Picture Stories

A Sri Lankan boy with Thurston syndrome (type V oro-facio-digital syndrome)

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Background
Oro-facio-digital syndromes are a group of related conditions characterised by malformations in the mouth, face and digits. There are as many as thirteen syndromes described thus far[1]. The oro-facio-digital syndrome type V (Thurston syndrome) is characterised by median cleft of the upper lip, polydactyly and oral manifestations. It is extremely uncommon, with only a handful of cases, mostly of Indian descent, reported to date. Here, we report a Sri Lankan Sinhalese boy with Thurston syndrome.

Case report
A 7-year old Sri Lankan boy was seen in the clinic for abnormalities in face, hands and feet. He was the only child of non-consanguineous parents without a family history of congenital malformations. His antenatal history was unremarkable with no teratogenic exposure and his birth and early neonatal periods were uneventful. His childhood growth was normal. He achieved all developmental milestones at appropriate ages, had normal intelligence and showed good progress in school.

Examination of face revealed a median cleft of the upper lip with no cleft palate (Figure 1). He had a duplicated frenulum and malformed teeth, but the rest of the oral examination including examination of the tongue, was normal. He had postaxial polydactyly in all four limbs (Figures 2 and 3).

Figure 1: ***Photograph demonstrating median cleft lip***

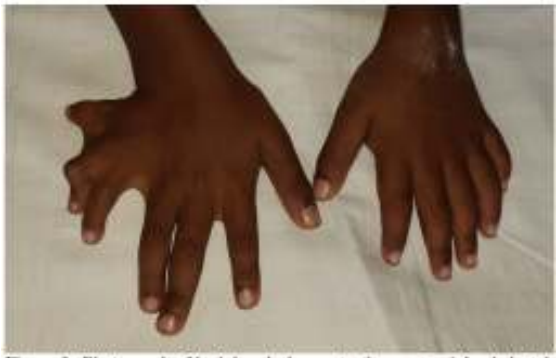

Figure 2: ***Photograph of both hands demonstrating post-axial polydactyly***

Figure 3: ***Photograph of both feet demonstrating post-axial polydactyly***

He had six toes each in both feet and six fingers in left hand with clinodactyly of sixth finger. Examination of the right hand revealed seven fingers with syndactyly of fifth to seventh digits. His head size, hair, eyes and ears were normal. Examination of other systems were clinically normal.

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Radiography of the left hand showed an extra digit at ulnar border with a metacarpal and three phalanges; middle phalanx was malformed and incurved (Figure 4).

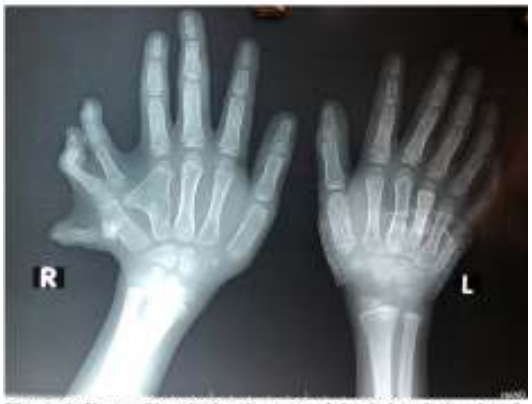

Figure 4: *X-ray of hands showing postaxial polydactyly, syndactyly of right 5th to 7th fingers and metacarpal abnormalities*

X-ray of the right hand depicted abnormal fourth and fifth metacarpals and seven fingers. Abnormally broad fourth metacarpal was shared by tri-phalangeal fourth and fifth fingers whereas fifth metacarpal was shared by hypoplastic sixth and seventh fingers. Echocardiogram and ultrasound scan of the abdomen were normal. The diagnosis of Thurston syndrome was made based on the presence of characteristic clinical features (median cleft lip, duplicated frenulum and postaxial polydactyly) and normal intelligence.

Discussion

Thurston syndrome was first reported by Owen Thurston in his description of two siblings of North Indian descent with median cleft lip and postaxial polydactyly[2]. Since then only a few cases have been reported. Dysmorphic features reported in the subsequent case reports include, duplication or hyperplasia of the frenulum, high arched palate, enamel hypoplasia, hypodontia, supernumerary teeth and clinodactyly, brachydactyly and syndactyly of digits[1,3-10]. Even though the exact genetic basis has not been established, Thurston syndrome is believed to be due to autosomal recessive mutations in the DDX59 gene in chromosome 1[11].

The differential diagnoses of Thurston syndrome include other types of oro-facio-digital syndromes. These syndromes are associated with deafness and abnormalities in the tongue, nervous system and respiratory system which were not present in our patient[8]. Thurston syndrome is the mildest of all oro-facio-digital syndromes with preserved intelligence and normal life expectancy[10,12,13]. The oro-facio-digital syndrome type I (Papillon-League-Psaume syndrome) is characterised by central nervous system anomalies, polycystic kidney disease, hypoplasia of alae nasi, hamartomas of tongue, bifid tongue and increased cranial base flexure[12,14]. It is also associated with digital anomalies including polydactyly, syndactyly, brachydactyly and clinodactyly[12]. Oro-facio-digital syndrome type II (Mohr syndrome) is autosomal recessive and is associated with cleft tongue, nodules of tongue, conductive deafness and partial duplication of hallux[12,14]. Oro-facio-digital syndrome type IV (Varadi-Papp syndrome) shows close similarity to Thurston syndrome but is associated with cerebellar anomalies and absence of oral frenulum[12,15].

This case report highlights a very rare oro-facio-digital malformation syndrome that should be considered in the differential diagnosis of median cleft lip and digital anomalies. Establishing the exact diagnosis of Thurston syndrome is vital for genetic counselling of this autosomal recessive disorder.

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