Dysplasia epiphysialis, with Ocular Anomalies

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A 36-week premature boy, birth weight 2,880 g, is demonstrated by means of slides. The child shows all the clinical and radiological characteristics of dysplasia epiphysialis punctata.

A better-known synonym for this condition is chondrodystrophia cal-cificans congenita. 112 cases have been described, in 30% of which eye anomalies have been found: cataract and atrophy of the optic nerve. The prognosis is particularly bad when the bone anomalies are combined with cataract; these children do not usually live longer than about two years.

In our case, there was bilateral cataract and indications of raised intraocular pressure. By using pilocarpine, it was possible to see the bilateral embryotoxon posterior. The child died of pneumonia a few months after birth. Histological examination of the eye revealed, in addition to the cataract, specific anomalies of the chamber angle: a very well-developed ring of Schwalbe; a broad adhesion arising from the area adjacent to Schwalbe's ring, spreading out to the atrophic iris; and a well-developed trabecular network at the chamber angle. These findings together form the well-known syndrome of Rieger.

This combination, dysplasia epiphysialis punctata with cataract and Rieger's syndrome, has not been described before. It is difficult to say whether this is a primary or secondary Rieger, as the differentiation is not clear to me.

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Discussion

Van Balen: Why do you use this unusual name for this condition?

Miss Hammond (in reply): When studying the bony anomalies

I used Rubin's textbook

'Dynamic Classification of Bone Dysplasias'. It is a well-known orthopaedic book that is of practical use

thanks to its simple and convenient classification.