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.

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Pachyonychia Congenita

(A Case Report)

By

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SUMMARY

A rare case of pachyonychia congenita syndrome is reported and the relevant literature reviewed.

INTRODUCTION

Pachyonychia congenita is a rare dominantly inherited ectodermal dysplasia, first described by Jadassohn and Lewandowsky in 1906.[5] It is associated with other hereditary dyskeratoses like palmar and plantar callosities, bullae, keratosis pilaris, icthyosis, hyperhydrosis, leukokeratosis oris and steacystoma multiplex.[10] Murray[8] first noticed the association of neonatal teeth and pachyonychia congenita. Association of the above two with steacystoma is still rarer. Diffuse polyposis of the gastrointestinal tract with ectodermal changes of nails and skin have been reported.[7]

CASE REPORT

N.R., a twelve year old boy, born of a non-consanguinous marriage, was brought with the history of horny growths of the nails and on the palms and soles since birth. There was history of recurrent shedding of nails. He also complained of photophobia, redness of eyes and diminished vision over the past three years. There was no history of neonatal teeth or early loss of teeth. There was no similar family history.

On examination, he had microcephaly, madarosis, keratitis, corneal opacities, dendritic ulcers in both eyes, horizontal nystagmus, dental malocclusion, pigmentation on the tongue, horny hyperkeratotic lesions on the nails, hyperkeratotic erosive patches on the palms and soles, (See Figs. 1 and 2 on page 188B), crazy pavement dermatoses and dry sparse hair. Systemic examination did not reveal any abnormality.

The investigations done were as follows: intelligence quotient was between 60 and 70. Exact intelligence quotient could not be assessed because of poor vision and lack of education. Plasma and urinary amino-acidogram showed a normal pattern. Roentgenograms of the hands and feet did not reveal brachyphalangy or any other abnormality.

DISCUSSION

In pachyonychia congenita, the principal manifestation is hyperkeratotic lesions of the nails and palms and soles.[4] At birth, the nail may appear normal, but shortly thereafter, a yellow brown discolouration of the nail bed and an uplifting of the nail is seen.[4] These changes progress until the nail bed is elevated and eventually the anterior half of the nail becomes hypoplastic or even absent. Skin changes include patchy to complete hyperkeratosis of palms and soles, callosities of feet that blister easily and keratosis pilaris with tiny cutaneous horny excrescences. Epidermal cysts

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filled with loose keratin are often seen on the face, neck and upper chest. Leukokeratosis of the mouth and tongue are also known. Occasional abnormalities include mental deficiency,[3] corneal thickening, cataracts, thickening of tympanic membrane, hyperhydrosis, dry sparse hair, osteomata and intestinal diverticula.[6]

This condition is known to be autosomal dominant with variability of expression.[9] The basic mechanism is unknown, however vacuolisation of the cytoplasm of nail matrix cells may be significant and needs to be studied further.[9]

The fully developed syndrome is seldom mistaken for anything else. The differential diagnosis includes hereditary onychogryposis, which is also inherited as a dominant trait, but there are no abnormalities other than the nail changes. In epidermolysis bullosa, blister formation may be associated with nail thickening, but the clinical picture is very different.

Topical application of iodochlorhydroxyquin powder (3%) in chloroform resulted in excellent control of pachyonychia in one patient.[5] Surgical removal of the nails have been reported to be useful in some cases.[4,9] However, any matrix left behind will reform abnormal nails.[1,2,9] Amputation of the entire distal phalanx is the only way to free the fingers of distorted, dystrophic nails and make them useful.[11]

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REFERENCES

1. Andrew, G. C.: Pachonychia congenita, *Arch. Dermatol. Syph.*, 32: 501-502, 1935.
2. Andrew, G. C.: Pachonychia congenita, *Arch. Dermatol. Syph.*, 33: 183-184, 1936.
3. Carakushansky, G.: Pachonychia congenita. In, "Genetic and Malformation Syndromes in Clinical Medicine," Editors: W. L. Nyhan and N. O. Sakati. Year Book Medical Publishers Inc., Chicago, 1976, pp. 225-227.
4. Heenan, P. J. and Quirk, C. J.: Transient acantholytic dermatosis (Grover's disease) in pachonychia congenita. In, "Dermatology in General Medicine. Textbook and Atlas", Editors: T. B. Fitzpatrick, A. Z. Eisen, K. Wolff, I. M. Freedberg, K. F. Austin. McGrow-Hill Book Company, New York, 1987, pp. 526-533.
5. Jadassohn, J. and Lewandowsky, F.: Pachonychia congenita keratosis disseminata circumscripta (folliculosis): tylomata; leukokeratosis linguae. *Iconographia Dermatol. Tab.*, 629, 1906. Quoted by Smith and Jones.[9]
6. Joseph, H. L.: Pachonychia congenita. *Arch. Dermatol.* 90: 594-603, 1964.
7. Manousos, O. and Webster, C. U.: Diffuse gastro-intestinal polyposis with ectodermal changes. Gut, 7: 375-379, 1966.
8. Murray, F.A.: Congenital anomalies of nails, (associated with erupted teeth at birth). *Brit. J. Dermatol.*, 33: 409-412, 1921.
9. Smith, D. M. and Jone, K. L.: Pachonychia congenita. In, "Recognizable Patterns Human Malformations," 3rd Edition, W. B. Sounders Company, Philadelphia, 1982, pp. 406-407.
10. Soderquist, N. A. and Reed. W. B.: Pachonychia congenita with epidermal cysts and other congenital dyskeratoses. *Arch. Dermatol.*, 97: 31-33, 1968.
11. Wright, C. S. and Guequierre, J. P.: Pachonychia congenita. Report of two cases with studies on therapy. *Arch. Dermatol. Syph.*, 55: 819-827, 1947.