



Pachyonychia Congenita Project

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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.

HEREDITARY ONYCHOGRYPHOSIS

By AAGE VIDEBÆK (*From the University Institute of Human Genetics, Copenhagen*)

Among the fairly large number of abnormalities of the nail, there are several hereditary conditions. The inheritance is often dominant and the changes symmetrical, perhaps involving all the nails of the hands and (or) the feet. Frequently they are accompanied by other changes of the ectoderm (multiple atheromata, dystrophy of the hair, changes in the pigmentation) and sometimes with malformations in the mesoderm or entoderm (patellar defect, various dislocations, clubfoot), or else they may be links in more generalized degenerations (eruption of teeth at birth, psychosis, psychopathy).

Reports have been published of only a few families affected with onychogryphosis and, even as an isolated phenomenon, it is rather uncommon.

Onychogryphosis is characterized by the following changes of the nails: marked thickening, increased consistency, marked convexity, laterally as well as longitudinally, and, as a rule, a dark colour. Disregarding the symptomatic cases, which rarely involve all the nails and the so-called idiopathic ones, the present writer is only concerned with the familial cases.

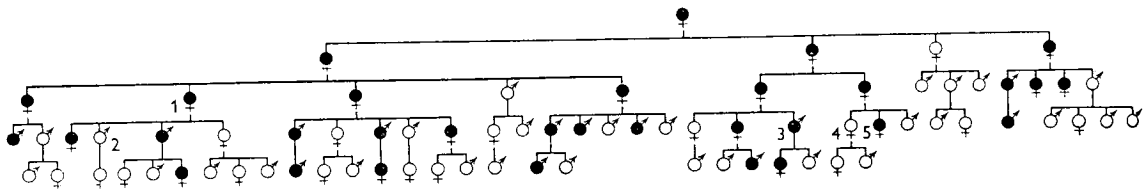
Blech, in 1846, reported onychogryphosis (confined to one finger) beginning at about the age of 9 years; a similar phenomenon was to be found on the corresponding finger of a brother and sister as well as the mother. Billroth (1869) also described familial onychogryphosis in a brother and sister. The abnormality did not appear until about the age of 17, and the parents are stated to have had normal nails. These two cases of familial nail changes do not belong to the usual type in which the deformity is congenital. It is therefore extremely likely that these were not cases of onychogryphosis. The changes described by Brochard-Rigaud (1883) in the nails of two brothers were marked by paronychia, and, in one of the brothers, the nail was atrophic. This does not correspond to the modern interpretation of onychogryphosis. Müller, in 1904, described a typical, total onychogryphosis in a girl, aged 14. The deformity was congenital and, in addition, the hair of the head was thin and the eyebrows absent. There was no information about familial occurrence. Koehler, in 1909, found onychogryphosis localized to the great toes of a woman, and similar changes in her mother, grandmother and daughter. He did not, however, perform detailed family investigations. In one family Mikula (1923) observed total onychogryphosis in two males and three females. The deformity had developed during the first month of life; at birth there had only been a yellow stripe along the nail-wall. Schmidt (1927) described total onychogryphosis, combined with an almost total and generalized alopecia, with dominant inheritance in thirteen males and twenty females in five generations. In the same family, two persons suffered from schizophrenia, one from other mental disorder, and one was hydrocephalic. Orel (1928) gave a four-generation pedigree of total onychogryphosis; there were eighteen children thus involved whose fathers or mothers had been affected with the deformity. Lastly, Tourdine & Soullignac (1937) describe onychogryphosis of all toes in a woman and in her father.

Accordingly, total onychogryphosis has only been reported in three families with thirty-three, eighteen and five affected members respectively. In all the families the inheritance was

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dominant. In cases where the deformity was limited to the great toe or all toes the inheritance was also dominant. In Schmidt's opinion, this anomaly has arisen by mutation. The family reported by him also exhibited other ectodermal changes.

The writer has observed onychogryphosis in thirty-two members of a family in Copenhagen, distributed among five generations. In all cases the onychogryphosis involves all nails of hands and feet. The deformity is congenital and rather more marked during the first years of life than later on. The lunula is preserved, but the entire nail is of a hard consistency and thickened, particularly towards its free margin. The colour is only slightly darker than normal. Some of the persons affected with the abnormality complained of paronychia, which, however, is in all probability due to the difficulties of manicure. The disease manifests itself in the form of a typical dominant affection. Presumably it has arisen by mutation. It is stated that the first known case was the youngest (a daughter) of a family of twelve. Unlike her sisters and brothers she



Genealogical table, showing a family with onychogryphosis: (1) pernicious anaemia, (2) acute myelogenous leukemia, (3) ectodactylism, (4) and (5) congenital deafness.

was usually kept in the kitchen and pantry because of her ugly nails. One day a suitor came to the house, but the daughter with the disfiguring nails was not presented to him like the others. He had, however, heard about all the daughters and missed one. At last she was fetched, and the suitor chose her as his bride. As evident from the figure she transmitted the deformity of the nails to thirty-one of her descendants.

In no case were both parents affected with the abnormality. No children acquired it unless it was present in either the father or the mother, for among those members of the family not affected with the deformity twelve have had a total of twenty-four children, all with normal nails. In other words, the inheritance is dominant. In the families where there is a possibility of acquiring onychogryphosis, i.e. among children with a father or mother affected with the deformity, there are twenty-six boys and twenty-five girls. Of them thirteen boys and eighteen girls are affected with onychogryphosis. Exactly 50% of the boys were born with onychogryphosis, while about 70% of the girls were affected. There is, however, no definite preponderance of females ($\chi^2 = 2.59$; $P = 0.189$). Skin and hair are said to be normal in all the relatives. One woman also suffered from pernicious anaemia, and her son, who developed acute myelogenous leukemia, had normal nails. In a male, affected with onychogryphosis, the middle, ring and little fingers of the left hand were missing. A female with onychogryphosis was born deaf just like her sister who was not onychogryphotic. Mental disorder did not occur in the family. The presence of the diseases or defects mentioned can, however, hardly be considered relevant to the onychogryphosis, as they were mainly isolated cases observed in members of the family with normal nails.

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SUMMARY

The paper reviews the previously reported cases of hereditary onychogryphosis.

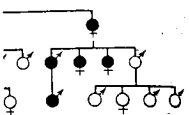
A family comprising seventy-six members exhibits total onychogryphosis in thirty-two instances, distributed among five generations. The abnormality is presumed to have arisen from mutation. The inheritance is dominant.

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