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 The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.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.

Familial Lupus: Spectrum of Disease in One Family

To the Editor.—In a recent study on lupus erythematosus (LE), Prystowsky and Gilliam separated their patients into the following three groups: a group of patients with LE limited to the skin; a group with active discoid lesions plus visceroi involvement; and a group with proliferative glomerulonephritis. We found this particularly interesting for we were observing at that time a case of familial lupus in which the three probands corresponded to these three distinct groupings.

Report of Cases.—Case 1.—A 25-year-old woman was seen in August 1975, with atrophic, tender, hypopigmented areas on her palms and soles, breasts, thighs, and scalp that were accompanied by arthralgia, morning stiffness, recent weight loss, and Raynaud phenomenon. The patient appeared ill and was hospitalized. A test for antinuclear antibody (ANA) was positive in titer of 1:1280 and was reported as 3+ shaggy. The anti-DNA was less than 1:10. Lupus erythematosus cell preparation was strongly positive. Rheumatoid factor was negative. The C3H0 was 50 units (normal, 25 to 57 units); C3 was 100 mg/100 ml (normal, 125 to 175 mg/100 ml), and the C4 was 23 mg/100 ml (normal, 20 to 50 mg/100 ml). Urine reaction for protein was 2+ and the 24-hour urine protein was 735 mg/100 ml. A skin biopsy specimen was taken that was typical of LE. HLA type 2,9,7,17. The EMA was undetectable. Severe pleuritic chest pain developed during the patient's hospitalization which was diagnosed as lupus pleuritis. She responded to bed rest and salicylate therapy.

Case 2.—This 34-year-old woman, the sister of patient 1, had typical discoid lesions on her scalp, ears, fingers, toes, abdomen, and nostrils, which on biopsy were interpreted as discoid LE. The results of all laboratory studies including the ANA, LE preparation, and complement were either normal or negative. HLA type was 2,9,7,17. The EMA was undetectable.

Case 3.—A 15-year-old female adolescent (another sister) was admitted to the hospital in January 1968, because of a one-week history of generalized edema. Physical examination revealed a cachectic appearing girl with 1+ generalized edema. The patient was given intravenous and a renal biopsy was performed that demonstrated lupus nephritis. Despite high-dose steroid therapy, her condition steadily deteriorated. Pleural effusion, pneumonia, and renal failure developed and she died in March 1968. An autopsy consent was not obtained, but the laboratory studies shortly before her death revealed the following: hemoglobin level, 6.4 mg/100 ml; urea nitrogen level, 97 mg/100 ml; creatinine clearance, 34 mg/100 ml; 24-hour urine protein level, 6 mg/100 ml; and ANA, 4+. HLA type was not available.

Comment.—Apart from adding another report of familial lupus to the growing literature of familial occurrence of LE,2 we thought that these cases were interesting because the entire spectrum of LE is demonstrated in one family whereas other familial reports tend to demonstrate a familial systemic or familial discoid lupus.2, 3

The mother and father were deceased; however, three other siblings were studied extensively. There was no clinical or laboratory evidence of LE. Because of the reported association of HLA A1 and HL-A8 with systemic lupus, HLA typing was done on all available family members. HLA types of the involved siblings were already presented. The following types were noted in the uninvolved siblings: HLA type 2,9,14,17; HLA type 2,9,17; and HLA type 2,9,17.

S. BARSKY, MD
D. KNAPP, MD
B. BENNIN, MD
Chicago


Pachychonia Congenita With Recessive Inheritance

To the Editor.—Pachychonia congenita (PC), a rare genodermatosis, to our knowledge was first described by Jadassohn and Lewandowsky in 1906. It is characterized by the typically thin or dystrophic nails, keratoderma of the palms and soles, hyperhidrosis, plantar blisters, keratosis pilaris, leukokaryotaxis or ichthyosis, steato-cytoma multiplex, epidemial cysts, and other features of dyskeratosis such as corneal dystrophy, cataract, hypoplastic hair, and changes in the teeth, nose, ears, and mucous membranes.

A majority of the cases were reported from Europe and America, and studies from several large families have demonstrated that PC is transmitted as a simple mendelian dominant gene with incomplete penetrance.1, 4-6 Asian cases were rarely reported, and the disease, as far as we are aware, has not been known to be inherited as an autosomal recessive trait.

Recently, an Indian girl from Western Malaysia was found to have PC, but the disease appeared to be a recessive disorder, as she was the product of a consanguious marriage. The inheritance of this interesting patient is briefly discussed.

Report of a Case.—A 4-year-old Indian girl from Jodhpur, Western Rajasthan, was recently seen at our clinic because of her abnormal nails since infancy. Several months after birth, her parents observed that the nails of all her toes and fingers were grossly thickened, distorted, and broke easily at the free edge. Frequent clippings did not prevent this reappearance. The nail beds were readily infected after minor trauma, and occasionally, the whole nail was shed to be replaced by another abnormal nail. She had a transient erythematous rash at the age of 6 months, and was suffering from recurrent "boils" and "dandruff". There was no residual pigmentation or hair loss. Her palms and fingers were further, and when she started to walk at approximately one year of age, small blisters and peeling of the skin appeared on the sides of the toes and feet. A year later, small, discrete calluses developed on the sides of the feet and the plantar surface of the toes. About two years ago, her parents noted that small horny lesions developed on both sides of her knees, her elbows, and one buttock. These caused mild limitation of motion, and she had no other significant symptoms. Her birth and milestones were normal and her physical and mental development, too, were normal. Physical examination showed that she was normal for her race, sex, and age. There were no systemic abnormalities, and the significant findings were confined to the nails, skin, and oral mucosa.

