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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.
megaly, aplastic anemia, and purpura. Because of these observations, some writers have attempted to link dyskeratosis congenita with Fanconi's syndrome. Cases are too few to establish a close relationship between these two syndromes; however, the anemias of the two are similar.

Variations. Costello and Buncke reported a case where they observed a transparent tympanic membrane. This was the only patient in which this change had been noted.

Cases revealing reticular hyperpigmentation of the skin and dystrophy of the nails but no leukoplakia, may be formed fromkses of dyskeratosis congenita unless leukoplakia develops later in life or is found subsequently in other areas not easily detected.

Complications. Malignancy is the complication to be feared and the usual cause of death. Cancers have usually developed on the buccal mucosa, tongue, or rectum. The patient described by Costello and Buncke developed squamous cell carcinoma on the dorsal surface of the left hand with metastases to the axilla.

Inheritance. Dyskeratosis congenita is probably transmitted as a sex-linked recessive characteristic.

Differential Diagnosis. Dyskeratosis congenita most closely resembles dystrophic epidermolysis bullosa from which it may be distinguished by the presence of lesions resembling poikiloderma vasculare atrophicans on the face and neck.

Prognosis. Due to the development of malignancy, patients with dyskeratosis congenita have a decreased life expectancy.

Pathology. Sections show the epidermis to be atrophic with flattening of the rete pegs. The stratum corneum is thickened, and, in the reticulated areas, the cells of the Malpighian layers contain granular aggregates of melanin. The upper portion of the dermis is slightly denser than usual and, in places, contains clusters of melanocytes.

Treatment. Garb and Rubin noted pronounced regression of the oral leukokeratotic patches following injection of testosterone propionate intramuscularly and large doses of brewer's yeast by mouth. Radical surgical or electrosurgical removal of the leukoplakia patches, wherever found, should be performed to prevent the development of cancer.

BIBLIOGRAPHY


Pachyonychia Congenita

Synonym. Pachyonychia ichthyosiformis.

Definition. Pachyonychia congenita is a hereditary dyskeratosis characterized by thickening of the nails in all cases and by inconstant associated involvement of the skin, oral mucosa, hair, and cornea.

Occurrence. The disease is extremely rare. It affects males chiefly and appears to be most prevalent among Slavs and Jews of Slavonic origin. Cases have been reported in the Negro.

Symptoms. The thickening of the nails is the invariable and constant finding in this syndrome. All of the associated findings have occurred in whole or in part in the various cases recorded. The nail changes may be present at birth or appear shortly thereafter. In exceptional instances, the primary manifestations have been thickenings of the palms and soles with subsequent development of pachyonychia.

The pathologic changes in the nails occur
in the distal half to two thirds. The proximal portion of the nail is apt to be smooth, shiny, and attached normally in the proximal and lateral grooves. The distal part of the nail is arched transversely and pushed upward from its bed by hard adherent keratinous material collecting under it so that it forms a 30 to 40 degree angle with the axis
of the phalanx. The thickness of the nail plate at the free edge appears normal in some; in others, the nail is thickened, extremely hard, and protrudes beyond the tip of the digit. The portion of the nail raised from its bed is opaque, lusterless, and yellowish-gray or brown in color. Removal of the nail reveals that much of its apparent thickness is due to the accretion of keratinous material from the anterior half of the nail bed rather than to any extreme change in the nail plate itself.

Plantar and, to a lesser degree, palmar keratoses usually accompany the nail changes. Most commonly there are symmetrical, well-circumscribed, yellowish, thick, round or oval, hyperkeratotic areas over the ball of the foot, head of the fifth metatarsal, on the plantar surface of the heel, and the under surfaces of one or more toes. The intervening skin appears normal. Similar changes occur in the palms. Sometimes the changes are less striking, and discrete, small to large wart-like keratotic lesions appear on the soles, palms, and palmar aspects of the fingers. At the other extreme, the entire plantar surface may be covered with yellowish hyperkeratotic material which in some places may reach a thickness of 2.5 cm. Palmar surfaces may exhibit similar material reaching 1.0 cm. in thickness with linear extension to the distal phalanges of each finger. One patient exhibited a striate form of palmar keratosis. Some patients exhibit characteristic plantar keratoses but the palms remain clear. Most of them will show hyperhidrosis of the palms and soles.

