Congenital Anonychia of the Toes with Absence of Underlying Phalangeal Bones

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Congenital absence of nails usually occur as a rare isolated anomaly or combined with other ectodermal defects. This anomaly is regarded as an inherited disorder either dominantly or recessively but quite a few cases were reported as sporadically developed.

The patient was a 2-month-old girl who had no nails on both her 2nd, 3rd toes but had rudimentary nails on her left big toe and both 4th toes at birth. We could not find any other congenital deformity, any family history of inherited diseases related to anonychia. Radiological findings revealed no visualization of both 4th distal phalanges, only.

We report this case as congenital anonychia of a sporadic type which may have developed independently from an underlying bone abnormality. We also review other reported cases in the literature.(Ann Dermatol 9:(3) 188~190, 1997).

Key Words : Congenital anonychia, Bone abnormality, Sporadic type

Congenital anonychia or congenital absence of nails is a very rare anomaly which may occur dominantly[1-8], recessively[9,10] or as an isolated condition[11-13]. Reported cases were seldom associated with various skeletal[14] or dental abnormalities[5]. They were however, associated with ectrodactyly, incomplete or partial congenital absence of one or more digits.[1,7,8,12,13]

Recently, a case of congenital onychodysplasia of the index fingers was reported by Choi et al[15], but no case of congenital anonychia has been described in Korean dermatology literature.

We present a case of congenital anonychia that occurred sporadically and discuss its relationship with underlying osseous abnormalities such as loss or hypoplasia of the phalanges.

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CASE REPORT

A two-month-old girl with absence of toe nails since birth visited our department. There were no nails on the 2nd and 3rd toes of both feet, and pin-head sized rudimentary nails(remnant of nail) were found on her left big toe and both 4th toes. On examination, nail structures such as nail plates and folds were completely absent in all of the affected toes. In addition to nail absence, both 2nd and 3rd ones of the affected toes were relatively shorter than normal appearing toes(Fig. 1).

The patient was born as a full-term baby by normal vaginal delivery, and the body weight was 3.0 kg, height 49.2 cm, head circumference 33.0 cm, chest circumference 32.5 cm. These were within the 25 to 50 percentile. The past medical history of her mother, who had been a nurse, during pregnancy was all insignificant. The parents were not consangineous and no family history of similar nail deformities or hereditary diseases related to ectodermal disorders were found.

The results of the following laboratory tests were within normal limits or negative: a complete

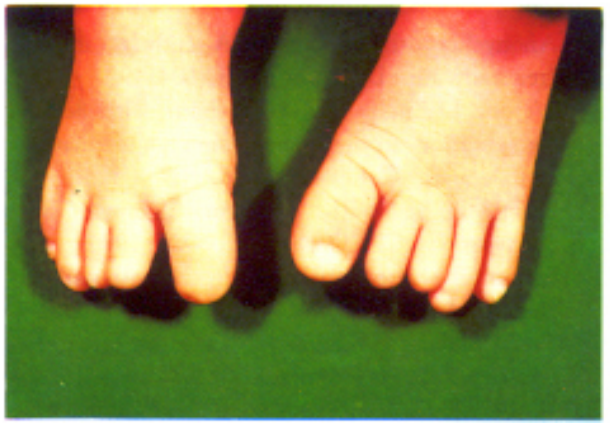

Fig.1. Feet of the patient showing absence of nails and shortening of both 2nd and 3rd toes, pin head sized rudimentary nails on the left big toe and both 4th toes.

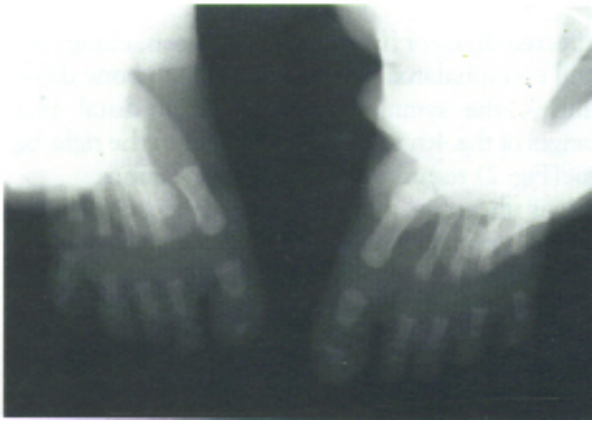

Fig. 2. Radiological findings of the 4th toes revealed no visualization of the distal phalangeal bone.

blood count, platelet count, liver function test, urinalysis and stool examination. The roentgenogram of the skull 4 series, chest, abdomen, elbow joint, wrist joint, hip joint, knee joint and hand were all normal and compatible with her age. However, on the examination of both feet, the 4th toe area revealed no visualization of distal phalangeal bones and the distal phalangeal bone of the right big toe was rather smaller than that of the left one(Fig. 2).

