Corneal Endothelial Polymegethism: A Neural Crest Defect

Paolo Nucci

Department of Ophthalmology, University of Milan, Scientific Institute San Raffaele Hospital, Milano.

Mandibulofacial dysostosis\(^1\) is characterized by marked malar and mandibular hypoplasia, antimongoloid slant of the palpebral fissures, lower eyelid colobomas, and auricular malformations. Cleft palate and conductive deafness are also present in this condition.

Mandibulofacial dysostosis is reported to have an autosomal dominant transmission with 60% of the cases representing new mutations. In 1975 Johnston\(^2\) proposed that the syndrome is caused by a defect in neural crest cell migration. In a previous paper we reported the occurrence of cornea guttata in two patients affected by mandibulofacial dysostosis, confirming a common embryologic origin of these two conditions.\(^3\)

Recently we had the opportunity to examine a 19-year-old boy with mandibulofacial dysostosis at the Pediatric Ophthalmology Unit of San Raffaele Hospital in Milan.

He had never worn contact lenses nor suffered from ocular diseases or trauma.

The routine examination did not show any abnormalities but the lower eyelid coloboma. The extension of the coloboma was not severe enough to expose the cornea. His corrected visual acuity was 6/6 (20/20). Specular microscopy of the corneal endothelium showed bilateral polymegethism. (Fig. 1) The coefficient of variation was 42%, whereas at this age it rarely exceeds 22%.

\(^{1}\) Mandibulofacial dysostosis
\(^{2}\) Johnston
\(^{3}\) Previously

---

Received November 8, 1989; revision received January 30, 1990

* Physician
The endothelial cell count was within normal limits (2550 SD: 88). This finding appears to be in contrast with the belief that polymegathism is a generalized response to endothelial stress or related to the decrease in oxygen tension at the cornea. Our hypothesis is that polymegathism can be related to anomalies of neural crest-derived cells and can be detected in patients suffering from neural crest anomalies such as mandibulofacial dysostosis. In these patients a careful examination of the corneal endothelium is mandatory before prescribing extended wear contact lenses, since investigators have reported that a correlation exists between the degree of polymegathism and a reduction of endothelial function.

REFERENCES


AUTHOR'S ADDRESS:
Paolo Nucci
Department of Ophthalmology
University of Milan
Scientific Institute San Raffaele Hospital
via Olgettina 60
Milano 20132
Italy