Nail Lesions.—All the nails of the
fingers and toes were markedly hyperkeratotic, deformed, and dystrophic. The affected nails appeared “pinched up,” with narrowing of the lateral nail edges and upward and outward growth of the free edges. Although the nail plate was normally attached to the proximal and lateral grooves, the distal part often protruded beyond the tip of the digits and was considerably raised and arched transversely. Because of the brittleness, the free edges were often jagged and cracked, exposing the piling up of hard keratous debris from the nail bed under the raised-up nails. Some nails, however, had the appearance of a claw-like incomplete cylinder or cone, eg, the big toes. Subungual keratosis and scars of previous infections were observed in several fingers and toes. On the sides of the toes and feet, peeling of skin from blisters and hyperhidrosis were found.

Skin Lesions.—Besides blisters and skin peeling, the planter surface of the feet had a number of callosities or keratoderma. These were found on the sites of pressure. Her palms, however, showed mild, diffuse thickening without callus formation. On her knees, elbows, and gluteal surface were numerous follicular keratotic grayish-black papules. The horny cones in the center of these papules fit into the crater-like depressions, and removal of the cones produced spots of slight bleeding. On her forehead were five to six small, asymptomatic, soft cystic lesions, and biopsy of one disclosed an epidermal cyst. The patient had no pigmentation, skin atrophy, skin tumors, or other skin lesions on other parts of the body.

Oral Lesion.—A few small patches of white mucosal lesions (leukokeratosis oris) were found in the buccal mucosa, but consent for biopsy was denied. No mucosal lesion was detected in the nose, throat, tongue, or rectum.

Her mental and physical status were normal, and no abnormalities were detected in her eyes, hair, teeth, or other major body organs.

Investigations.—Results of the hematologist, urinalysis, and biochemical tests were all normal. A blood VDRL test was negative. No roentgenological abnormalities were found in the skull and chest. Microscopic examination of the hair did not yield any abnormal findings.

Family Study.—Four generations in this family were examined but no one was found to have the features of PC. The family members were all born and reared in Western Malaysia. The patient's father (aged 32) and mother (aged 30) were first cousins. They as well as the patient's younger sister (aged 2) did not show any nail, skin, or other lesions of PC. All of them were well and healthy (Figure).

Comment.—The diagnosis of PC in this Indian girl was established by the clinical picture of the classical dystrophic nails, the presence of hyperhidrosis, plantar blisters, callosities of the soles and palms, keratosis pilaris lesions on the knees, elbows, and one buttlock, the epidermal cysts on the forehead, and the leukokeratosis in the buccal mucosa. According to the classification of Kumar and Loos,1 she belongs to the type II category of PC as she did not have the cornal lesions. Other uncommon associations with this disease but not found in this patient are hypoplastic or kinky hair, nail teeth, microphthalmos, cataract, corneal dystrophy, scrotal tongue, intestinal diverticulosis, and leukoplakia of mucosal membrane. The rarity of this disease in Asians is not known. Lack of documentation aside, the nail and skin lesions of PC might often be mislabeled as some common local conditions unless one is aware of the disease and unless a family history is obtained. Most studies in the literature have demonstrated that PC is transmitted as a simple autosomal dominant gene with incomplete penetrance.2-6

The disease transmission in our patient is most likely that of the autosomal recessive trait since she was the product of a consanguineous marriage between two first cousins and both of them and the other siblings were unaffected by the disease. The other family members in the four generations were also not affected. However, PC is a very heterogenous disorder, probably involving multiple genes, and hence the possibility of a spontaneous mutation in this family cannot be entirely ruled out.

TAY CHONG-HAI, FRCGP
Republic of Singapore
K. RAJAGOPALAN DDM, MRCP
Kuala Lumpur, West Malaysia


Erythema Multiforme Resulting From Insecticide Spray

To the Editor.—Recently, we have come across an interesting case, in which erythema multiforme developed in a woman 25 years of age merely a few hours after coming in contact with an insecticide, mephalathrin (Dalfy) (2% fanthion), sprayed for killing bedbugs. She had multiple erythematous papulossquamous iris pattern lesions all over her body, with the greatest amount occurring over the extensor aspects of her legs, hands, chest, back, and abdomen that persisted for 20 days. She was not taking any other drugs, had not had a recent vaccination, and had no allergic reactions to food. She did not have a history of similar lesions. All the investigations were within normal limits, except for mild albuminuria and an increase in the eosinophil value. Skin biopsy specimen taken were consistent with the diagnosis of erythema multiforme. She responded well to antiallergic tablets and vitamin C.

Re-exposure to a provocative dose of metronidazole, multifactorial insecticide yet be...