DISCUSSION

The sporadic type of congenital anonychia occurs without any other defects but is sometimes associated with ectrodactyly,[1,11-13] or is combined with onychoatrophia or rudimentary nails without a skeletal anomaly[4].

Over sixty years ago, Cockayne[16] classified anonychia into four types. Type 1, the most common type, showing recessive inheritance mainly due to consanguineous marriage; Type 2, a dominant inheritance pattern; Type 3, named anonychia pollicum, described as an abscence of thumb nails with red, sensitive nail beds; Type 4, compatible with nail-patella syndrome. Thereafter, another clinical entity of anonychia, a sporadic type was reported[1,11,13].

On review of previously reported cases by Baran et al,[12] Verbov[2], Kumar et al[3], Cooks et al[7], and Nevin et al[8] anonychia usually involves both fingers and toes, simultaneously. However, the cases of Yesudian et al[1] and Hopsu-Havu et al[9] presented the isolated deformity on finger nails which compares with our presenting case as well as the case of Ortonne et al[11] where congenital anonychia manifested on the toes, exclusively. In most cases, the distal phalangeal bones of affected fingers or toes were usually absent or hypoplastic, shortened of terminal phalanges(brachydactyly) or hypotrophy. In addition to complete absence of nail structure-nail beds, nail folds and terminal creases[3], the thumb or great toes also mainfest bulbous tip and digitalization[7]. We also found a deformity that affected the toes showing hypoplasia, brachdactyly and bulbous tips which resemble the case report of Cooks et al[7](Fig. 1). Other clinical findings related to anonychia as a hereditary disorder described by Verbov[2] were sore, crackled soles and hypo-, hyperpigmentation of the axillae, groins and hair abnormalities. Sequeiros and Sack[14] also presented the congenital anonychia with various cutaneous manifestations such as linear skin atrophy, depressed scar like alopecia areata and an abnormality of the tongue in a monozygotic twin. Those cases well suggested that anonychia usually occurs along with other inherited dermatosis involving ectodermal development rather than an isolated anomaly. Compared to hereditary congenital anonychia reported before, it seems likely that our case is a typical sporadic type because none of the family members including relatives of three generations evaluated had a similar nail anomaly as well as any other hereditary disorders.

Radiological examinations observed by others [1,3,7,11,13] showed the total absence of distal phalanges in

affected digits or hypoplasia, synostosis, elongation and extraphalanx. We also found such bone deformities, the symmetrical absence of distal phalanges of the 4th toes and hypoplasia in the right big toe(Fig. 2) regardless of normal appearance.

It is uncertain that the underlying cause of congenital anonychia either hereditary or sporadically have developed. Baran and Juhlin[12] suggested the close relationship between the developing nail and underlying bone. We agree with their conclusion that `there will be no nail if the distal phalanx is lacking. When the distal phalanges are hypoplastic the nails may be normal, hypoplastic or absent'. However, our presenting case have different aspects which disagree with the role of the phalanx, especially the middle phalanx proposed by Baran and Juhlin[12], in developing anonychia.

Furthermore, as shown in our case presentation, congenital anonychia could happen without any underlying osseous abnormality(2nd, 3rd nail), independently. This phenomenon represents the possible hypothesis that ectodermal development for the nail and underlying bone structure share a common stage during the differentiation process. In addition, the extent of the anomaly may be greatly influenced by the degree of individual bone deformity(Lt. big toe nail), not by a single dominant bone abnormality responsible for all nail abnormalities.

We do need to observe this case on a progessive basis in the future. On the other hand, it was revealed that most patients with the sporadic type[11-13] maintained normal healthy status throughout the life.

Congenital anonychia is rarely combined with other skeletal[7] or dental anomaies[5] but it may not correlate with any other abnormalities like in our case. In such a sporadic case, further investigation would be necessary to solve the role of the underlying bone to find out if it directly influences nail formation or not.

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