



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.

Pachyonychia Congenita Associated with 46, XYq- Karyotype

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Introduction

Pachyonychia congenita, which is characterized by the thickening of the finger- and toenails, was first described by Jadassohn and Lewandowsky in 1906 [1]. The disease is considered to be transmitted by a simple autosomal dominant gene with incomplete penetrance.

Deletion of the long arm of the Y chromosome (Yq-) may show several characteristics including short stature, hypospadias, muscular dystrophy and mental retardation [2-5].

We report a male infant with an unusual association of pachyonychia congenita and 46, XYq- karyotype.

Case Report

The patient was born at 38 weeks of gestation by a healthy gravida 3 para 3 mother after an uneventful pregnancy. His birth weight was 2940 g and length was 50 cm. His mother noticed thickening of all his nails since birth. Otherwise he grew well with apparently normal appetite until one month of age, when he

weighed 3650 g. During the next 3 months, he gained only 9 g per day, and he was referred to our hospital at the age of 4½ months for evaluation of poor weight gain and thickening of nails.

Physical examination revealed a mildly floppy boy with a weight of 4550 g (< 3 percentile) and a height of 59.5 cm (< 3 percentile). Head control was incomplete, and his postnatal development was considered to be slightly retarded for his age. The most prominent clinical feature was a symmetrical thickening of the distal portion of the finger- and toenails, with moderate uplifting (Fig. 1). Yellowish discoloration was not marked, and there was no plantar keratosis, leukokeratosis or hyperhidrosis. His external genitalia were normal, and both testicles were descended. He also had bilateral ptosis which did not respond to edrophonium injection. His mother, elder brother and sister also presented ptosis, without showing any changes in the nails. Otherwise physical examination showed nothing in particular. Radiologic examination of the skull, brain CT scan, and routine laboratory studies were all negative. The patient was diagnosed as having pachyonychia congenita and congenital ptosis.

Chromosomal analysis of peripheral blood leukocytes revealed deletion of the long arm of Y (46, XYq-) (Fig. 2). Studies were not performed on blood from his sibs or parents.

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Fig. 1: Characteristic thickening of the nails of the patient.

Discussion

Pachyonychia congenita is a rare congenital ectodermal dysplasia, which is characterized by hyperkeratotic lesions of the nail. Other manifestations include leukokeratosis, keratosis palmoplantaris, follicular keratosis, hair abnormalities and hyperhidrosis [6]. Although our patient did not show diverse symptoms of the disease, the diagnosis was evident by the characteristic changes of the nails.

Since the patient was mildly floppy, and showed poor weight gain and mild developmental retardation, chromosome study was performed. Unexpectedly, there was a deletion of the long arm of the Y chromosome (46, XYq-). The clinical features associated with this chromosomal aberration range from normal development to various features such as short stature, hypospadias, oligo- or azoospermia, muscular dystrophy and mental retardation [2-5]. Since the external genitalia of the patient were normal, it can be assumed that expression of the male phenotype may be achieved even if the long arm of the Y chromosome is not complete.

The present case is characterized by the coexistence of pachyonychia congenita, congenital ptosis and deletion of the long arm of the Y chromosome. The patient also showed insufficient weight gain, which may be attributed to the chromosome abnormality. There are several reports which describe uncommon symptoms of Yq deletion. Association of absent Yq and short nails is reported by Meisner and Inhorn [3], although the characteristic is not explained in detail. Fitch et al described a male patient with deletion of the long arm of the Y chromosome who had ptosis [5]. Since bilateral ptosis was observed in sibs of our patient, there might be the same chromosomal abnormalities in those family members. Unfortunately, chromosomal analysis was not performed due to refusal.

It is difficult to explain how this change in karyotype, pachyonychia congenita and congenital ptosis occurred in the same patient. To our knowledge, no case of the association of these conditions has been reported.



Fig. 2: G-banded karyotype of the patient (46, XYq-).

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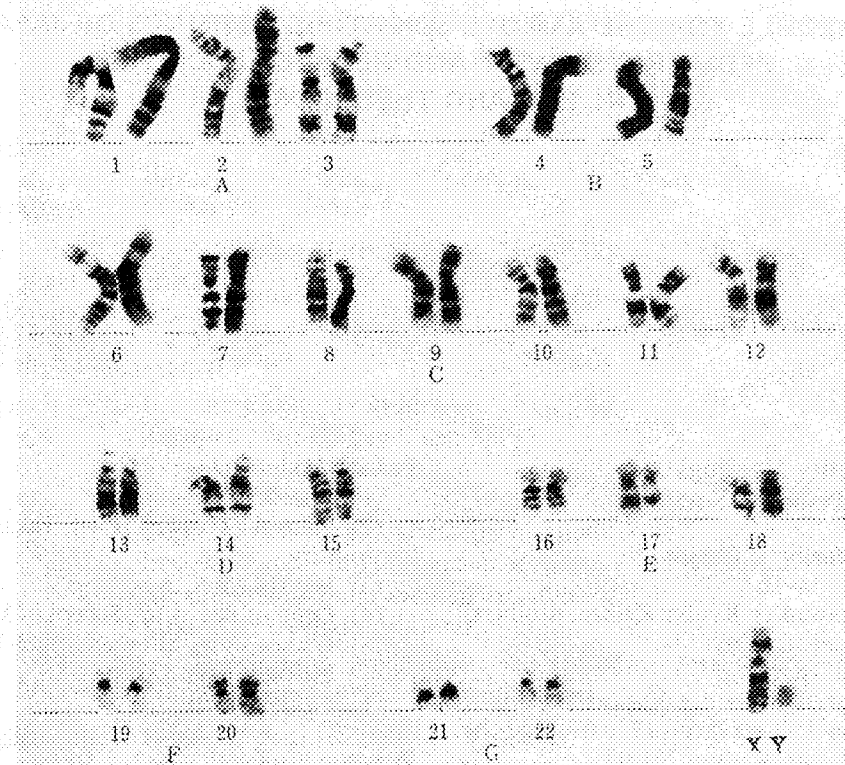


Fig. 2: G-banded karyotype of the patient, showing deletion of the long arm of the Y chromosome (46, XYq-).

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