital condition of mandibulo-facial dysostosis was published in 1889 by Berry[1]. It was however, the 2 further cases described and published by Treacher Collins in 1900[2] that led to the nomenclature of the Treacher Collins syndrome. Whereas Berry reported on the eye signs of the syndrome ie (a) colobomata of the lateral aspects of the lower lids and (b) an antimongoloid slant of the palpebral fissures, Treacher Collins went on to also emphasise another chief feature ie that of pronounced underdeveloped (hypoplastic) malar bones.

The nomenclature was further complicated after the published works of Franceschetti and co-workers in 1944 and 1949[3,4]. They produced extensive reports and devised a classification system for the syndrome. Out of their work, the term mandibulo-facial dysostosis was coined. However, because of the habit of medical historians for attaching names to a syndrome, further synonymous terms have been tagged to the condition: "Franceschetti-Klein", "Franceschetti-Zwahlen" or more comprehensively the "Franceschetti-Klein-Zwahlen" syndrome[3,4].

FEATURES AND CLASSIFICATION

The Treacher-Collins syndrome as initially described[2] entailed 2 main features:

a. the notching of the lower eyelids
b. underdevelopment of the malar bones

However, after the work of Franceschetti and Zwahlen (1944) a more complete classification was put forward which included many other features (described below). These included, amongst others, deformities of the mandible and ear. They also stressed that the barest minimal single clinical manifestation of the syndrome may be the mere presence of a slight anti-mongoloid slant of the palpebral fissure.

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Franceschetti's classification has 5 categories - complete, incomplete, unilateral, abortive and atypical [3-5].

a. The Complete Form

The complete fully developed syndrome consists of all or most of the following features:

(1) Anti-mongoloid slant of the palpebral fissure
(2) Colobomata or notching of the outer portion of the lower lids (the upper lid may sometimes be involved)

These eyelid features are frequently combined with deficient or absent eyelashes of the medial two-thirds or four-fifths of the lower eyelid.

(3) Hypoplasia of the facial bones especially the malar and mandible bones
(4) High palate, macrostomia and abnormal dentition with malocclusion of the teeth
(5) Abnormal growth of hair from the temporal area towards the cheek
(6) Malformation of external pinna (deformed, crumpled forward or misplaced)

This is sometimes associated with malformation of the middle and inner ears with frequent conduction deafness.

(7) Blind fistulae between the ears and the angles of the mouth. Occasional ear tags
(8) Other anomalies, skeletal deformities, facial clefts, large appearing nose with narrow nares

b. The Incomplete Form

In this category the deformity is less marked and less extensive. Deafness is often present even though the external ear may be normal. Franceschetti's classification includes in the incomplete form category those cases that are characterised solely by the antimongoloid oblique palpebral fissures, eyelid colobomata and hypoplasia of the malar bones (ie original cases reported by Treacher Collins).

c. The Abortive Form

In the abortive form ONLY the eyelid anomalies are present.

d. The Unilateral Form

In this form the abnormalities are confined to only one side of the face.

There is some debate among several authors that this form should rightly be excluded as the studies of Stark and Saunders[6] suggest that these unilateral cases can be classified either into (i) the first branchial arch syndrome or (ii) the first and second branchial arch syndrome. Whereas the mandibulo-facial dysostosis syndrome should be reserved for cases with bilateral abnormalities. The matter remains unresolved and will probably remain so till the exact aetiology of the syndrome is elucidated.

e. The Atypical Form

This form includes incomplete forms of the syndrome in which one or more of the principal characteristics of the complete, fully developed syndrome is missing, whereas other abnormalities which do not belong to the complete syndrome may be present.

Among the atypical features described are the following (Hurwitz in 1954):

