Terminal Transverse Defects with Orofacial Malformations (TTV-OFM): Case Report with Mandibular Prognathism and Submucous Cleft Palate

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Terminal transverse defects of varying severity, ranging from aplasia of phalanges and digits to hemimelia, have been reported in association with various orofacial malformations. Temtamy and McKusick (1978) introduced the term "terminal transverse defects with orofacial malformations (TTV-OFM)" as a formal genesis syndrome (etiologically undefined) to include the following clinical entities: (1) aglossia-adactylia syndrome; (2) ankyloglossum (superius) syndrome; (3) Hanhart syndrome; (4) ectrodactyly with OFM; and (5) Möbius syndrome with digital malformations (including the Charlie "M" syndrome). This report describes a patient whose phenotypic characteristics qualify for inclusion under this heading, but with facial features somewhat different from those previously reported in the literature including the presence of an eccentric submucous cleft of the hard palate and zona pellucida of the soft palate.

KEY WORDS: Terminal transverse defects, orofacial malformations, submucous cleft palate

Patients with craniofacial anomalies in association with malformations of the extremities characterized by aplasia have been reported in the literature under the collective headings "oromandibular limb hypogenesis syndromes (OMLH)" (Hall, 1971; Grolin et al., 1976) and "terminal transverse defects with orofacial malformations (TTV-OFM)" (Temptamy & McKusick, 1978). The craniofa-
included the amniotic band syndrome in his table of OMLH syndromes.

The term TTV-OFM syndrome is preferred because it is clear when it refers to the type of limb malformations. Implied is a total amputation across the distal end of the limb or a portion of it (i.e., digits, fingers or toes). The facial malformations encountered are wide-ranging and non-specific. The term OMLH syndromes, as suggested by Hall (1971), is limiting because it is based on the assumption that hypoglossia is present in all cases. Review of the literature indicated that the tongue may be affected in different ways, ranging from macro to microglossia, or it may not be affected at all (Cosack, 1953; Garner & Bixler, 1969; Wexler & Novark, 1974; Herrmann et al., 1976).

Considerable overlap exists in the phenotypic characteristics displayed by the patients described under the different diagnostic headings that may be considered TTV-OFM syndromes. Such clinical entities include the hypoglossia-hypodactyly (aglossia-adactyly) syndrome, ankyloglossum (superius) syndrome, Hanhart syndrome, Möbius syndrome, Charlie “M” syndrome, glossopalatine ankylosis-microglossia-hypodontia-limb anomalies syndrome, etc. (Spivack & Bennett, 1968; Herrmann et al., 1976; Gorlin et al., 1976; Temtamy & McKusick, 1978).

The purpose of this paper is to present a patient that may be included with the TTV-OFM syndromes and whose facial characteristics are somewhat different from those described in the literature including an eccentric submucous cleft of the hard palate and zona pellucida of the soft palate.

**Case presentation**

CCFA #2912 This black female was born prematurely at six months of gestational age with multiple congenital facial and limb anomalies. Birth weight was 1342 gms. She was the fifth child born to non-consanguineous parents. The mother was treated for an undetermined infection during the fifth month of pregnancy (unknown medication, probably an antibiotic). The growth and development of the child have been adequate, requiring no special care.

On clinical examination at 12 years, 3 months of age, the facial skin had a normal appearance and texture. The right eye was microphthalmic, with opaque microcornea and no vision. The left eye was normal and well positioned. The midface appeared hypoplastic but symmetrical. The lips were normal in appearance except that the lower lip was relatively protrusive. The mandible appeared symmetrical and prognathic (Figure 1). Dental development was in the late mixed dentition stage, with a class III molar relation and bilateral buccal crossbite. In occlusion, she appeared overclosed with reduced facial vertical dimension. The right side of the hard and soft palate was smaller. An eccentric submucous cleft of the hard palate with zona pellucida of the soft palate was observed (Figure 2). On sustained phonation /a/, the soft palate elevated and deviated to the left. The
tongue was apparently normal in size, position, tone, posture, and function.

Examination of the neck, back and chest was normal. She presented normal pubertal secondary sex characteristics. The range of motion and length of the extremities were normal; however, abnormalities of the distal portions were evident (Figure 3). On the right hand the second finger had clinodactyly and a normal nail. The fingers were absent on the left hand with small fleshy nubs in the first and fifth finger positions. The right foot showed absence of the great toe and hypoplastic second toe. The left foot had hypoplastic great, second, and third toes. Chromosome analysis yielded a normal female karyotype, 46 XX.

Radiographic findings: Marked digital impressions on the endocranial surface of the frontal bone were noted. Although suggestive of craniosynostosis, there was no record of symptoms suggestive of increased intracranial pressure. There was absence of many permanent teeth (13, 12, 11, 21, 22, 23, 44, 46, 47-F.D.I. nomenclature). The maxilla was retruded in relation to the cranium. The mandible appeared normal in form and structure, but was prognathic in relation to the cranium. The anterior nasal spine was identified radiographically indicating that the premaxilla was present (Figure 4). Superimposition of serial tracings demonstrated a facial growth pattern (Figure 5) in which the original maxillary-mandibular disharmony was accentuated by virtue of greater incremental growth of the mandible. The lateral cephalometric radiographs revealed adequate velar movement and velopharyngeal closure during sustained production of the /s/ sound but not in the /u/ sound where a 5 mm. gap was measured.

