



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.

MULTIPLE CONGENITAL ANOMALIES

A DIAGNOSTIC
COMPENDIUM

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to cutis verticis gyrata, on the eyelids (leading to ptosis), and on the hands. Finger clubbing is caused by soft tissue hyperplasia. The main clinical problem is usually hyperhidrosis.

Features

- Acne
- Clubbing of fingers (drumstick)
- Coarse facial features
- Cortical hyperostosis/thickening
- Cutis gyrata of scalp
- Hyperconvex/clubbed nails
- Hyperhidrosis
- Hyperkeratosis
- Keloids
- Large nose
- Papules
- Ptosis of eyelids
- Subperiosteal new bone formation
- Thick skin
- Thickened nails

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Venande PY, Boffa GA, Delmas PD et al. Pachydermopentostosis with gastric hypertyrophy, anemia, and increased serum bone Gla-protein levels. Arch Dermatol 1988; 124: 1831-1834.

Pachyonychia congenita

Inheritance Autosomal dominant

Abstract This condition may be heterogeneous. Many cases manifest with pachyonychia congenita (opaque, thickened nails), palmo-plantar hyperkeratosis and hyperhidrosis, oral leukoplakia and follicular hyperkeratosis. In the so-called Jadassohn-Lewandowsky type the above lesions breed true. In the Jackson-Lawler type oral leukoplakia is not present, but neonatal teeth and epidermoid cysts can be a feature.

Features

- Cataract
- Hyperhidrosis
- Hyperkeratosis
- Leukoplakia
- Neonatal teeth
- Other tumours of skin
- Papules
- Skin cysts
- Sparse hair/aloppecia areata
- Thickened nails

References

Besser FS, Moynahan EJ. Pachyonychia congenita with epidermal cysts and teeth at birth: 4th generation. Br J Dermatol 1971; 84: 95.

Inheritance Uncertain

Abstract Two unrelated boys were reported with septo-optic dysplasia, developmental delay, short stature and symmetric defects of the hands that were similar to amniotic band dysplasia. The hand anomalies consisted of absence or hypoplasia of middle or distal phalanges of fingers 2, 3, 4 and the great toe. There were also constrictions and syndactyly.

Features

- Absent fingers or oligodactyly
- Absent nails
- Absent phalanges
- Constriction rings of arms
- Constriction rings of digits
- Hypoplasia/dysplasia of optic nerve
- Hypoplastic phalanges
- Mental retardation
- Nystagmus
- Septum pellucidum/falx, abnormal
- Short stature, proportionate
- Skin syndactyly of fingers
- Small/hypoplastic/deepset nails

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