Cleft lip and palate, lower lip pits, and limb deficiency defects

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SUMMARY Cleft lip or palate and lower lip pits are typical features of the autosomal dominantly inherited Van der Woude syndrome. Limb defects have not been reported in this syndrome so far. A girl with a unilateral complete cleft lip and palate, bilateral lower lip pits, and amniotic deformities of all four limbs is reported and the possibility of chance occurrence of cleft lip and palate, lower lip pits, and limb defects is discussed.

Symmetrical lower lip pits with or without cleft lip and palate are characteristic features of the Van der Woude syndrome. There is also often hypodontia, usually affecting the second incisors and second molars. There have been no additional symptoms reported, particularly no limb defects. We have observed a child with cleft lip and palate, bilateral

lower lip pits, and congenital deformities of all four extremities.

Case report

The family history was unremarkable. The female patient was the first child of healthy, unrelated parents. There was premature rupture of membranes in the 31st week of gestation. Delivery was in the 35th week of gestation with a birth weight of 2420 g, a length of 45 cm, and a head circumference of 32.5 cm. Physical examination after birth showed a right sided cleft lip and palate and typical bilateral raised lower lip pits. Limb anomalies of all extremities were noted (figs 1 and 2). On the left hand there was syndactyly of the second and third fingers, brachydactyly of the second, third, and fourth fingers, absent nails on the second and third fingers, and a hypoplastic nail on the fourth finger. On the right hand there was syndactyly of the first, second, and third fingers, a shortened second finger with missing middle and distal phalanges, an absent distal phalanx of the third finger, and a dysplastic nail on

FIG 1 Hands of the patient after surgical correction of syndactyly showing distal amputations, nail dystrophy, and constriction rings of both thumbs.

FIG 2 Feet of the patient showing distal amputations of the right second to fourth toes and constriction rings of the right big toe.

FIG 3 Hand X ray. Note absent or hypoplastic distal phalanges and missing middle phalanges.

FIG 4 Surgically corrected right sided cleft lip and conical elevations of the lower lip with shallow pits and tiny openings.

Case reports

the thumb. There was right sided club foot and absent second to fourth toes with a small soft tissue dimple in the place of the second toe. The left foot showed no visible abnormalities. Constriction rings could be seen on both thumbs, on the left third finger, and the right big toe.

Fig 3 shows an x ray of the hands. In the left hand the middle and distal phalanges of the second finger are absent except for a small pea-like bone. Both middle phalanges of the third and fourth fingers are missing. In the right hand there is a rudimentary distal phalanx in the first finger and proximal phalanx in the second finger. There are only two phalanges in the third finger. The proximal phalanges of the right second to fourth toes are present with total cutaneous syndactyly. The middle phalanx of the right fifth toe is missing and the distal phalanx is rudimentary. The right distal phalanges of the first to fourth toes are hypoplastic and the distal phalanx of the fifth toe of the left foot is absent.

Physical examination of the girl showed no further abnormalities. The cleft lip and palate as well as the syndactyly in both hands have been corrected surgically. Without operative correction the lower lip pits became smaller and shallower during early childhood. At present only conical elevations can be seen at the position of the pits (fig 4).

Discussion

Cleft lip and palate and lower lip pits are the main symptoms of the syndrome of lip pits and clefts or Van der Woude syndrome.¹ The clinical expression of the pits and clefts is highly variable. The typical lip pits are bilateral, symmetrical, raised openings on the upper border of the lower lips, but unilateral or median lip pits have been reported.² Pits can be observed as microforms, similar to conical elevations, without any deeper sinuses. Clefting can be unilateral or bilateral, complete or partial, cleft lip alone or with cleft palate. The incidence of hypodontia is increased in patients with Van der Woude syndrome. Inheritance of this syndrome is autosomal dominant. Reduced penetrance has been reported, but is probably caused by incomplete examination of the families.¹ The incidence of Van der Woude syndrome was estimated by Rintala *et al*³ in Finland to be 1 in 33 000 among livebirths. To the best of our knowledge congenital anomalies of the extremities have not been reported previously.

The limb deficiencies of the patient described above with constriction rings, amputation, and nail defects are features which are characteristic of the amniotic band sequence or ADAM (Amniotic Deformities-Adhesions-Mutilations) sequence. The clinical manifestations of the amniotic band sequence range from single constriction rings to complex anomalies with abdominal wall defects and craniofacial abnormalities. The incidence of congenital constriction rings was calculated as 1 in 1300 to 1 in 15 000 pregnancies.^{4 5}

Cleft lip/palate and limb defects are cardinal features of the EEC (Ectrodactyly-Ectodermal dysplasia-Cleft lip/palate) syndrome. However, lip pits are not known in EEC syndrome. The limb anomalies of EEC syndrome are highly variable, but usually of the lobster claw type without constriction rings.⁶

From the reported incidence of Van der Woude syndrome and amniotic band sequence, the probability of the coexistence of both syndromes by chance would be a rare but possible event. Because the appearance of lip pits, clefts, and limb defects has not been described so far, although hundreds of patients with only one of these defects are known, this could be the case in our patient, but the limb defects could also represent a previously unrecorded effect of the Van der Woude syndrome gene.

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