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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
Pachyonychia congenita with steatocystoma multiplex.  
A report of two cases and a discussion of the classification

Pachyonychia congenita is a rare syndrome in which the main and most common clinical sign is onychodystrophy of all finger and toe nails.  
The most frequent type of transmission seems to be autosomal dominant, but recessive forms have also been described.  
Typical onychodystrophy can be associated with other clinical manifestations. The most recent literature refers to descriptions of about 250 cases up until 1993.  
Numerous classifications of pachyonychia congenita have been suggested by several authors over the years.  
We report two cases of pachyonychia congenita in association with steatocystoma multiplex in a mother and son. (Key words: genetic disease, onychodystrophy, pachyonychia, steatocystoma.)

Pachyonychia congenita (PC) is a rare, genetic disease with an autosomal dominant inheritance and a high degree of penetration [1], which may be the product of two adjacent genes [2]. However, some authors have hypothesized an autosomal recessive mode of inheritance for this rare disease [3]. The main sign, pachyonychia, usually arises at birth or in childhood [4].  
The presence of onychodystrophy of all finger and toenails, suggests the diagnosis of pachyonychia congenita, even in the absence of other clinical signs [5].  
Associated anomalies include palmo-plantar hyperkeratosis, follicular keratosis, hyperhidrosis, leukokeratosis and precocious dentition.  
Steatocystoma multiplex (SM) is frequently associated with PC [6-8]. On the basis of the whether or not this associated symptom is present, numerous subdivisions have been proposed. The first classification was carried out in 1935, by Kummer and Loos [9]. The most recent was undertaken in 1980 [1] by Schonfeld, who distinguished three forms.  
We report two cases of PC in association with SM, in a mother and son.

Cases reports

P.A., a 29-year-old woman, had a malformation of all finger and toenails from birth (Figs. 1 and 2). They appeared short, thickened, hardened, yellowish-brown, with subungual hyperkeratosis and distal nail plate hyperconvexity.

Figure 1. Onychodystrophy of all fingers.

Figure 2. Onychodystrophy of all toenails.
Moreover, she presented numerous nodular lesions from 0.5 to 2 cm in diameter, spread over her scalp, the nape of her neck (Fig. 3), the cervical region (Fig. 4), her trunk, abdomen and upper limbs (Fig. 5). These lesions had appeared at around the age of 12 years and, over the had course of the years, most of them had increased in number and size while two had spontaneously disappeared. Histological examination of two of these lesions led to a diagnosis of steatocystoma.

The woman also had a linear verrucous nevus in the cervical region of the right shoulder blade.

The woman reported that she was born with two of her inferior incisor teeth already erupted and that she had suffered from scalp alopecia at the age of 17.

Our second patient was her 2-year-old son, who presented the same nail dystrophies from birth, symmetrically distributed over finger and toenails.

His hair appeared fine, fragile and woolly; microscopic examination under direct and polarized light revealed the presence of torsion and crushing of the hair shaft.

The boy presented considerable hyperhidrosis of the palms and soles, follicular hyperkeratosis and he was born with two of his incisor teeth already erupted like his mother.

**Discussion**

Pachyonychia congenita is a rare syndrome in which the main and most common clinical sign is onychodystrophy of all finger and toe nails. They show thickening and hardening, a yellow-brown discoloration, subungual dyskeratosis, often angulate toward the center with hypercurvature, distal friability and fragmentation of the free edge. The most frequent mode of transmission seems to be autosomal dominant, but recessive forms have also been described [3].

The typical ungual changes described above can be associated with other clinical manifestations. The most common are palmo-planar hyperkeratosis, mucous leukokeratosis, follicular keratosis and hyperhidrosis [9].

PC was first described by Muller [10] in 1904, but in 1716 Carl Musaeus reported the case of a young woman “with monstrous nails” in his doctoral thesis [11].

In 1906, Jadassohn and Lewandowsky [12] observed another case and since then the syndrome has been known by their names.

In 1943, Ormsby and Montgomery [13] were the first to classify the numerous clinical manifestations associated with pachyonychia congenita. They classified the most frequently encountered clinical signs, including: dystrophic nail alterations, palmo-planar keratosis, hair anomalies, mucous leukokeratosis, follicular keratosis, blister formation, corneal dyskeratosis, the presence of teeth at birth or precarious dental eruption, cataracts, congenital white sponge nevus, alopecia, angular cheilitis, laryngeal lesions and mental retardation.

The most recent literature refers to descriptions of about 250 cases up until 1993 [4-8].

Since then, other associations have been mentioned (but they were considered less frequent), such as multiplex steatocystoma, dermoid cysts, vitiligo and pathologies such as schizophrenia, depression, epilepsy, migraine headache, polydactyly, microcephaly, Lesch-Nyhan Syndrome and Kyrie disease [8].

Abnormal laboratory changes, included eosinophilia with elevated levels of IgM and hypogammaglobulinemia, have frequently been reported [1].

Numerous classifications of PC have been suggested by authors over the years.
One of the most recent, suggested by Schonfeld [1] in 1980, is composed of three groups. Type A: symmetric ungual dystrophies, palmo-plantar hyperkeratosis, follicular keratosis, blister formation, oral leukokeratosis, hoarseness, hair alterations, hyperhidrosis (Jadassohn-Lewandowsky Syndrome). Type B: findings of type A with steatocystoma multiplex and neonatal dental eruption (Jackson-Sertoli Syndrome). Type C: findings of type A with corneal dyskeratosis (Schaffer-Brunaer Syndrome).

Another classification, suggested by Feinstein [8], goes back to 1988. In it, Type I includes the most frequent forms (56% of all cases), characterised by ungual dystrophy, palmo-plantar keratoderma, follicular keratosis, leukokeratosis. Type 2 presents clinical findings of Type 1, plus palmo-plantar blisters, palmo-plantar hyperhidrosis and steatocystoma multiplex. In Type 3, the previously mentioned manifestations are associated with angular cheilitis, corneal dyskeratosis and cataracts.

Finally, Type 4 presents the symptoms described in the other three forms plus laryngeal lesions, hoarseness, mental retardation, hair anomalies and alopecia.

PC remains a syndrome characterised by extreme phenotypic variability. For example, in 1994, a case was described of ungual alterations in the absence of other anomalies. This was interpreted as a monosymptomatic form, which could be determined by phenotypic presentation of a less penetrant allele of the same gene [5].

We can cite another case, reported in 1995, to support this hypothesis: an entire family affected by late onset PC [14]. Another case, described in 1995, involved a man presenting PC and multiple trunk dermoid cysts, with the diagnosis of Type 2 PC [15] in association with axonal polyneuropathy and cutaneous signs of neurofibromatosis. This case also reveals the complex and still unelucidated correlation that exists between the various neurocutaneous syndromes.

In as far as our cases are concerned, it seems interesting to report evidence of dominant autosomal transmission (mother and son). The associated findings helped us to classify our cases as Type 2 with universal onychodystrophy, with concomitant SM, neonatal teeth in both patients, alopecia and linear verrucous nevus in the woman, and hair alterations in the son.

Some of these symptoms have been reported in the literature as being less frequently associated (according to Feinstein, alopecia is present in 2.4% of the cases, SM in 15.4%, hyperhidrosis in 19.8%) with the main symptom, pachyonychia. Perhaps we could suggest another sub-type, taking into consideration our observation of a further association with verrucous linear nevus.

References