Pachyonychia Congenita Project

15 March 2005

Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

> Sec. 107. - Limitations on exclusive rights: Fair use
> Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, **teaching (including multiple copies for classroom use), scholarship, or research**, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

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.

2386 East Heritage Way, Suite B, Salt Lake City, Utah 84109 USA
Phone +1-877-628-7300 • Email—Info@PC-Project.org
www.PC-Project.org

), necessitating amputation of the upper
of slowly progressive ulcerated cutaneous
igin, for which the name malignant histio-
nployed by the French school. Montpellier
l tumour of the female breast under that
ıeir case differs somewhat from ours. The
uibert and Marre (1942) and for Dupont's
.as, which showed a good deal of xantho-

large pale stellate cells with one or more
ıcleoli frequently combined with or even
ımmatory cells does not, *per se*, help in
ttern may be seen in various diseases. A
ient of Pautrier and Woringer (1937), who
esions and who was cured of his granulo-
llowed reappearance of the lesions, which
ern and gave the impression of a lympho-
ıtrier and Woringer classify the large cells
ɜmohistioblast of Ferrata.
mely Letterer-Siwe disease, may be pre-
n with a microscopic picture not too dis-
lohistiocytoma (Case 7 of Ponseti, 1948).
n are the cases of chronic irritation afte-
8), among others, drew attention. In this
ical features resembling our material, yet
)le. The cases of cutaneous reticulosis of
), whose slides I had occasion to see,* and
ɜ (1949) and of Cobane (1950) are different
self-evident that the clinical findings (size
be evaluated before microscopic diagnosis

JMMARY.

is used for a dermato-histological entity
oticed. It is felt that the cutaneous
and O'Leary represents the same process.
ɜtological appearance are presented, as well
me aspects and may well represent a pro-
f low grade malignancy. The differential
ɜs are discussed.

n and Dr. Haber of London for their co-operation.

References.

ALLEN, A. C. (1948) *Arch. Derm. Syph., Chicago*, **57**, 19.
——— (1948) *Amer. J. Path.*, **24**, 367.
AMICIS, A. DE (1938) *Rinasc. med.*, **15**, 119.
CALDARERA, E. (1934) *Riv. sanit. sicil.*, **22**, 1532.
COBANE, J. H., STRAITH, C. L. and PINKUS, H. (1950) *Arch. Derm. Syph., Chicago*, **61**, 442.
DUPONT, A. (1933) *Bull. Soc. franç. Derm. Syph.*, **40**, 674.
——— (1939) *Bull. Ass. franç. Cancer*, **28**, 456.
GOUGEROT, H., and DREYFUS, B. (1937) *Bull. Soc. franç. Derm. Syph.*, **44**, 1699.
GUIBERT, H. L., and MARRE, P. (1942) *Bull. Ass. franç. Cancer*, **30**, 52.
MONTGOMERY, H., and O'LEARY, P. A. (1934) *Arch. Derm. Syph., Chicago*, **29**, 26.
MONTPELLIER, J., LAFFARGUE, P., and NICOLAI, R. (1938) *Bull. Ass. franç. Cancer*, **27**, 784.
PAUTRIER, L. M., and WORINGER, F. (1937) *Bull. Soc. franç. Derm. Syph.*, **44**, 1549.
PINKUS, H., *et al.* (1949) *Amer. J. Dis. Child.*, **77**, 503.
PONSETI, I. (1948) *J. Bone jt. Surg.*, **30A**, 811.
RENTIERS, P. L., and MONTGOMERY, H. (1949) *Arch. Derm. Syph., Chicago*, **59**, 568.
ROBB-SMITH, A. H. T. (1944) *Brit. J. Derm. Syph.*, **56**, 151.
TILLMAN, W. G. (1949) *ibid.*, **61**, 179.
TOBIAS, N. (1949) *J. invest. Derm.*, **12**, 255.
VOLTERRA, M. (1928) *Riv. clin. Med.*, **29**, 447.
WIGLEY, J. E. M. (1949), *Proc. R. Soc. Med.* (Sect. Derm.), **42**, 139.
WINER, L. H., and STRAKOSCH, E. A. (1941) *J. invest. Derm.*, **4**, 249.

