Correspondence

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Holoprosencephaly and postaxial polydactyly: another observation

SIR,

In the November 1987 issue of your journal, Young and Madders[1] reported the hitherto unreported observation of the association of holoprosencephaly and postaxial polydactyly in a stillborn male infant. In addition to the holoprosencephaly sequence with premaxillary agenesis, bilateral microphthalmos, alobar holoprosencephaly, and bilateral upper limb postaxial polydactyly, this first child of healthy, unrelated parents also had ventricular and septal heart defects and a small penis with cryptorchidism. We have observed a similar association in a stillborn female fetus, the second born child of healthy, unrelated parents.

The prenatal history was unremarkable. Ultrasonographic diagnosis of holoprosencephaly was made at 31 weeks' gestation. Amniocentesis showed an alfafetoprotein level of 1·3 μg/ml and a 46,XX normal female karyotype (confirmed on postnatal skin fibroblast culture). Induction of labour was performed at 31½ weeks.

The family history is negative. The index patient is the second child of healthy, unrelated parents, the father aged 57 years and the mother 34 years. The first child, a boy, is normal. Necropsy (fig 1) showed ocular hypotelorism, suborbital proboscis (cebocephaly), frontal hirsutism, and no cleft palate. There was bilateral postaxial upper limb polydactyly. The following internal abnormalities were also found at necropsy (fig 2): alobar holoprosencephaly with hydrocephalus, neocerebellar hypoplasia, absence of the anterior lobe of the pituitary with secondary hypoplasia of the thyroid and surrenal cortices, and microphthalmia with retinal dysplasia. No abnormalities were noted in the lungs, heart,

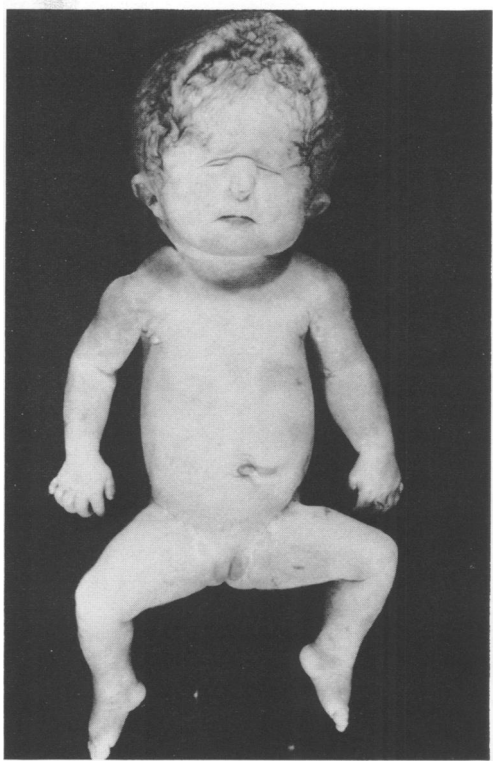

FIG 1 *The proband at necropsy.*

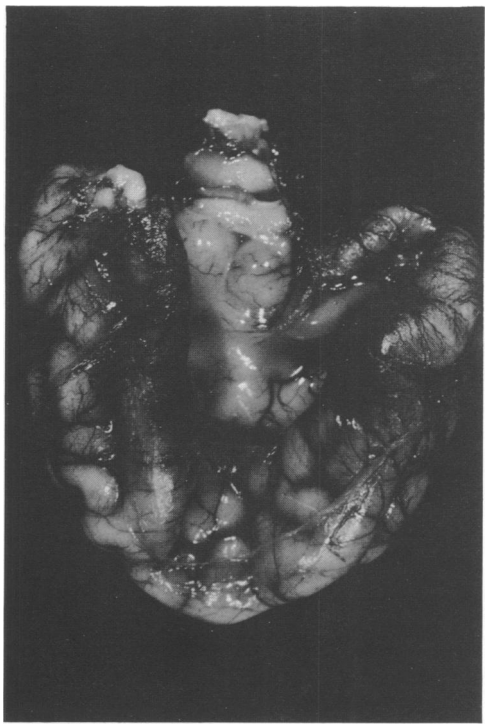

FIG 2 *The brain at necropsy.*

oesophagus, intestinal tract, liver, pancreas, spleen, or kidneys.