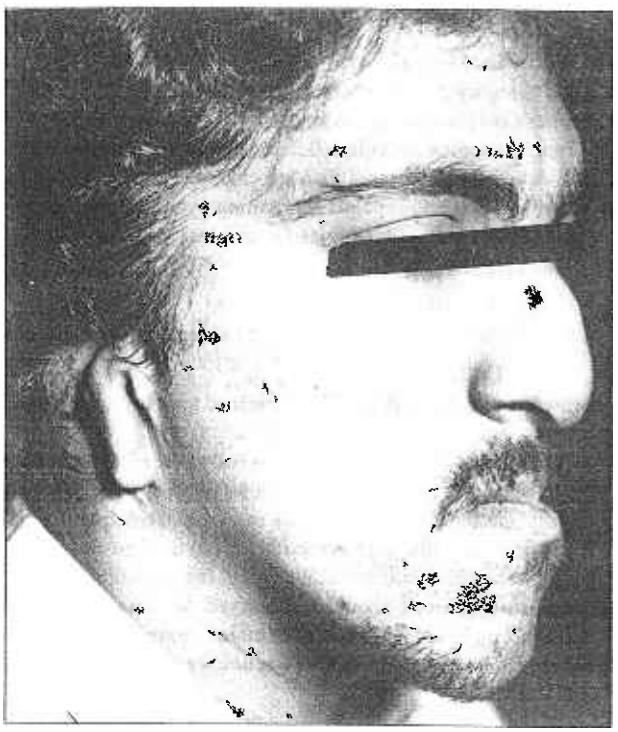
(1) Eye - microphthalmos, cataract, lacrimal canal atresia, double row of lower lid lashes, ectopiae pupillae, esotropias, orbital hyoplasia
(2) Oral - cleft lip, cleft palate, underdeveloped epiglottis, prognathism
(3) Nasal - partial atresia of the nasal fossae, absence of the frontal nasal angle, enlarged frontal sinuses, absence of frontal-nasal angle and rudimentary sphenoid sinuses
(4) Aural - Absent external auditory canal, absent mastoid cells
(5) Skull - hypoplasia of petrous bone, temporo-mandibular malarticulations, narrow sella turcica
(6) Others - agenesis of the frontalis muscle, club foot, synostosis of joints, agenesis of homolateral lung, congenital heart malformations

CASE REPORT

Mr AC, an 18-year-old Indian, was first diagnosed as having Treacher-Collins syndrome in early childhood. A brief summary of his congenital anomalies is as follows (Fig 1):

(1) Bilateral malar hypoplasia
(2) Cleft lip and cleft palate - repaired before first year
(3) Atresia of the right pinna (cosmetic surgery was done at age 15)
(4) Bilateral sensori-neural partial deafness
(5) Scaphocephaly
(6) Nocturnal enuresis (improved by age 13)
(7) Ventral septal defect (followed up by cardiologist till age of 17)
(8) Scoliosis of thoraco-lumbar spine
(9) Dental deformities

Fig 1 – Right profile – Malar hypoplasia and deformed pinna



Although Mr AC did not demonstrate any lower eye lid colobomata or absence of medial cilia, his other congenital anomalies support strongly the diagnosis of the Treacher-Collins syndrome, in the atypical category as classified by Franceschetti[3-5].

Of particular interest was Mr AC's ocular findings. An extensive literature search has not revealed any report of such similar ocular anomalies associated with the mandibulo-facial dysostosis syndrome.

What follows is a review of these ocular findings as were noted when Mr AC presented for a routine pre-military eye screening at the age of 18 years.

(1) Visual Acuity

Unaided (R) < 6/60 Aided (R) 6/12
(L) < 6/60 (L) 6/12

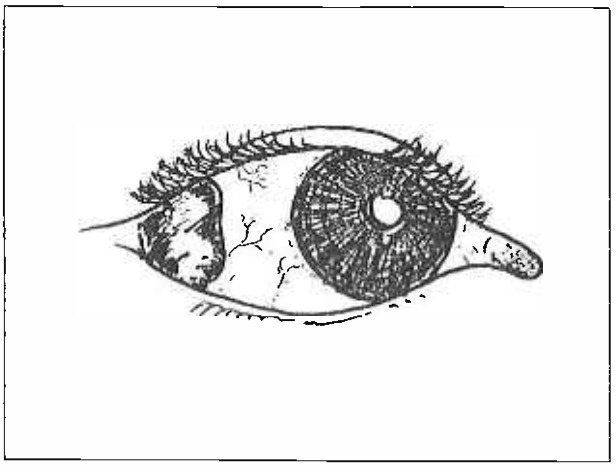
(2) Refraction

(R) -10.00/-1.75 x 90 —> 6/12
(L) - 11.75/-3.00 x 90 —> 6/12

(3) Anterior Segment Examination

A temporal dermolipoma was demonstrated in the right eye (Fig 2). This was made more evident on adduction of the eye.

Fig 2 – Line drawing of temporal dermolipoma



Bilateral shallowing of the anterior chamber was noted. This was assessed clinically by comparison with corneal thickness. The shallowing of the anterior chamber was more evident nasally in both eyes. Iridodonesis was evident.

On dilatation of the pupils, bilateral subluxation of lenses was found (Fig 3).

(4) Fundal Examination

On fundal examination, no retinal pathology was found. However, an enlarged cup-disc ratio was noted in both eyes. The CD ratio was 0.8 bilaterally and nasalisation of the blood vessels was present.