ITAMIN A IN A CASE OF ACQUIRED LOCALIZED KERATOSIS PALMARIS ET PLANTARIS AND ONE OF ACQUIRED PACHYONYCHIA.

DR. A. D. PORTER AND DR. H. HABER.

THE difficulty of treating these disorders successfully makes us think it worth while to record satisfactory results in one case of each treated with vitamin A.

These two cases are reported together because both are essentially manifestations of abnormal keratinization, both responded to vitamin A, and both were under observation at the same time.

CASE 1.—A female dressmaker, aged 19, complained of skin trouble all her life on the palms and soles.

Past history.—She had never suffered from any illness that she could remember, apart from colds.

Family history.—This was entirely negative, except that her father's skin had always been dry.

Examination revealed the presence of well-marked keratosis palmaris et plantaris. On the soles the horny layer was much thickened and was surrounded by an erythematous margin. The whole surface of the palms was erythematous, but hyperkeratosis was less marked. Sweating on the affected areas was excessive. Mild xerodermia was also present, with slight keratosis pilaris on the outer surface of the arms. The W.R. and Kahn reactions were negative, and no fungus was detected microscopically. Plasma vitamin A averaged 88 i.u./100 ml.

Treatment.—Ointments containing up to 6% salicylic acid were used for many months without benefit. Higher concentrations appeared to irritate the skin. Tar and other substances were also applied without altering the condition.

While under active treatment numerous hyperkeratotic nodules resembling large

plantar warts appeared on the palms (Fig. 1), which were entirely unaffected by external applications.

It was finally decided to test the effect of vitamin A, and 100,000 i.u. daily was ordered. After three months it was found that the hyperkeratotic nodules and

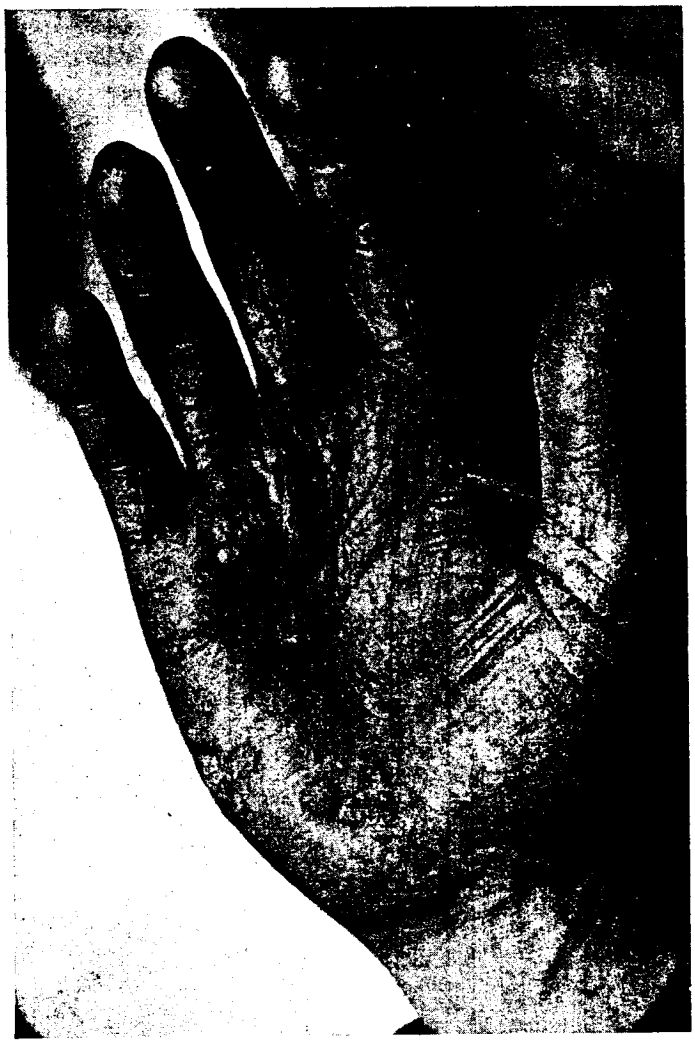

FIG. 1.

slight hyperkeratosis had entirely disappeared, and that the palms were covered with soft erythematous sweating skin. The soles remained as before. The vitamin A was increased to 200,000 i.u. daily for three months and then stopped. Six months later there had been no recurrence of hyperkeratosis on the palms, while the skin of the soles was softer.