Radiographs demonstrated normal carpal and metacarpal bones in both hands, with complete aphalangia with the exception of the curved right second finger (Figure 6).

Speech: Her speech has been recorded as “amazingly good” with an intelligibility of 60–65%, despite the presence of a submucous cleft palate and the absence of maxillary anterior teeth.

Discussion

As previously stated by Gorlin et al. (1976), syndromes of oromandibular limb hypogenesis are confusing because of the variable pattern of craniofacial and limb malformations encountered. The limbs may be affected in different ways and degrees (from hypoplastic toes and fingers to complete limb absence) and there is no pattern regarding what part, or how many limbs may be affected. This diversity in expression appears to occur in the orofacial region and does not follow a typical pattern either. As described in excellent reviews by others (Gorlin et al., 1976; Herrmann et al., 1976; Temtamy and McKusick, 1978) it is possible to find orbital hypertelorism, cranial nerve involvement, clefts of the face and palate, micrognathia, microglossia,

FIGURE 2. CCFA #2912, female, age 10 years, 11 months. Note absence of several maxillary teeth. The arrow on the maxillary cast shows the eccentric position of the right submucous cleft palate.
FIGURE 3. CCFA #2912, female, age 12 years, 3 months. The only digit present was a clinodactylic second finger on the right hand. Note small nubs in the first and fifth finger position on the left hand. The right foot shows absent great toe and hypoplastic second toe. The left foot presents hypoplastic great, second and third toes.

FIGURE 4. CCFA #2912, female, age 10 years, 11 months. The lateral cephalometric radiographs reveals endocranial digital impressions, a long and thin anterior nasal spine (arrows), maxillary antero-posterior deficiency and mandibular prognathism with the teeth in occlusion. The posteroanterior cephalometric headplate is not remarkable.
rather than micrognathia was present. Our patient would fit under the broader umbrella defined by Herrmann et al. (1976) for the Hanhart syndrome. The hypoglossia-hypodactyly syndrome was excluded on the basis of the presence of a normal tongue and a prognathic mandible. The Charlie “M” syndrome was excluded because of the absence of hypertelorism, antimongoloid slant of the eyes and facial paralysis. Also, Möbius syndrome was excluded because there was no involvement of cranial nerve VI. The mimetic musculature and eye motility in the right eye were normal and there was a normal chest wall structure.

Cosack (1953) and Shear (1956) reported cases with premaxillary hypoplasia and absence of maxillary incisors, as in our case. The relative prognathism of the mandible in relation to the anterior cranial base could be due to a short anterior cranial base, an intrinsically longer mandible, or a combination of both. Unfortunately, normative values for blacks matched for age and sex were not available. When compared with matched data on white females (Riolo et al., 1974), the length of the anterior cranial base was within normal limits, whereas the length of the mandible was greater than the normal value, reinforcing our impression of mandibular prognathism. Since mandibular micrognathia was the usual mandibular characteristic reported (Garner & Bixler, 1969; Wexler & Novark, 1974; Gorlin et al., 1976; Herrmann et al., 1976; Temtamy & McKusick, 1978), the finding of mandibular prognathism makes the present case exceptional. However, Lustmann et al (1981) recently reported a 17 year black female with unilateral hypoglossia, ankyloglossia, oligodontia hypodactyly and prominent mandible.

Eye anomalies observed in this patient have also been reported in Möbius syndrome (Evans, 1955).

The atypical anatomical and functional characteristics in the palate area are of interest. The deviation of the soft palate to the left during phonation may be due to a muscular derangement in the soft palate as a result of the submucous cleft palate, or to possible partial deficiency of cranial nerves VII, IX and X (Nishio et al., 1976, a,b) innervating the right side of the soft palate. The latter possibility brings our patient closer to Herrmann’s (1976) definition of Hanhart
syndrome which includes cranial nerve involvement.

Most of the cases affected with TTV-OFM syndromes are sporadic and because of the extreme variability in the type of limb and facial anomalies observed, it is thought that these conditions are probably etiologically heterogeneous (Cohen et al., 1971; Gorlin et al., 1976; Herrmann et al., 1976; Temtamy & McKusick, 1978).

**Summary**

This report presents a patient affected with limb and orofacial congenital malformations. The patient may be included under the general diagnostic heading of TTV-OFM syndromes (Temtamy & McKusick, 1978) or under Herrmann's (1976) definition of Hanhart syndrome. The limb anomalies are similar to those reported in the literature. The facial characteristics are somewhat different in that our patient presents true mandibular prognathism instead of the commonly reported mandibular hypoplasia or retrognathism. Submucous cleft of the hard palate and zona pellucida of the soft palate were associated findings.

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**References:**