Painful blisters and oozing are other features of this disorder. Sometimes the initial changes on the soles are erythematous patches which develop into blebs and these in turn give rise to calluses. Not only may the clear watery blebs initiate the appearance of hyperkeratotic plaques but they continue to occur on the palms and soles after the calluses have reached their maximum development. There is a predilection for the toes and adjacent parts of the soles, heels, and along the borders of the feet. Exceptionally, blebs occur on the elbows and knees. In many instances, they are more severe during the hot summer months. Although often extremely uncomfortable, the bullae heal without scarring.

Various other cutaneous manifestations occur in pachyonychia congenita. One of the more common is the presence of bright red papules with central horny follicular plugs on the extensor surfaces of the elbows and knees. Keratosis pilaris occurs on the outer aspects of the upper and lower extremities and sometimes on the back, buttocks, and lumbar region. Another annoying feature is the occurrence of verrucous lesions on the knees, elbows, popliteal spaces, buttocks, legs, and ankles. Calluses may occur on the extensor surfaces of the forearms. As a rule, the entire skin is dry and there may be a variable degree of ichthyosis.

The majority of cases show leukoplakia oris. Whitish plaques occur on the buccal mucosa, particularly along the interdental lines posteriorly, tapering off gradually toward the buccal commissures. In some, the anterior two thirds of the tongue exhibits irregular patches of smooth white infiltration. In others, there are transverse whitish striations on the posterior two thirds of the tongue or white streaks on the borders and ventral surfaces. Occasionally, the gingivae show whitish thickening.

On occasion, the nasal mucosa may be similarly affected while the tympanic membranes may be so thickened as to cause total deafness.

The hair of the scalp may be sparse or normal. Corneal dystrophy may lead to opacities and partial blindness. Bilateral cataracts have also been reported. The teeth are usually normal.

Sufferers of this disease usually have average intelligence. Many are able to walk only with great difficulty and discomfort because of the blebs and thick calluses on the soles.

Complications. Among the associated findings have been atrophy of the tongue, abnormally long bones, twinning of the incisor teeth, and excessive motility of the
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joints. The common complication is paronychial infection which follows slight injury and leads to shedding of the affected nail.

Inheritance. The cause of pachyonychia congenita is not known. It appears to be transmitted as a dominant characteristic but it is not certain whether a single gene or two dominant genes acting together are necessary.

Pathology. Histologic examination confirms the fact that the disturbance of the nail is due to the accumulation of keratinous material beneath the distal portion of the nail. The pathologic findings in other manifestations of the disease will vary according to location, stage, and type of lesion. A specimen prepared from a knee lesion was reported as exhibiting hyperkeratosis, parakeratosis, acanthosis, and dyskeratosis. The stratum granulosum was present throughout. The rete showed granular degeneration, deeply stained nuclei, and intracellular edema. The basal cell layer was irregular and the cells were swollen. Horny plugs occupied the follicular orifices and some sweat pores. The sebaceous glands were small but the sweat ducts were large. The blood vessels were dilated and often surrounded by lymphocytes, mast cells, connective tissue cells, and an occasional plasma cell.

TREATMENT. Vitamin A has been recommended for pachyonychia congenita although it has not been universally successful. It is administered orally (100,000 units daily) and applied in ointment form to the beds of the nails that have fallen off or have been removed surgically. A rubber base foot mold has been used successfully to relieve the effects of the plantar calluses. Hypnosis has also been employed to produce improvement in the hyperkeratosis of the soles.

A successful surgical procedure involves the removal of the nails followed by thorough scraping of the matrix to prevent regrowth. In one case, removal of the distal phalanges permitted the patient to remain with useful fingers. Skin grafting to relieve the plantar calluses has usually given poor results.

BIBLIOGRAPHY

