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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.
Pachyonychia Congenita

SALIL K PANJA*, P K DATTA**
A K JAISWAL***

PACHYONYCHIA congenita or Jadassohn-Lewandowsky syndrome is a rare congenital and sometimes familial malady characterised by distinctive thickened nails of fingers and toes, palmpoplantar hyperkeratosis, follicular keratosis of skin and leucokeratosis of mucous membrane.

Pachyonychia congenita was first reported by Jadassohn and Lewandowsky, (1910). Since then about 60 cases have so far been reported in the world literature.

The object of reporting this case is the extreme rarity of the syndrome.

CASE REPORT

A middle aged man of 36 years presented with deformities of nails of fingers and toes since birth. He also complained of painful corns and callosities with occasional bullae over soles of about 4 years duration and recurrent attacks of paronychia along with whitish oral patches since childhood. There was no history suggestive of palmpoplantar hyperhidrosis, hoarseness of voice, eye involvement, teeth malformation or alopecia.

Family history—There was no history of consanguinity in the parents. He has 2 sons and 1 daughter. Nail changes and leucokeratosis were seen in the daughter and in 1 of his sons. The other son was normal. He has 6 brothers and 4 sisters. None of his parents, brothers and sisters were afflicted with the disease.

Examination—On examination, the patient was of fair build and well nourished. The nails of the fingers and toes were affected and appeared greatly thickened, opaque, lustrless and folded longitudinally (Fig 1). The distal edges of the nail plates were elevated due to thick horns mass of subungal keratosis.

Patient also had corns and callosities of soles with moderate degree of palmpoplantar hyperkeratosis. There were follicular blackish grey papules with central keratotic plugs over extensor surfaces of extremities and umbilicalar region. Occasional epidermal cysts were present over root of the neck. Buccal mucous membrane showed leucokeratosis with angular stomatitis. There were 12 carious teeth in upper jaw, 6 on each side. No cornal dyskeratosis, cataract, alopecia, teeth malformations, pigmentedary changes or mental deficiency were detected. Systemic examination did not reveal any gross abnormality. About 18 years back all nail plates were removed under general anaesthesia but this was soon followed by reappearance of dystrophic thickened nail plates.

Fig 1—Showing Greatly Thickened, Laterally Curved and Distally Elevated Nails in Pachyonychia Congenita

DISCUSSION

Pachyonychia congenita is a term applied to 2 distinct syndromes with several features in common, viz., thickened nails and palmpoplantar keratoderma with hyperhidrosis. In type I (Jadassohn-Lewandowsky syndrome) mucosal leucoplakia occurs later (Jackson and Lawler, 1951; Joseph, 1964). In type II there are no mucosal lesions, but sebaceous glands develop on the trunk, and epidermal cysts in the scalp (Soderquist and Reed, 1968). It is inherited as an autosomal dominant trait.

The condition is sometimes associated with features such as keratosis pilaris, leucokeratosis, ichthyosis, bullae, corneal dyskeratosis, cataracts, hoarseness of voice and hair abnormalities such as hypotrichosis. Teeth may erupt precociously or may be carious.

Pachyonychia congenita has to be differentiated from dyskeratosis congenita. Salient features of differentiation between dyskeratosis congenita and pachyonychia congenita are: (1) autosomal/sex linked recessive inheritance in the former whereas autosomal dominant in the latter; (2) nails—dystrophic or absent in dyskeratosis congenita, but nails are greatly thickened in pachyonychia congenita; (3) verrucose lesions are usually absent in dyskeratosis congenita whereas in pachyonychia congenita the lesions may be present in elbows, popliteal areas, legs; (4) in dyskeratosis congenita, there is frequent association with Fanconi's disease whereas no such association in pachyonychia congenita; (5) complications like squamous cell carcinoma of mouth, anus, are present in dyskeratosis congenita but complications are absent in pachyonychia congenita.

