

## Keratoconus and Congenital Hip Dysplasia

Paolo Nucci, M.D.,  
and Rosario Brancato, M.D.

Clinica Oculistica dell'Università di Milano, Istituto Scientifico H. S. Raffaele.

*Inquiries to Paolo Nucci, M.D., Clinica Oculistica dell'Università di Milano, Ospedale S. Raffaele, via Olgettina 60, 20132 Milano, Italy.*

Congenital hip dysplasia denotes a delayed or defective development of the hip joint and its associated structure. If untreated, the condition heals, persists as a subluxation, or progresses to a frank dislocation. Genetic, ethnic, and uterine factors play a role in its pathogenesis. Congenital hip dysplasia occurs once in every 110 births, with a female sex predilection of approximately 5 to 1. A family history of the condition is noted in 12% to 33% of patients.<sup>1</sup>

Two sisters, aged 9 and 12 years, were examined by one of us (P.N.) for blurred vision and photophobia. They were the only children of healthy, consanguineous parents whose mothers were sisters.

Both girls were delivered vaginally at term and had Apgar scores of 9/9. Both girls had bilateral congenital hip dysplasia, related to an acetabular dysplasia, diagnosed by ultrasonography at age 6 months in the older sister and at age 3 months in the younger sister. There was no family history of hip dysplasia.

The younger sister had best-corrected visual acuity of 20/20 in each eye with refraction of R.E.: +1.00 +1.25 × 30 and L.E.: -1.00 -1.25 × 15. The older sister had best-corrected visual acuity of R.E.: 20/40 and L.E.: 20/30 with refraction of R.E.: -2.00 -3.25 × 40 and L.E.: -1.75 -2.75 × 35. Keratometric readings disclosed distorted mires in each girl. Biomicroscopic examination disclosed a cone-shaped ectasia of the cornea in each girl. Vogt's striae were noted in the deep stroma and Descemet's membrane of the older girl. The endothelial cell count was within normal limits (2,890 ± 121 cells/mm<sup>2</sup> in the younger sister and 2,914 ± 321 cells/mm<sup>2</sup> in the older sister).<sup>2</sup> The appearance of the corneas was consistent with bilateral keratoconus. No evidence of other corneal dystrophies was noted. The ophthalmic history was unremarkable for atopic diseases, ocular trauma, eye rubbing, or hard contact lens use.

Keratoconus usually becomes manifest during adolescence. Most cases of keratoconus are sporadic, but it is sometimes familial. Kennedy, Bourne, and Dyer<sup>3</sup> described keratoconus occurring in less than 6% of the blood relatives of an affected proband. Greenfield and associates<sup>4</sup> reported a familial occurrence of keratoconus and osteogenesis imperfecta and hypothesized an autosomal recessive pattern of inheritance. Several systemic tissue disorders are reported in association with keratoconus, including Marfan's syndrome and Ehlers-Danlos syndrome.<sup>3</sup> Reports of consanguinity have been cited as evidence of a recessive mode of inheritance. Wynne-Davies<sup>5</sup> suggested a multiple gene heredity defect in congenital hip dysplasia with acetabular dysplasia, especially when environmental factors had not played a role.

The incidence of keratoconus ranges from 50 to 200 per 100,000 live births, whereas congenital hip dysplasia occurs once in 110 live births. Therefore, congenital hip dysplasia and keratoconus is an association of two independent events whose incidence ranges from 4 to 18 per 1,000,000 live births. Mathematically, the probability that this association occurs twice by chance is extremely low. The early development of keratoconus and the consanguinity of the parents support the hypothesis that this is most likely an autosomal recessive association.

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### References

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