The present patient is another example of non-chromosomal association of holoprosencephaly and postaxial hexadactyly limited to the hands. It is interesting to note that in the patient of Young and Madders[1] and in the present case the postaxial polydactyly is limited to the hands. In trisomy 13 a holoprosencephaly sequence may be present and is associated with postaxial hexadactyly of the hands and feet. As discussed by Young and Madders, the association of holoprosencephaly and postaxial polydactyly of the hands does not fit into any hitherto delineated MCA syndrome.

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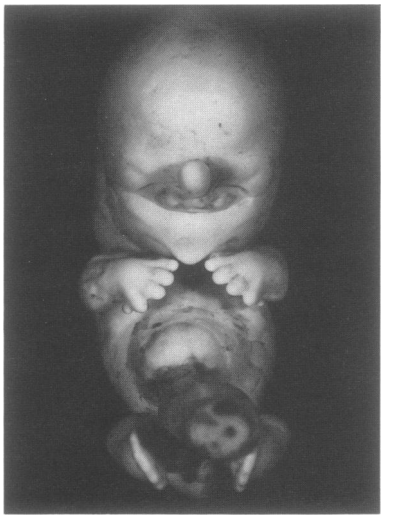

FIG 1 *AP view of the embryo. Note the proboscis and closely set eyes.*

Reference

[1] Young ID, Madders DJ. Unknown syndrome: holoprosencephaly, congenital heart defects, and polydactyly. *J Med Gen* 1987;**24**:714–5.

Holoprosencephaly, ventricular septal defect, and postaxial polydactyly in a human embryo

SIR,

In the November 1987 issue of the Journal, Young and Madders[1] reported a stillborn male infant with holoprosencephaly, cardiac anomalies, and postaxial polydactyly. This is a new dysmorphic syndrome which has not been fitted into any established clinical entity.

We report here a six week male embryo with a similar syndrome. The embryo was studied after induced abortion on a healthy 25 year old woman for socioeconomic reasons (Eugenics Protection Law of Japan). Her husband was 31 years old and the couple was non-consanguineous. Their family history was unremarkable. They had one normal child and one previous induced abortion. The mother had regular menstrual cycles and her pregnancy was uneventful. She took no alcohol, cigarettes, or medication during pregnancy. The pregnancy was terminated on the 45th day after estimated ovulation by dilatation and curettage.

The embryo was at Carnegie stage 20[2] and its crown-rump length was 18·8 mm. Externally, ethmocephaly with a proboscis and closely set eyes

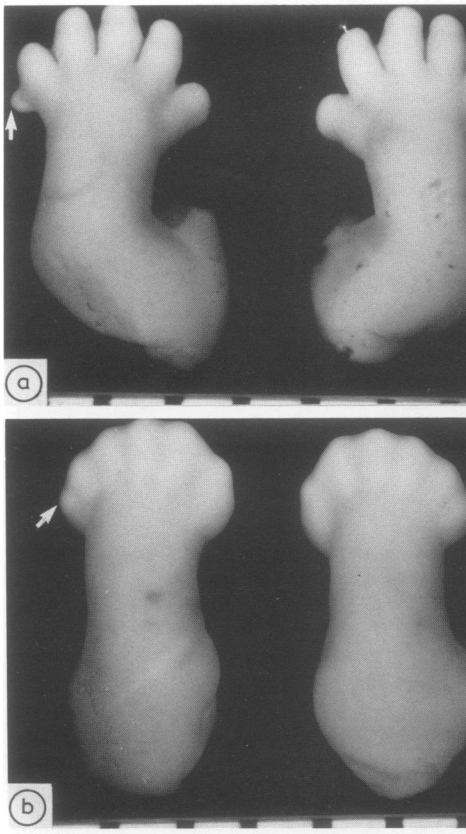

FIG 2 *Upper (a) and lower (b) limbs showing supernumerary digits on the postaxial side (arrows).*