Some workers however, believe that both maladies are variations of the same disorder.

In this case there were thickened nails, palmpoplantar keratodermia, though it is a common feature. The presence of 12 carious teeth is also common in general and is uncommon in this case.

No treatment is available for this complex factor. In this case, leucokeratosis of nails and skin is common. Special oral vitamin A induces keratodermia.

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Family History - There was no history of consanguinity in the parents. He has 2 sons and 1 daughter. Nail changes and leukokeratosis were seen in daughter and in 1 of his sons. The other son was normal. He has 6 brothers and 4 sisters. None of his parents, brothers and sisters were afflicted with the disease.

Examination - On examination, the patient was of fair build and well-nourished. The nails of the fingers and toes were affected and appeared greatly thickened, opaque, burred and folded longitudinally (Fig. 1). The distal edges of the nail plates were elevated due to thick horny mass of subungual keratosis.

Patient also had corns and callosities of soles with moderate degree of palmar plantar hyperkeratosis. There were follicular blackish grey papules with central keratotic plugs over extensor surfaces of extremities and lumbar-sacral region. Occasional epidermal cysts were present over root of the neck. Buccal mucous membrane showed leukokeratosis with angular stomatitis. There were 12 carious teeth in upper jaw. 6 on each side. No corneal dyskeratosis, cataract, alopecia, teeth malformations, pigmentation changes or mental deficiency were detected. Systemic examination was unremarkable.

Fig. 1 - Showing Gravely Thickened, Laterally Curved and Distally Elevated Nails in Pachyonychia Congenita

DISCUSSION

Pachyonychia congenita is a term applied to 2 distinct syndromes with several features in common, viz. thickened nails and palmar plantar keratodermia with hyperhidrosis. In Type I (Jadassohn-Lewandowsky syndrome) mucosal leukoplakia occurs later (Jackson and Lawler, 1951; Joseph, 1964). In Type II there are no mucosal lesions, but sebaceous cysts develop on the trunk, and epidermal cysts in the scalp (Soderquist and Reed, 1966). It is inherited as an autosomal dominant trait.

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Some workers however, believe that both maladies are variations of the same disorder.

In this case there were thickened nails, palmo-plantar keratodermia and fissures in it falls under pachyonychia congenita. Though our patient was of Type II, he was positive for the presence of 12 carious teeth, which is common in Type I.

No treatment for this syndrome is available. In this case, leucokeratosis and alopecia were treated with vitamin A.

Soderquist NA, Reed RG, Arch Dermatol 1966; 94: 31.

Prolymphocytic Leukemia

GALTON'S definition of lymphatic leukemia (1944). It is rare disorder, though has been reported to have been reported to be in children. It is considered it to be a variant of lymphocytic leukemia. Due to its frequency, we will consider it as a separate entity.

A 30-year-old male patient with a history of lymphocytic leukemia for 7 years, was referred to us for further management. The patient had been treated with CHOP regimen for 5 years and was in complete remission for 2 years. However, he developed progressive symptoms of lymphatic leukemia with weight loss of 20 kg, fever, and persistent night sweats.

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plantar keratoderma and mucosal leukokeratosis. Hence it falls into Type I of pachyonychia congenita. Though epidermoid cysts are usually seen in Type II, they were present in this case also. Presence of 12 carious teeth in this patient is unusual and is commonly found in dystkeratosis congenita.

No treatment is available; oral vitamins and B complex factors play a doubtful role. In the present case, leukokeratosis improved with vitamin A injections. Special shoes may relieve the pressure which induces keratoderma (Garb, 1959).

SUMMARY

A middle aged male presented with gross nail deformity with thickening, palmoplantar hyperkeratosis, leukokeratosis, carious teeth, follicular keratosis, epidermal cyst, recurrent paronychia, occasional bullae and painful corns and callucities of soles of both feet and the case was diagnosed as pachyonychia congenita. Nail changes were present since birth, while others developed gradually. No associated palmoplantar hyperhidrosis, eye changes, abnormality of voice, chilblains, teeth malformations or alopecia were detected. Leukokeratosis improved with vitamin A injections.

