Anonychia with absent phalanges and brachydactyly: A report of two unrelated cases

Sir,

Congenital anonychia or its milder phenotypic counterpart, hyponychia, is a relatively rare abnormality of nail development affecting all or some of the finger and toe nails, characterized by the complete absence of nails or the presence of only rudimentary nails, respectively. It could occur as an isolated abnormality, termed anonychia simplex or as part of a syndrome.[1] We report two cases of partial hyponychia/anonychia with underlying absent distal phalanges in both, and in addition hypoplastic/absent middle phalanges in one of them.

A 35-day-old female child, second born to non-consanguineous parents was brought with absent finger nails since birth. The mother denied any intake of medication or illness during the antepartum period. There was no similar history in the family. The child had short digits along with rudimentary nails of digits 2, 3, and 5 of the right hand and digit 5 of the right hand with absence of nails on digit 4 of the right hand and digits 3 and 4 of the left hand; the nails of the index finger of left hand, both thumbs and all toes were normal [Figure 1a and b]. The head circumference was 35 cm (50th centile for age) and there was no facial dysmorphism. The skin did not reveal any pigmentary abnormality. The plain radiograph of both the hands revealed the absence of distal phalanges of digits 2-5 [Figure 1c and d]. The child’s pelvis X-ray did not show iliac horns, the renal function was normal and the ultrasound of abdomen did not show any renal abnormality. The child’s brainstem-evoked response audiometry (BERA) was also normal.

Our second case was a 11-year-old girl, second born of a non-consanguineous marriage, with congenital absence of nails of the right hand. There was neither a positive family history nor a history of maternal drug intake during pregnancy. Her right hand had shortened digits 2-4 with poorly defined finger creases. Digits 3 and 4 had anonychia, whereas the little finger and index finger nails were hypoplastic [Figure 2a and b]. The nails of the right thumb, all digits of the left hand and all toes were normal. X-ray showed absence of middle and distal phalanges of digits 3-5 and absence of distal phalanx with hypoplasia and tapering of the middle phalanx of digit 2 of the right hand. There were rudimentary epiphyses seen in digits 3, 4 and 5 of the right hand [Figure 2c]. She had no other cutaneous abnormalities.

Congenital anonychia/hyponychia is a spectrum of nail abnormalities which often occur concurrently in an individual. It is inherited as an autosomal dominant or recessive trait or can occur sporadically.[1] Simple anonychia has been identified to be due to mutation in R-spondin 4- gene and some cases of SOX9 duplication are reported as causes of brachydactyly-anonychia. Onychoatrophy, on the other hand, is distinct from hyponychia in that the nails are reduced in size and thickness. Table 1 lists the associations of anonychia/hyponychia and causes of onychoatrophy.
Cooks et al. had initially described a distinctive syndrome occurring in two generations of a family of anonychia, brachydactyly of the 5th digit of the hands and digitalization of the thumbs with absence and/or hypoplasia of the distal phalanges of the hands and feet.\(^2\) Since then, some cases have been identified akin to the syndrome.\(^3,4\) Our cases had partial hyponychia/anonychia involving some of the finger nails with normal toe nails along with brachydactyly of the affected digits. They both had in common absence of phalanges (distal phalanges in both the cases along with tapering/absent middle phalanges in the second) on radiographs of the hands. They had no other associated abnormalities associated and no family history of a similar abnormality. Genetic analysis was not performed in our cases.
Letters to the Editor

Aplasia cutis congenita, group 5 without fetus papyraceus in two newborns

Sir,

Aplasia cutis congenita is an uncommon disorder characterized by the congenital absence of a portion of the skin, the most common presentation being a solitary lesion on the vertex of the scalp, just lateral to the midline (70%). Lesions are usually non-inflammatory, well-demarcated and have a variable appearance depending on the size (0.5–10 cm), extent, depth and degree of healing and scarring.

The various causative pathophysiological mechanisms include genetic factors, intrauterine trauma, fetus papyraceus, intrauterine infections by varicella or herpes viruses, drugs including methimazole, carbimazole and misoprostol, feto-fetal transfusion, early rupture of amniotic membrane forming amniotic bands and abnormal elastic fiber biomechanical forces.

Truncal aplasia cutis congenita is usually associated with fetus papyraceus, classified as group 5. We report a rare presentation of aplasia cutis congenita, group 5 in two newborns without any history of twin gestation/fetus papyraceus.

**Case 1**

A 2-day-old healthy male child with a birth weight of 2.9 kg, delivered by full-term normal vaginal delivery to a multigravida mother, presented with congenital absence of skin on bilateral flanks. Ultrasonography done at 18 and 32 weeks antenatally had revealed a single live fetus with normal parameters and with no obvious congenital malformations. On examination, the lesions had a gelatinous appearance with a glistening red base and visible capillaries covered with a thin transparent membrane. They were 6.3 cm × 4.2 cm, well-defined and symmetrical, present on bilateral flanks joined with a linear streak [Figure 1a].

**Case 2**

A newborn healthy male child, weighing 2.8 kg. delivered by full-term normal vaginal delivery, presented with absence of the skin on the trunk, arms and legs. His mother had a history of typhoid fever at 6 weeks of gestation which was managed conservatively; the antenatal period was otherwise uneventful. Ultrasonography at 18 and 28 weeks antenatally revealed a single live fetus with healthy parameters and no twin gestation was noted. On examination, the lesions were well-demarcated, symmetrical, linear/elliptical and stellate. They were covered with a thin, gelatinous, shiny membrane and were present on both flanks, upper arms and thighs, distributed in a H-shaped pattern measuring 7.1 cm × 4.4 cm [Figure 2a].

There was no history of any intrauterine infection/trauma or exposure to any teratogenic drug in both cases. Scalp and nail examination, radiological examination and ultrasonography revealed no abnormalities in both children. Lesions healed within 2.5 months and 1 month of conservative treatment with

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