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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.
This young girl has shown keratotic growths since infancy. Manifested in the first few weeks of life as subungual thickening, this hyperkeratosis later became evident as discrete acuminate horn excrescences on the knees and elbows. After she began to walk, sharply circumscribed thick calluses and corns with underlying deep bullae appeared and have persisted precisely at the pressure points on her soles. At present the buttocks, posterior thighs and palms show numerous follicular hyperkeratoses and cutaneous horns. Lentokeratotic changes are present on the palate and lower lip. She also exhibits hyperhidrosis of the palms and soles.

She is an only child and there is no history of consanguinity nor any nail or skin problems in the past three generations.

Pachyonychia Congenita

This child was born with the gene for claws and horns! An extremely rare event, it has been recorded in the world’s medical annals less than a hundred times. This is pachyonychia congenita, a genodermatosis. It is one in which the inheritance pathway can be singularly well studied, since the gene marker of thickened nails is fully apparent to all. And the pathway has proved to be an autosomal dominant one, traced in some instances for over six generations. In our patient, however, the syndrome has sprung full blown, de novo, and we assume a mysterious mutation has occurred.

What can we make of this hoof, nail and follicle disorder? The unifying theme is hyperkeratinization in response to physical stress. This is a pressure keratoderma, named for the first site to be affected, the nail bed. This is a disease of the keratinocyte, the major cell of the epidermis, responsible for the elaboration of the protective and insoluble surface protein, keratin. It is one of controlled yet superabundant production of stratum corneum. Clinically, it is initiated in the crib with simple acts such as scratching. The nail plate pull serves as a stimulus to the hyponychium to overproduce keratin. The result is a nail plate lifted high by subungual keratin. As might be expected, all of 20 nails are involved. Secondly, the nail plate becomes deformed, warped, twisted...
or abnormal as the keratin mass exerts an effect on the nail matrix. This is the thickened nail of pachyonychia - a very precise change, not just a thick nail.

As our infant grows, new points of trauma behave in an identical fashion, producing minarets of keratin at focal sites of follicular trauma. These are the giant magnified versions of keratosis pilaris, i.e., cutaneous horns. Even the leukokeratoses of the mucosa record the tendency to keratinize excessively. In early infancy these may be confused with thrush until the total picture is realized, and the scrapings for mycelia are repeatedly unsuccessful. We should be aware, incidentally, that pachyonychia congenita is a mucocutaneous genodermatosis inasmuch as the nasal, laryngeal, corneal, vaginal and rectal mucosa may be similarly involved. Probably the fact that some of these affected infants are born with a few fully erupted teeth is also relevant to an altered keratinizing potential.

The most dramatic change, however, comes with walking. The consequent pressure on relatively few plantar sites induces enormous calluses of keratin on the soles. Understandably, these can literally shear away the epidermal attachments to the dermis when she runs. It is this that is responsible for the sublesional bullae of the soles. These are not the lesions of epidermolysis bullosa despite a superficial resemblance. The calluses and corns of her feet reflect the intermittent pressures exerted there and not a local defect, since skin from elsewhere grafted to the sole has been shown to produce the same massive keratin growths. Her entire epidermis has, as it were, the capabilities of producing "keloids" of keratin.

A great deal can be done to help this child, although we can do nothing to reverse the mutant gene or to prevent its transmission to any children she might have. We can help because we know that her problem is triggered by pressure and physical trauma. It cannot be overemphasized that the keratin growths serve as a vicious self-perpetuating mechanism. The rubbing of any keratotic excrescence or simply sitting on a cutaneous horn provides a selective stimulus for even greater keratinocyte activity. The little become big, and the big grow larger. This self-amplification of lesions is nowhere better shown than on the feet, once localized thickenings occur at the normal pressure points, the entire body weight is virtually transferred to these alone, with progressive keratodermal growth and inevitable consequence. Her feet need our initial attention to break this vicious cycle. Moreover, the bullae which have followed are a source of her subsequent symptoms, which range from mild dactylytis to skin grains, and to distribute the weight better, the keratin as a part of good management.

