Dysmorphology report

Journal of Medical Genetics 1989, **26**, 202–203

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-

FIG 1 Facial view of the patient with the CHARGE association.

Received for publication 8 June 1988.
Accepted for publication 11 July 1988.

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

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

tion which has not yet been adequately defined or whether it is characteristic of a distinct entity within the non-specific CHARGE association. As we have learned in the case of the VATER association, which shows some overlap with the CHARGE association, distinct syndromes may possibly be delimited in the future from the probably heterogeneous CHARGE association. This would be of particular importance for practical questions such as genetic counselling. Whereas we still assume at present that the CHARGE

association usually occurs sporadically and accordingly has a low recurrence risk,^{1 4 6} it might be shown that there is a higher recurrence risk in some cases.

P MEINECKE*, ANKE POLKE*, AND P SCHMIEGELOW†
 *Altonaer Kinderkrankenhaus, Hamburg; and
 †Pathologisches Institut, Klinikum Stadt Karlsruhe,
 West Germany.

References

- ¹ Koletzko B, Majewski F. Congenital anomalies in patients with choanal atresia: CHARGE association. *Eur J Pediatr* 1984;**142**: 271-5.
- ² Rabaeus M, DeLozier C, Klauser P. Examen diagnostique et conseil génétique dans certains cas d'atrésie choanale avec malformations multiples (association "CHARGE"). *J Genet Hum* 1983;**31**:67-72.
- ³ Davenport SLH, Hefner MA, Mitchell JA. The spectrum of clinical features in CHARGE syndrome. *Clin Genet* 1986;**29**: 298-310.
- ⁴ Oley CA, Baraitser M, Grant DB. A reappraisal of the CHARGE association. *J Med Genet* 1988;**25**:147-56.
- ⁵ Siebert JR, Graham JM Jr, MacDonald C. Pathologic features of the CHARGE association: support for involvement of the neural crest. *Teratology* 1985;**31**:331-6.
- ⁶ Pagon RA, Graham JM, Zonana J, Yong SL. Coloboma, congenital heart disease and choanal atresia with multiple anomalies: CHARGE association. *J Pediatr* 1981;**99**:223-7.

Correspondence to Dr Peter Meinecke, Abteilung Medizinische Genetik, Altonaer Kinderkrankenhaus, Bleickenallee 38, D-2000 Hamburg 50, West Germany.