Correspondence

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Hirschsprung's disease, distinctive facies, and microcephaly

SIR,

I read with interest the report by Hurst *et al*[1] in the July issue of the Journal of three children with Hirschsprung's disease, distinctive facies, and microcephaly. The two sibs and an unrelated child were matched by the London Dysmorphology Database and presented as a new syndrome. There was no discussion of similar case reports.

The POSSUM syndrome information system[2] lists 10 syndromes with Hirschsprung's disease.[3–11] Eight of the syndromes could be quickly dismissed on the basis of associated findings. The sibs reported by Goldberg and Shprintzen[3] had the same facial appearance and common features. I believe that this is the same syndrome. Goldberg and Shprintzen[3] had also proposed this to be of recessive inheritance and the result of an error in neural crest development. Although the case of Brunoni *et al*[5] shares some of the same features it is likely to be a different condition. The features of the five cases from Goldberg and Hurst are listed in the table together with the features of the case reported by Brunoni.

All five patients had Hirschsprung's disease, a distinctive facies, mental retardation, microcephaly, and short stature. The facies was characterised by dense eyebrows, apparent hypertelorism, large nose with broad nasal root, relative prognathism, and anteverted, 'bat' ears. Similar brain CT scan abnormalities were present in four of the patients. Iris colobomata are reported in only one of the sibs and the third patient reported by Hurst *et al*. Submucous cleft palate was seen in the sibs of Goldberg's report. It would be desirable to review Goldberg's patients for ocular colobomata and Hurst's patients for submucous cleft palate. Goldberg's patients had a spastic gait and neonatal hypotonia; the third case in Hurst's report had spasticity.

Although the karyotypes in the three patients tested were reported to be normal, it is of interest that a similar pattern of features, including Hirschsprung's disease, developmental delay, and coloboma of the iris, was reported in a patient with de novo interstitial deletion of 2p22 and reciprocal

TABLE *Phenotypic features of Hirschsprung's disease, distinctive facies, and microcephaly.*

	Goldberg and Shprintzen[3]		*Hurst et al*[1]			*Brunoni et al*[5]
	Case 1	*Case 2 (Sibs)*	*Case 1*	*Case 2*	*Case 3*	
Hirschsprung's disease	+	+	+	+	+	+
Mental retardation	+	+	+	+	+	+
Short stature	+	+	−	+	?	?
Microcephaly	+	+	+	+	+	+
Cleft lip	−	−	−	−	−	+
Cleft palate (submucous)	+	+	−	−	−	+
Dense eyebrows	+	+	+	+	+	−
Curled eyelashes	+	+	?	?	?	−
Hypertelorism	+	+	?+	?−	+	+
Iris coloboma	−	−	+	−	+	−
Big/'bulbous' nose	+	+	+	+	−	−
Broad nasal root	+	+	+	+	+	−
Sparse scalp hair	+	+	−	−	+	−
Prognathism	+	+	−	+	+	+
Anteverted ears	+	−	+	+	+	−
Scoliosis	+	−	−	−	−	−
Inguinal hernia	+	−	−	−	−	−
Cryptorchidism	+	−	−	−	−	−
Neonatal hypotonia	+	+	?	?	−	−
Abnormal gait	+	+	?−	−	−	−
CT ventricular dilatation	+	+	−	+	−	−
'Cerebral dysgenesis'	+	+	+	+	−	−
Autosomal recessive	+	+	+	+	+	?
Karyotype	N	?	N	?	N	

translocation (3;7)(p21;q22).[12] Review of karyotypes with high resolution banding may be of value.

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