**Dysmorphology report**

**Orofaciodigital syndrome type IV (Mohr-Majewski syndrome) with severe expression expanding the known spectrum of anomalies**

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**Abstract**
We present a male infant with hypertelorism, median pseudo-cleft of the upper lip and cleft palate, lobulated tongue, hypoplastic larynx and epiglottis, mesomelic shortening of limbs with particularly short and broad tibiae, polydactyly of the upper limbs, severely hypoplastic external genitalia with anorchidism, anal atresia, severe congenital heart defect, and renal agenesis. These features show considerable overlap with severe Majewski type short rib-polydactyly syndrome and so expand the known spectrum of anomalies in orofaciodigital syndrome type IV.

**History**
_Prenatal._ Oligohydramnios and intrauterine growth retardation noted in the third trimester of pregnancy. Caesarean section at 36 weeks’ gestation. Severely cyanotic and asphyxiated male infant died shortly after birth.

_Family._ First child of young, healthy, and unrelated parents.

**Clinical examination**
_**At birth.**_ Weight 1620 g (−3 SD), length 37 cm (−5 SD). Head circumference not recorded. Abundant lanugo hair on frontal region, lateral face, and back. Microcephalic skull, hypertelorism, short, broad nose, median pseudo-cleft of upper lip, small and dysmorphic ears (fig 1). Small, lobulated tongue tethered to the lower gum. Cleft palate. Severe choanal stenosis. Short neck. Severe hypoplasia of penis and scrotum, cryptorchidism. Anal atresia. Mesomelic shortening of arms and legs. Short, broad hands with postaxial hexadactyly (six nails), severe brachydactyly, and syndactyly. Owing to severe shortness and syndactyly of toes with severe hypoplasia of the nails, exact number not determined. Karyotype 46,XY.

**Radiological findings (fig 2a,b,c)**
Small facial bones with ocular hypotelorism and small mandibles. Narrow upper thorax, but no significant shortening of ribs. Hypoplastic iliac wings. Mesomelic shortening of limbs.

![Figure 1](http://jmg.bmj.com/)

_Figure 1_ Newborn male with orofaciodigital syndrome type IV. Note median pseudo-cleft of upper lip and lobulated tongue.
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Figure 2a  Note narrow upper thorax but no shortening of ribs.

Figure 2b  Note mesomelic shortening of upper limb.

Figure 2c  Note mesomelic shortening with very short and broad tibiae.

mesomelic and acromelic shortening of upper and lower limbs with moderate bowing of the ulnae and severely shortened and broad tibiae. Hands: only four well ossified metacarpals and extremely short fingers with syndactyly bilaterally. Feet: only four metatarsals ossified bilaterally and extremely short toes with syndactyly without any ossified centres.

Necropsy

Discussion
The orofacial findings described in combination with mesomelic shortening, predominantly of the lower limbs, and polydactyly are typical features of the orofaciodigital (OFD) syndrome type IV.

This probably autosomal recessive disorder was first described by Temtamy and McKusick1 followed by two sibs reported by Baraitser et al2 and Burn et al.3 Most recently, a fifth case from Ireland was published by Nevin and Thomas.4 Two cases reported by Silengo et al5 showed a transitional type between
the Mohr and Majewski syndromes; however, these patients lacked the short tibiae that are a characteristic feature of OFD syndrome type IV. Finally, an interesting family with an X linked recessive condition resembling OFD type II but with tibial shortening was observed by Edwards et al.6

Compared with the five cases reviewed by Nevin and Thomas,4 our patient shows an unusually severe expression of this syndrome with regard to internal malformations. Apart from the severe congenital heart malformation, which was also seen in case 1 of Temtamy and McKusick1 and in the patient of Nevin and Thomas, our case showed absent adrenals, renal agenesis with absent ureters and bladder, anorchidism, and anal atresia. This observation expands our current knowledge of the clinical spectrum of OFD type IV and proves that there may be a considerable overlap with severe Majewski type short rib-polydactyly syndrome.

Our case lends further support to the hypothesis1,2 that OFD IV represents a genetic compound for the Majewski syndrome and the Mohr syndrome (OFD II) genes. However, genetic heterogeneity within this group of disorders has to be taken into consideration since the crucial observation by Edwards et al.6

An alternative but less likely explanation would be that there is a distinct OFD syndrome, the Majewski syndrome and the Mohr syndrome representing the extreme ends of its very broad clinical spectrum.

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