(5) Intraocular Pressure and Visual Fields

Because of the disc changes the intraocular pressures were taken. At the initial visit the readings were right 21 and left 21. Phasing was subsequently done and the results were as follows:

Time	Right	Left
0900	21	22
1000	18	20
1100	18	26
1200	14	14
1300	15	16
1400	14	14
1500	16	16
1600	16	16
1700	10	10

Fig 3 – Line drawing of left and right eye showing subluxation of the lens

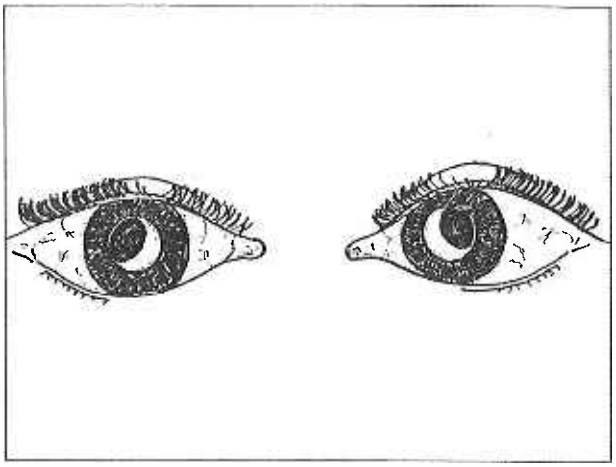
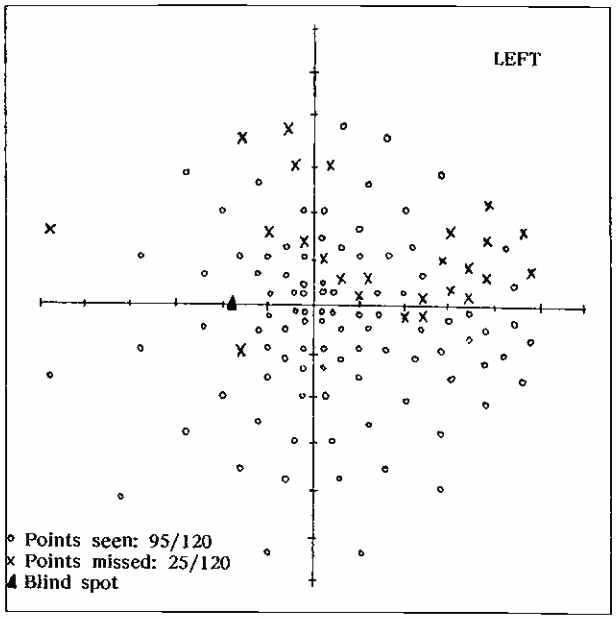


Fig 4 – Left visual field



Visual field plotting revealed a full field in the right eye. The visual field for the left eye however showed a Bjerrum's scotoma with nasal break through (glaucomatous changes) (Fig 4).

Gonioscopy revealed open angles in both eyes with marginal narrowing of the temporal quadrants (secondary to lens subluxation).

The optic disc changes, gonioscopy, visual field changes and morning rise in intraocular pressure are supportive of a diagnosis of secondary open angle glaucoma.

Genetics

The syndrome is transmitted as an irregularly dominant gene whose power of expression is variable and sometimes weak in its penetrance. This leads to the multiple combinations of possible presentations and also to the difficulty in classification as highlighted above. Further to this over 50% of the cases are due to new mutations.

An attempt to study the family history of Mr AC was hindered by the reluctance of his relatives to present for examination. However, through interviewing Mr AC and his father, 2 maternal great uncles were said to have poor hearing.

Mr AC's elder brother also has a history of bilateral ear operations. All relatives were however apparently normal in physical appearance. Hence no further comment can be made on the significance of the family history of "hearing problems".

DISCUSSION

Berry, Treacher-Collins, Franceschetti-Zwahlen, Franceschetti-Klein syndromes, are all representative of the varied presentations of a more comprehensive hereditary syndrome more suitably termed mandibulo-facial dysostosis.

In addition to the main ocular findings of lower eyelid colobomata and an antimongoloid slant of the palpebral fissure, many other ocular anomalies have been described in the present literature[3 5,7-14]. Mr AC however displays additional eye findings as of yet not reported.

These include:

(1) Dermolipoma
(2) Subluxation of lens
(3) Secondary open angle glaucoma
(4) High myopia

Besides being additional ocular anomalies associated with this syndrome, they are especially important in that they warrant referral to an ophthalmologist for further assessment and treatment. This contrast with the other ocular findings which are essentially anomalies which do not progress with time nor avail themselves to any form of treatment.

High myopia however requires assessment to ensure proper refraction is done. This is especially important in youth to prevent amblyopia. Retinal degenerations, which are found more commonly in high myopia, must also be actively sought and treated lest they progress to retinal detachments.

Secondary open angle glaucoma also requires close monitoring and treatment to ensure that intraocular pressure is maintained at reasonable levels. This will prevent the field change losses of glaucoma. Initially treatment should be with topical eyedrops. If intraocular pressure still cannot be controlled a filtration procedure may have to be considered.

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