The orofaciodigital (OFD) syndromes

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There are now four separate orofaciodigital (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 (COMARED WITH TYPE II)**

<table>
<thead>
<tr>
<th></th>
<th>I</th>
<th>II</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tongue nodules</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Bifid tongue</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Midline lip cleft</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Frenulae hypertrophy</td>
<td>+</td>
<td>-</td>
</tr>
<tr>
<td>Thick alveolar bands</td>
<td>+</td>
<td>-</td>
</tr>
<tr>
<td>Lateral incisor absent</td>
<td>+</td>
<td>-</td>
</tr>
<tr>
<td>Central incisors absent</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td>Aplasia of nasal alae</td>
<td>+</td>
<td>-</td>
</tr>
</tbody>
</table>

**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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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).
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.

Differential diagnosis

The Joubert-Bolthauer syndrome is characterised by cerebellar vermis aplasia, mental retardation, rotatory nystagmus, alternating apnoea, and hyper-apnoea. 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 al9 falls into the category but it is not yet resolved whether they form part of the OFD phenotype or that of Joubert-Bolthauer.

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

Syndrome of the month

Temtamy S, McKusick VA. The genetics of hand malformations. *Birth Defects* 1978; XIV: No 3.


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