Dysmorphology report

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Limb anomalies in the CHARGE association

SUMMARY We report a male infant with iris coloboma, choanal atresia, postnatal retardation of growth and psychomotor development, genital anomaly, ear anomaly, and anal atresia. In addition, there was cutaneous syndactyly and nail hypoplasia of the second and third fingers on the right and hypoplasia of the left second finger nail. Comparable observations have rarely been reported and possibly represent genetic heterogeneity.

History

Prenatal. Premature contractions at 35 weeks of gestation, tocolytic treatment. Spontaneous delivery at 36 weeks. The newborn infant required resuscitation and was referred to the children's hospital.

Family. Second child of healthy unrelated parents. Father aged 32 years, mother aged 20 years. Healthy two year old sister.

Clinical examination

Birth weight 2370 g (between 25th and 50th centile), length 49 cm (97th centile), and head circumference 31-5 cm (25th centile). Right iris coloboma, anteverted nostrils, long philtrum, moderate retrognathia, and low set, posteriorly rotated ears with preauricular pits bilaterally (fig 1). Choanal atresia bilaterally and narrow palate resulting from broad alveolar ridges. Cutaneous syndactyly of the right second and third fingers and nail hypoplasia of the left index finger (fig 2). Pes adductus and sandal gap bilaterally. Anal atresia with rectourethral fistula. Shawl scrotum and shortened foreskin (fig 3).

Normal G banded karyotype on cultured peripheral lymphocytes.

Medical history

Artificial anus on the second day of life, 10 days later surgical correction of the choanal atresia. Throughout infancy severe diarrhoea with tendency towards hyponatraemic dehydration. Predominantly parenteral feeding with subsequent growth retardation and dystrophy. Psychomotor development severely retarded. Death at 11 months owing to protracted cardiac failure. Length 67 cm (-3.5 SD), weight 6480 g (10th centile for length), and head circumference 41.5 cm (-3 SD).

Necropsy. No internal anomalies, particularly no histomorphological explanation for the severe, prolonged diarrhoea. Neuropathological studies normal.

Discussion

We have assigned this characteristic pattern of multiple congenital anomalies to the CHARGE association. Anal atresia such as was present in our patient may be part of this association.1 Limb anomalies apparently are an unusual feature of the CHARGE association. Whereas slight anomalies, such as sandal gap, mild syndactyly, clinodactyly of the fifth fingers, or hypoplastic nails were seen in some patients,1-4 syndactyly as in our patient has only been reported so far by Siebert et al.5

Our observation once more poses the question as to whether this is simply a rare manifestation of an associa-
Dysmorphology report

Figure 2: Note cutaneous syndactyly and nail hypoplasia of the right second and third fingers and nail hypoplasia of the left index finger.

Figure 3: Note mild genital anomaly (shawl scrotum and shortened foreskin).

The association usually occurs sporadically and accordingly has a low recurrence risk,1,4 it might be shown that there is a higher recurrence risk in some cases.

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References


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