CASE 2.—A girl, aged 9, complained of thickening and deformity of the toe nails and bluish discoloration of the margins of the finger nails.

Past history.—Pneumonia, chickenpox, mea

Family history.—Mother has thickened ski

Examination showed a healthy girl with from the toe-nails and thickened skin over pr The distal ends of all the toe-nails were thi and raised from the nail bed. On the fourth "claw" formation (Fig. 2). No sign of fung fungus or monilia were found microscopicall

Treatment.—Vitamin B, 1 mg. *t.d.s.*, was Vitamin A, 30,000 i.u. daily, was then added

FIG. 2.

month later; within two months improveme the nails had become normal, excepting the f showed slight hyperkeratoses. The thickened unaffected.

COMMEN

It is, of course, possible that both of th without vitamin A, and that the improveme It is known that localized hyperkeratosis the first time late in life, may respond scribed by Brooke (1891). In Case 1, had resisted five years' treatment by ou

(Fig. 1), which were entirely unaffected by

fect of vitamin A, and 100,000 i.u. daily was found that the hyperkeratotic nodules and

FIG. 1.

appeared, and that the palms were covered The soles remained as before. The vitamin r three months and then stopped. Six month yperkeratosis on the palms, while the skin

l of thickening and deformity of the toe nail s of the finger nails.

Past history.—Pneumonia, chickenpox, measles and mumps.
Family history.—Mother has thickened skin over pressure areas on the soles.
Examination showed a healthy girl with no ectodermal or other defects apart m the toe-nails and thickened skin over pressure areas on the soles.
The distal ends of all the toe-nails were thickened, deformed, of a yellowish colour l raised from the nail bed. On the fourth and fifth toes there was a tendency to law" formation (Fig. 2). No sign of fungus infection was seen clinically, and no ngus or monilia were found microscopically.
Treatment.—Vitamin B, 1 mg. *t.d.s.*, was tried for one month without effect. amin A, 30,000 i.u. daily, was then added, and increased to 75,000 i.u. daily one

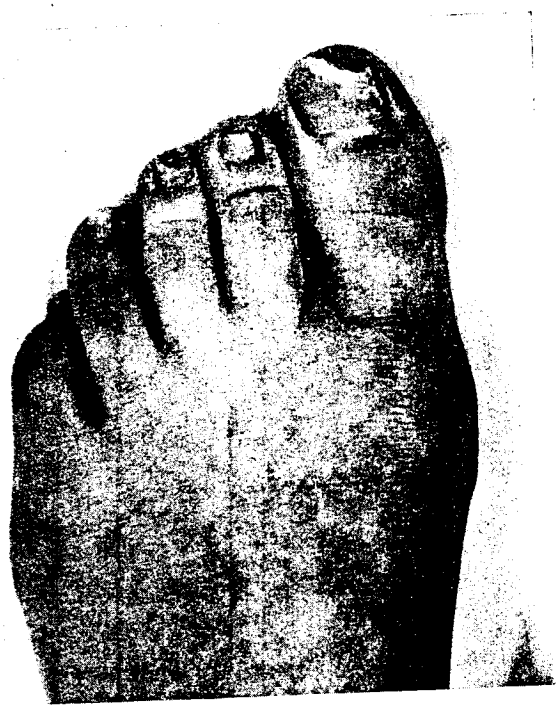

FIG. 2.

nth later; within two months improvement was noted, and after four months nails had become normal, excepting the free margin of the right big toe, which wed slight hyperkeratoses. The thickened skin on the soles, however, remained affected.

COMMENT.

