Correspondence

This letter was shown to Drs Baraitser and Patton, who reply as follows.

We would like to thank Drs Corona and Cantú for drawing our attention to their article.1 We did not include the condition on the London Dysmorphology Database as a separate condition because we thought that the authors were describing Noonan syndrome. The similarities between their report and ours are obvious and they probably refer to the same condition. Whether this is distinct from Noonan syndrome is still not clear. The recently published CFC syndrome by Reynolds et al2 could also be the same condition and it is uncertain whether it too is distinct from Noonan syndrome. We have decided for the time being to keep the CFC syndrome and the condition described by Cantú et al and ourselves separate, rather than lump them together in anticipation of the time when we have a definite test for Noonan syndrome.

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References


Mental retardation with blepharophimosis

Sir,

We have read with interest the article by Ohdo et al1 presenting two sisters and their cousin with mental retardation, blepharophimosis, blepharoptosis, and hypoplastic teeth. We recently studied a similarly affected three year old Caucasian male (figure), the only child born to a young, unrelated couple. The pregnancy was normal and free of exposure to known teratogens. Our patient, in addition, had myopia and horizontal nystagmus, a depressed nasal bridge, mid-facial hypoplasia, small, low set, round ears, and a small oral cavity. Other findings included laterally displaced, inverted nipples, hypoplastic scrotum with undescended testes, hyperextensible joints, clinodactyly of the fifth fingers bilaterally, and hypoplastic thenar eminences. There was generalised muscular hypotonia and the right patella was dislocated. No heart murmurs were heard. Psychometric evaluation using the Bayley Scales of Infant Development showed a developmental lag of over 50% in all areas. There was no-one on either side of the family with any form of birth defect.

We believe this is a distinct entity and should be considered among the differential diagnoses in patients with mental retardation, blepharophimosis, and dental abnormalities. Congenital heart disease may or may not be a frequently encountered component of this newly described entity.

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Reference