

Syndrome of the month

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The orofacioidigital (OFD) syndromes

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There are now four separate orofacioidigital (OFD) syndromes, designated as I to IV. Unfortunately, there is considerable overlap in the features and precise clinical identification is still difficult.

Type I

The original description was by Papillon-Leage and Psaume in 1954.¹

FACIAL FEATURES (COMPARED WITH TYPE II)

	I	II
Tongue nodules	+	+
Bifid tongue	+	+
Midline lip cleft	+	+
Cleft palate	+	+
Frenulae hypertrophy	+	–
Thick alveolar bands	+	–
Lateral incisor absent	+	–
Central incisors absent	–	+
Aplasia of nasal alae	+	–

DIGITAL FEATURES

Polydactyly: mostly unilateral or asymmetrical. Bilateral preaxial polydactyly has been reported once.

Syndactyly: skin or bone.

Brachydactyly.

Clinodactyly.

SKIN

Facial milia, coarse thin hair, sometimes alopecia.

CENTRAL NERVOUS SYSTEM

Mental retardation in just over half. Pathological features include cerebral atrophy, porencephaly, hydrocephaly, hydranencephaly.

OTHER

Polycystic renal disease.

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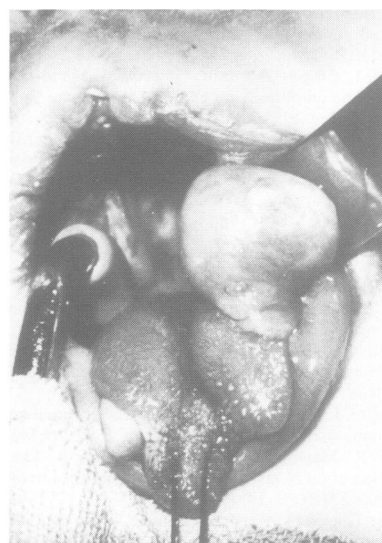


FIG 1 (a) OFD type I. Bifid tongue with nodules.



FIG 1 (b) OFD type I. Bifid tongue, less severe than in (a).



FIG 1 (c) OFD type I. Complex polysyndactyly.



FIG 1 (f) OFD type I. Unilateral brachydactyly.



FIG 1 (d) OFD type I. Midline cleft lip.



FIG 1 (g) OFD type I. Postaxial polydactyly.

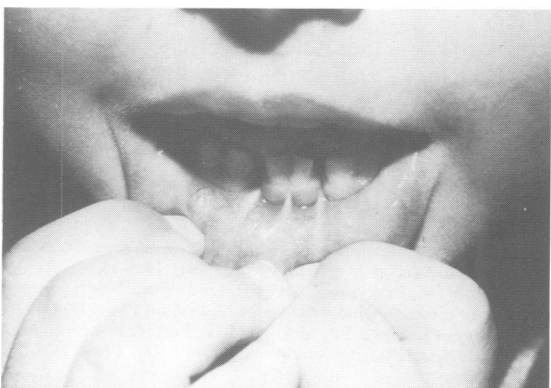


FIG 1 (e) OFD type I. Multiple frenulae.

Type II

The original description was by Mohr in 1941.² Mohr reported four males in a single sibship. Only one male was fully described; the three others had died previously. Subsequently, a male cousin was reported as being similarly affected (the parents were first cousins). One patient had unilateral syndactyly of fingers 3 and 4. The first and fifth toes were broad and the distal phalanges were duplicated. Rimoin and Edgerton³ reported three sibs (two male, one female) with the same condition and designated it as Mohr syndrome (or OFD type II).

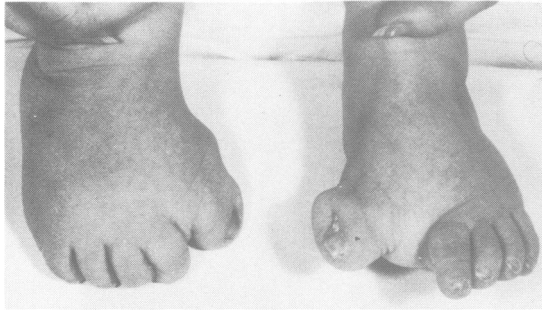


FIG 2 OFD type II. Preaxial polydactyly.

Major features which differentiate type II and I are bilateral duplication of the hallux (sometimes only visible radiologically) and occasionally of the thumbs,⁴ short stature, hearing loss, absence of milia, and absence of severe brain pathology. (Intelligence is usually normal but some members of the original family described by Mohr were retarded.) Minor differences include broad nasal tip and flat nasal bridge.