The keratodermas of the retinoids, which are the only topical agents currently approved for use in the treatment of keratoderma, are also valuable for the athetoidal areas, is also valuable for the athetoidal areas, is also valuable for keratoacanthomas. A liquid preparation 15 parts propylene glycol, 2 parts propylene glycol acetate and 83 parts propylene glycol applied under Saran Wrap. This aids in debulking the thickened nails can be treated similarly by fashioning the nails after prolonged use of pumice and clippers. We are reminded of this syndrome at the turn of the century with Reinke's description of hyperkeratosis of the nails of hands and feet.

A few patients may require the use of a skin matrix and nail bed to achieve even greater cosmetic and functional result. Their failure of their fingertips as a waxing and waning affair, and at times infected nails, can be treated with the sites of bullae on the bones.

Be aware of the fact that pachyonychia congenita is a neurocutaneous one. It is associated with neurocutaneous ones. Coloratofibrofatty well as other neurocutaneous ones. Coloratofibrofatty well as other cutaneous conditions.


World literature describes pachyonychia congenita as well as other neurocutaneous conditions.

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followed are a source of painful disability and, because of her hyperhidrosis, can easily become infected. Special shoes should be made for her which have a latex molded sole to reduce pressure on the calloused areas and to distribute the weight more evenly. Regular shaving of excess keratin as a part of good podiatric care is essential.

The keratinous horns elsewhere can be suppressed by topical retinoic acid (Retin-A) applied daily. Salicylic acid, if used in restricted areas, is also valuable for removal of the horn growths. We find that a liquid preparation 15 parts salicylic acid, 80 parts ethyl alcohol and 5 parts propylene glycol is also well tolerated and effective, especially if applied under Saran Wrap. The feet may be safely treated each night with 60 per cent propylene glycol in water, likewise applied under Saran Wrap. This aids in debridement of the excess keratin. The thickened nails can be treated similarly but are less responsive. Regular cutting of the nails after prolonged soaking is best done with large professional nail clippers. We are reminded by this that Jadassohn, who first described this syndrome at the turn of the century, had to use a hammer and chisel on the hardened nails of his patient.

A few patients may require plastic surgical excision of the entire matrix and nail bed to achieve permanent total removal of the nail. This is certainly preferable to yielding to the patient who pleads for amputation of his finger tips as a way to be rid of his grotesque, misshapen, painful and at times infected nails. Others who develop massive chronic ulcers at the sites of bullae on their feet may require grafting.

Be aware of the fact that, although this is a birth defect, it is not a neurocutaneous one. It does not involve the melanocytes and hence is not associated with neurologic problems or mental retardation. Also, it should be recognized that the lesions are not premalignant, nor even the leukokeratotic ones. Confusion has arisen in this regard, since the diffuse keratodermas of the palms and soles may be associated with mucosal carcinoma, particularly in the esophagus. Pachyonychia congenita is a local punctate keratoderma and is not premalignant.

Pachyonychia congenita is a remarkable experiment of nature from which we should learn much more about the keratinocyte and how it responds to physical forces. For this is truly a piezo-keratoderma.


World literature describes only 92 cases of pachyonychia congenita: pachyonychia in
90 of these: keratoderma of sole, 67, and of palm, 44: oral leukokeratosis, 50: bullae, 35; hyperhidrosis, 22; sparse thin hair, 11; premature dentition, 11; granulosis rubra nasii, 4.


Not to be confused with pachyonychia congenita; develops late in childhood, involves only a few digits, skin and hair normal.


Ten cases of hyperkeratosis of hands and feet; good results with 10 per cent urea cream.


Four cases of keratosis palmaris et plantaris responding to 60 per cent propylene glycol in water, under Saran Wrap occlusion.


Thyroid therapy associated with temporary alleviation.


Vitamin A in large dosage helpful.


May have to totally remove nail bed and matrix; graft plantar ulcers.


How to remove a nail—permanently.