

# MANDIBULO-FACIAL DYSOSTOSIS

## A Case Report

F.J. DAMATO  
M.D., D.O., D.O.M.S., F.R.C.S.

A. GRISCTI SOLER  
M.D.

Though the Mandibulo-Facial Dysostosis is a relatively rare congenital abnormality we thought this case deserved reporting because of the tender age of the patient — five months. Most reported cases were in considerably older patients. Moreover this is presumably the first case to be described in Malta. The syndrome is also known as the Treacher Collins or Franceschetti's Syndrome though it was first mentioned by Allen Thomson in 1847 and later by Berry in 1889. The genetic homogeneity of the syndrome was established by Franceschetti and Klein in 1949.

The fully developed syndrome shows the following features:-

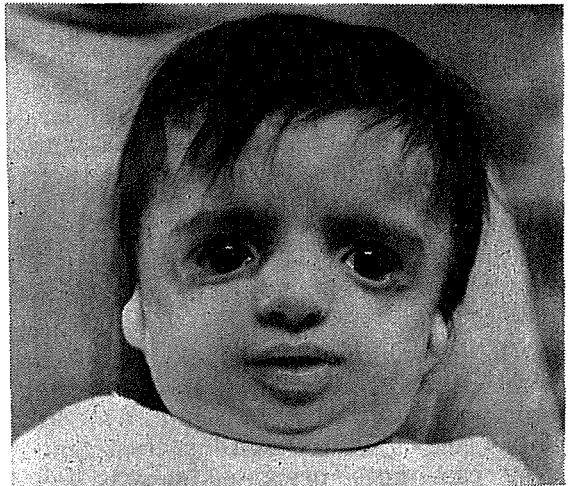
1. An antimongoloid obliquity of the palpebral fissures, with a coloboma in the outer parts of the lower lids and sometimes of the upper.
2. Hypoplasia of the facial bones particularly of the malar bones and of the mandible.
3. Malformation of the external ears and sometimes of the middle and inner ears.
4. Macrostomia, a high arched palate and an abnormal dentition.
5. Blind fistulae between the angles of the mouth and the ears.
6. Tongue shaped projection of the hairline on the cheek.
7. Associated anomalies such a facial clefts and skeletal deformities.

Atypical, incomplete and unilateral forms have been described.

Our case presents all the main features except those that one would not expect to find at such an early age, such as dental abnormalities, tongue shaped projection of

the hairline on to the cheek, etc. The general appearance is of a fishlike face with sloping palpebral fissures, (antimongoloid obliquity of the palpebral fissures), flattened cheek bones, a large mouth (Macrostomia), a receding chin (Micrognathia) and absence of the Fronto-Nasal Angle. A striking feature is the deformity of both ears.

There is atresia of the pinna except for the tragus and a fold of skin extending upwards. There is also atresia of the external auditory meati. Hearing seems to be present on both sides but the child is too young for full auditory testing. The palate is highly arched and there is a bifid uvula. X-ray of the skull shows a small mandible with



**Fig. 1: Sloping anti-mongoloid palpebral fissures. Colobomata at the outer parts of the lower eyelids with bilateral ectropion. Macrostomia, flattened cheek bones and the malformed pinnae can also be seen.**



**Fig. 2: Showing absence of fronto nasal angle, micrognathia, atresia of the pinna of the auricle, and the typical fish-like physiognomy.**

small rami and with especially small condylar processes. The inner ear appears normal with normal horizontal semilunar canals. X-rays of the left wrist showed some flaring of the lower end of the ulna. Lateral views of the lumbar spine show possibly some elongation of the pedicles of the lumbar vertebrae. The child seems otherwise normal. It is interesting to note that the only abnormality in the child's facies noticed by the parents was the abnormal shape of the ears.

The syndrome is described as having clearly established hereditary characters. It is inherited as an autosomal dominant trait with variable penetrance. This variability in penetrance of the gene is shown by

the fact that the features of the parents of this child do not show any abnormalities.

The mandibulo-facial dysostosis is caused by a retardation of the differentiation of the maxillary mesoderm derived from the first visceral arch. There is a defective ossification of the bones of the face derived from the visceral mesoderm. An inhibiting process seems to become effective about the seventh week of foetal life when the supporting bony structures of the face are being laid down. There would follow secondarily as a result of the delayed ossification of the walls of the orbit, particularly in the temporal region, a displacement of the bones, and because support is missing, a hypoplasia and malposition of the associated soft tissues. Moreover an arrest of development at this stage accounts for the micrognathia, the macrostomia and the downward displacement of the entire ear.

As regards treatment it has been suggested that the malformation of the lid could be remedied by a plastic operation meant to raise the level of the outer canthus by transferring skin from the upper to the lower lid. At the same time the deficient malar region could be filled up with cartilage implants.

**Summary: A case of the mandibulo-facial dysostosis at the age of 5 months is reported. All the main features described in the syndrome are present in this case.**

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