REFERENCES


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Prolymphocytoid Transformation of Chronic Lymphatic Leukaemia

JAYEN C SHAH*

Galton’s prolymphocytic leukaemia is a rare type of lymphatic leukaemia described by Galton et al (1974). It is now a well defined lymphoproliferative disorder, though only a few well documented cases have been reported. To date, most of the authors consider it as a variant of chronic lymphatic leukaemia. Due to its rarity a case is reported here.

CASE REPORT

A 69-year-old male was seen in May 1990 with a history of weakness and recurrent chest in-

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*MS, Resident in Surgery

fection for 6 months. The patient had a marked splenomegaly but no lymphadenopathy. He had a minimal hepatomegaly.

Investigations—A chest x-ray showed an elevated left hemidiaphragm and a left basal atelectasis. White cell count was 35,700 per cmm with 78% lymphocytes. There were no blast cells or atypical cells in the peripheral blood. Haemoglobin was 16.2 g/dl and haematocrit 47.0%. Bone marrow studies showed erythroid hyperplasia and iron deficiency and infiltration with well differentiated lymphocytes. The overall percentage was less than 20%. Therapy was not started at that time. His white count went on increasing and reached up to 63,000 per cmm. The patient clinically became worse. The spleen went on getting bigger.

Diagnosis—A provisional diagnosis of an atypical chronic lymphatic leukaemia was made.

Management—He was started on a combination chemotherapy including prednisolone 40 mg daily and chlorambucil 3 mg daily.

Progress—In December 1990, patient was readmitted. Repeat studies showed prolymphocytes in peripheral blood (Fig 1. Wright's stain, x1500). Electron microscopic studies confirmed their presence. A liver scan showed mild enlargement without any defects. A spleen scan showed a large spleen with a generalised decreased uptake and a localised defect in lower lateral portion. A revised diagnosis of prolymphocytic leukaemia was made.

Fig 1—Showing Prolymphocyte in Peripheral Smear

In August 1991, his spleen was huge, reaching a level of 15 cm below the left costal margin and there was elevation of left hemidiaphragm. The patient also complained of heaviness in left flank and a dragging pain there. His platelet count dropped to 69,000 per cmm with a poor clot retraction. Total white count was 56,000 per cmm with peripheral smear showing 76% prolymphocytes. Serum albumin was 2.4 g/dl, producing pedal oedema.

A splenectomy was performed at this juncture. Spleen was adherent to the diaphragm and the
plantar keratoderma and mucosal leukokeratosis. Hence it falls into Type I of pachyonychia congenita. Though epidermolytic cysts are usually seen in Type II, they were present in this case also. Presence of 12 carious teeth in this patient is unusual and is commonly found in dyserkeratosis congenita.

No treatment is available; oral vitamins and B-complex factors play a doubtful role. In the present case, leukokeratosis improved with vitamin A injections. Special shoes may relieve the pressure which induces keratoderma (Garb, 1989).

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CASE REPORT

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fection for 6 months. The patient had a marked splenomegaly but no lymphadenopathy. He had a minimal hepatomegaly.

Investigations—A chest x-ray showed an elevated left hemidiaphragm and a left basal atelectasis. White cell count was 33,700 per cmm with 78% lymphocytes. There were no blast cells or atypical cells in the peripheral blood. Haemoglobin was 16.2 g/dl and haematocrit—47.0%. Bone marrow studies showed erythroid hyperplasia and iron deficiency and infiltration with well differentiated lymphocytes. The overall percentage was less than 10%. Therapy was not started at that time. His white count went on increasing and reached up to 62,000 per cmm. The patient clinically became worse. The spleen went on getting bigger.

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A splenectomy was performed at this juncture. Spleen was adherent to the diaphragm and the