Of all of these criteria, only the presence of preaxial bilateral polydactyly of the big toes, in a male, is, in the presence of the oral pathology, very suggestive of type II.

Type III

The designation of type III has been given to the sibship of Sugarman *et al.*⁵ They reported mentally retarded female sibs with a lobulated hamartomatous tongue (in one), a bifid uvula, extra small teeth with malocclusion, and a bulbous nose. Facial clefts and hypertrophied frenulae were not present. They had bilateral postaxial polydactyly of hands and feet. One had ceaseless see-saw winking of the eyelids; each wink lasted five seconds and alternated from one eye to the other.

It is only the presence of the unusual eye movements that differentiates type III from type I.

Type IV

Temtamy and McKusick⁶ published two cases with features of both the Mohr syndrome (tongue tumours, multiple frenulae, broad flat nose, and hypertelorism) and short rib polydactyly with tibial dysplasia (Majewski syndrome). Subsequently, an inbred family with two affected members was reported.⁷

It is the presence of the severe tibial aplasia that differentiates type IV from type I.

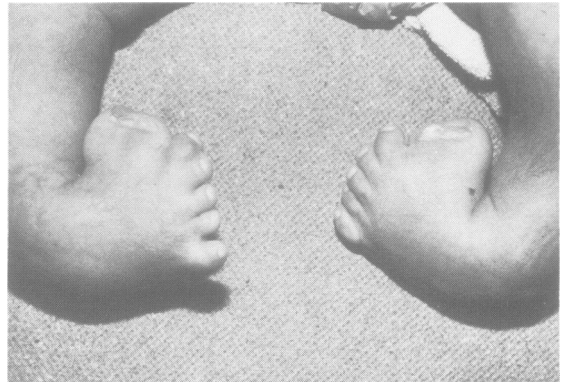


FIG 3 OFD type IV. Pre- and postaxial polydactyly in limbs with severe tibial aplasia.

Differential diagnosis

The Joubert-Boltshauser syndrome is characterised by cerebellar vermis aplasia, mental retardation, rotatory nystagmus, alternating apnoea, and hyperapnoea. Inheritance is autosomal recessive. The occasional patient has polydactyly and tongue tumours which make differentiation from OFD difficult.⁸ The sibship reported by Gustavson *et al*⁹ falls into the category but it is not yet resolved whether they form part of the OFD phenotype or that of Joubert-Boltshauser.

Number of reported cases/incidence

- OFD I — by far the most numerous.
 II — rare, fewer than 25 reported.
 III — only two reports.
 IV — only four reports.

Inheritance

- OFD I — X linked dominant.
 II — autosomal recessive.
 III — autosomal recessive.
 IV — autosomal recessive.

References

- Papillon-Leage (Mme), Psaume J. Une malformation hereditaire de la maqueuse buccale: brides et fret anormaux. *Rev Stomatol (Paris)* 1954;55:209-27.
- Mohr OL. A hereditary sublethal syndrome in man. *Skr Norske Vidensk Akad I. Mat Naturv Klasse* 1941;14:3-18.
- Rimoin DL, Edgerton MT. Genetic and clinical heterogeneity in the oral-facial-digital syndromes. *J Pediatr* 1967;71:94-102.
- Townes PL, Wood BP, McDonald JV. Further heterogeneity of the oral-facial-digital syndromes. *Am J Dis Child* 1976;130:548-54.
- Sugarman GI, Katakia M, Menkes J. See-saw winking in a familial oral-facial-digital syndrome. *Clin Genet* 1971;2:248-54.

- ⁶ Temtamy S, McKusick VA. The genetics of hand malformations. *Birth Defects* 1978;XIV:No 3.
- ⁷ Burn J, Dezateux C, Hall CM, Baraitser M. Orofaciodigital syndrome with mesomelic limb shortening. *J Med Genet* 1984;21:189-92.
- ⁸ Egger J, Bellman MH, Ross EM, Baraitser M. Joubert-Boltshauser syndrome with polydactyly in siblings. *J Neurol Neurosurg Psychiatry* 1982;45:737-9.
- ⁹ Gustavson KH, Kreuger A, Petersson PO. Syndrome character-

ised by lingual malformation, polydactyly, tachypnea and psychomotor retardation (Mohr syndrome). *Clin Genet* 1971;2:261-6.

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