It is, of course, possible that both of these patients would have got better thout vitamin A, and that the improvement while taking it was a coincidence. is known that localized hyperkeratosis on the palms, when appearing for first time late in life, may respond to simple remedies, as in the case scribed by Brooke (1891). In Case 1, however, the nodules on the palms d resisted five years' treatment by ourselves and others with keratolytic

and other ointments, and the malady had been present on the feet, and on the palms in a modified form, since birth. Although the palmar lesions which responded to vitamin A were of recent origin, the disorder was clearly of the congenital type, which is generally resistant to any kind of therapy. Marre (1949), however, gave vitamin A to a mother and children with congenital tylosis and noted that the palms became softer, though there is no mention of the hyperkeratosis disappearing.

The second case was probably one of pachyonychia congenita, although we have referred to it as " acquired " because the nail changes began later than usual. Wright and Guequierre (1947), in describing two cases of this nature, say that they found vitamin A and emollients of value in treating the dry keratotic skin in an infant, but apparently the nails were unaffected.

It is noticeable that the hyperkeratotic nodules in Case 1 and the nail changes in Case 2 were of comparatively recent origin and short duration for such disorders, and were actively progressive. Possibly these factors may help to account for the response to vitamin A.

It is not suggested that deficiency of vitamin A is an aetiological factor of importance in tylosis, but it is possible that the malady so disturbs the utilization of vitamin A that the process of keratinization is affected.

SUMMARY.

The beneficial effect of vitamin A in a case of keratosis palmaris et plantaris and in one of pachyonychia is described.

We wish to thank Dr. R. T. Brain for permission to publish the second case.

REFERENCES.

BROOKE, H. G. (1891) *Brit. J. Derm.*, **3**, 335.
MARRE, I. R. (1949) *Ibid.*, **61**, 144.
WRIGHT, C. S., and GUEQUIERRE, J. P. (1947) *Arch. Derm. Syph., Chicago*, **55**, 819.

LEPRA, PSORIASIS, OR THE WILLAN-PLUMBE SYNDROME?

BRIAN RUSSELL, M.D., M.R.C.P.

THE ancient history of the condition known to-day as " psoriasis " has been obscured by the unfortunate terminological confusion that arose not only between the Greek, Latin and Arabic schools of medicine, but also between succeeding generations of physicians. The precise detailed description of the management of the " rising, scab or bright spot " in Leviticus XIII shows the age-long difficulty of differential diagnosis between various skin diseases, from the serious to the banal. Biblical leprosy (Zaraath), of which Naaman was healed by dipping himself seven times in Jordan, was probably the same

as our psoriasis, and the term " leprosy " used by Bateman in 1817 for the first diseases, an eruption apparently identical Confusion also for long existed owing (λεπρα = scaly) for a large number of ' macular leprosy, psoriasis, squamous ec (ψωρα = the itch) was apparently applied psora ") and various forms of pityriasis (" rough or leprous psora "), and bore les did lepra. Paulus Aegineta, early in th disorders as " characterized by a roughn deliquescence of the body which originate lepra affects the skin deeply in circular pat of large fishes ; whereas psora is more s throws off bran-like substances." Anothe term αλφος (= white), which was apparen also came to be applied to vitiligo and m of leprosy. Hippocrates, however, rega blemish, and probably referred to lepro distemper of the most fatal kind." The scaliness) was applied to a slowly develo of children, with very white scales, and n with depigmentation. Lepra nigricans of cold, damp, and a precarious or improp it as " generally believed to be induced the system."

In Green's *Diseases of the Skin* a table mous with alphos, vitiligo, morphoea, l Greek elephantiasis ! When psoriasis is melas, ecthyma, psora leprosa, pityriasis (= lichen), terminological " confusion no

Psoriasis.

Robert Willan (1808) gave the first clea to-day, under the title " lepra vulgaris has a more distinct form and character He adopted " psoriasis," " which seems i scaly psora by a different appellation," w psora from the ulcerated psora, placed b The eleven types of scaly tetters he included psoriasis guttata, psoriasis gy correspond to those conditions in our ter