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CASE REPORT

# Brachydactyly, anonychia and a deformed nasal tip in a 16-year-old girl: A case report

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**KEYWORDS**
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**Summary** The current report describes a case of a 16-year-old girl with a rare combination of nasal dysmorphology, anonychia and brachydactyly of hands and feet. The combination of hand and nasal malformations suggested a type B brachydactyly. Genetic investigation, however, revealed Cooks syndrome with unexplained facial dysmorphology. Concerning her cosmetic complaints, options for surgical treatment were discussed. It was decided to treat only the nasal deformity by open rhinoplasty, leaving the nail deformities undisturbed.


Anonychia and hyponychia are rare anomalies. Congenital anonychia is the total absence of the nail in one or more fingers or toes. In hyponychia, there are some remnants of the nail present. These conditions are often accompanied by brachydactyly or even the complete absence of terminal phalanges.[1–5] Isolated cases of anonychia combined with facial dysmorphology, with deformation of the nose in particular, have been described in genetic literature.[6,7] It is very likely, however, that these patients are referred to a plastic surgery practice to seek advice for their cosmetic complaints. In this case report, we present a patient who consulted our plastic surgery clinic with a rare combination of anonychia, brachydactyly and nasal deformity.

## Case report

A 16-year-old girl, recently migrated from Iraq, consulted our clinic with short fingers, anonychia and a deformation of the tip of her nose, which were present since birth.

All finger- and toenails, including the nail matrices, were absent. Only small nails on the left third finger and the left fifth toe were seen (Figure 1). X-rays of the hands revealed that digits 2–5 had only two phalanges, slightly elongated. Tips of the most distal phalanges did somewhat resemble processi unguiculares. In the left third finger, where a small nail was present, a rudimentary distal phalanx was seen. In the feet, all distal phalanges were missing, including the distal phalanges of the halluces (Figure 2).

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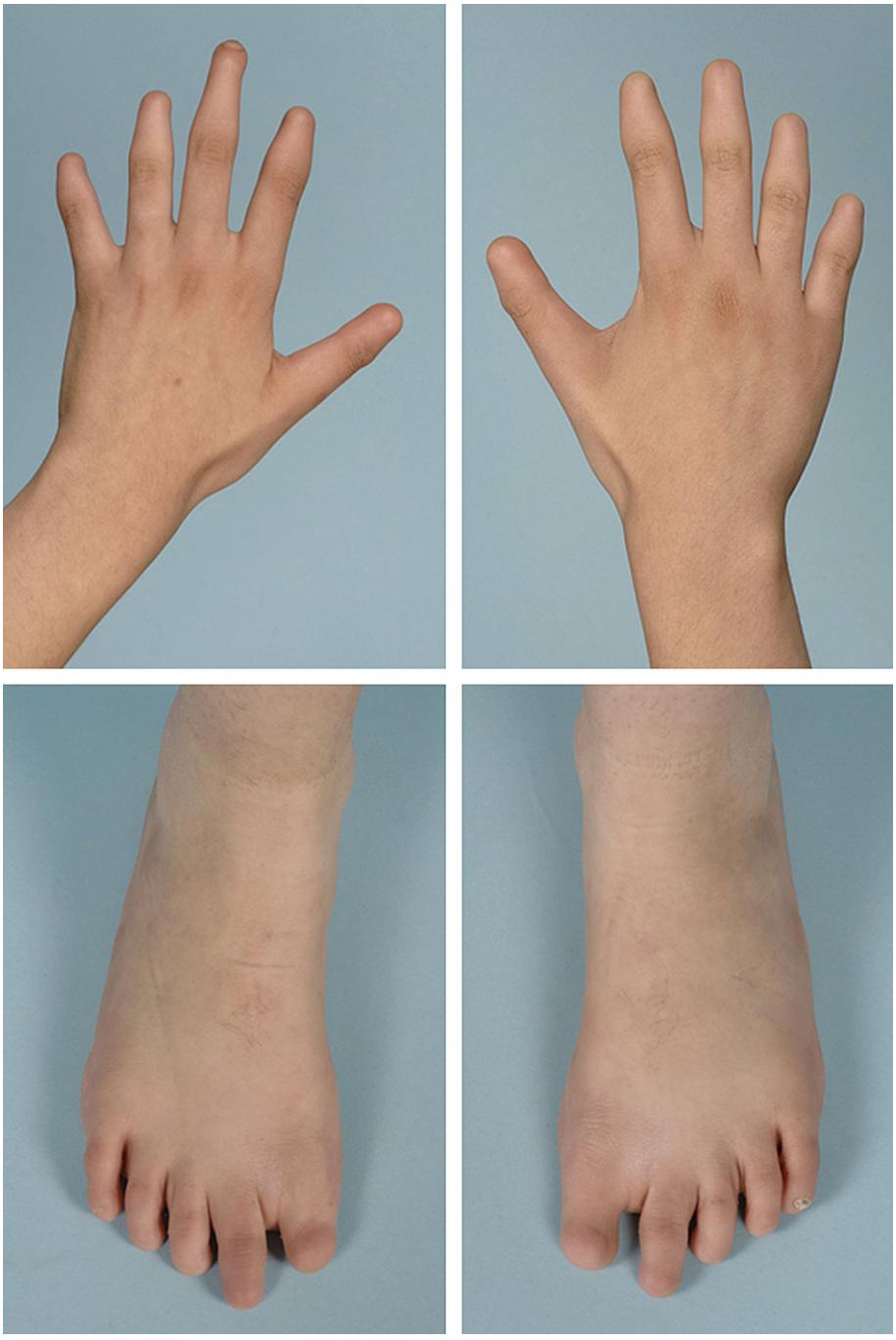

**Figure 1** Left hand: nails are absent. The middle finger has a nail remnant. Right hand: total anonychia. Right foot: all nails are absent. Left foot: nails are absent, digit 5 has a nail remnant.

The nasal tip was bulbous and flattened, with a normal although broad nasal bone (Figure 3). The columella was short. No other congenital abnormalities were present.

There was no history of anonychia or facial deformities in the family; her brothers had normal fingers and facial characteristics. There was no consanguinity in the family. Her mother neither used any drugs nor suffered from diseases during her pregnancy. It is unclear whether the use of chemical weapons in Iraq could have influenced the embryologic development. Her parents had been in Iraq at the time weapons were used, but they did not know what kind of weapons these were.

## Genetics

Facial defects and deformities of the digits, including the nails, fit in only a few syndromes. Brachydactyly type B is one of them, the phenotypes of which are described by Houlston[6] in 1993. Patients with type B brachydactyly have