

Corneal Endothelial Polymegethism: A Neural Crest Defect

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Mandibulofacial dysostosis¹ is characterized by marked malar and mandibular hypoplasia, anti-mongoloid 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 Johnston² 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%.

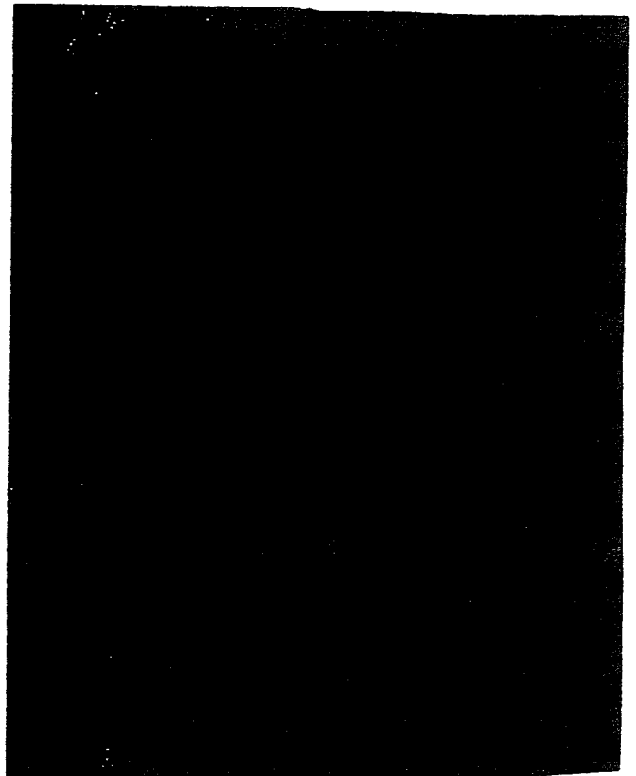


Figure 1. Polymegethism in a patient with mandibulofacial dysostosis.

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The endothelial cell count was within normal limits (2550 SD: 88). This finding appears to be in contrast with the belief that polymegethism is a generalized response to endothelial stress⁴ or related to the decrease in oxygen tension at the cornea.⁵ Our hypothesis is that polymegethism 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 polymegethism and a reduction of endothelial function.⁶

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