Pachyonychia Congenita Project

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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.

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Multiple Telangiectases (Hereditary Telangiectasia of Rendu-Osler-Weber?). Presented by DR. NATHAN SOBEL.

C. H., a white girl aged 5, is presented from the Skin and Cancer Unit, where she was first seen on Jan. 13, 1947, with an eruption which has been present since the age of 15 months. It appears first on her face and gradually spread to other areas. She has never had any hemorrhages. One aunt gives a history of nosebleeds, cause unknown.

On the face and hands are numerous small areas of telangiectasia. These are also present on the vermilion border of the lips. On the right buccal mucosa there are a few pinhead-sized, red areas. Over the whole body, but especially noticeable on the legs, there are numerous large pea-sized, blanched, flat spots, in many of which is a central dilated blood vessel without any radiating branches. On the body and arms there are port wine marks of various sizes.

DISCUSSION

DR. MAURICE J. COSTELLO: I think the lesions on the legs are punctate telangiectasis with avascular areas around them.

DR. DAVID BLOOM: The remarkable feature is the numerous, round, depigmented lesions on the extremities which have a small red or brown punctum in the center resembling leukoderma centrifugum.

DR. WILBERT SACHS: My contention is that it was capillary and not arterial, or the lesions would not bleed as easily.

DR. NATHAN SOBEL: I felt that the central puncta were dilated blood vessels; consequently, I do not believe they were "halo" nevi. As far as telangiectasia due to syphilis is concerned, a serologic examination is indicated. I have also seen 1 patient with telangiectasis on the palm in late acquired syphilis.

Pachyonychia Congenita with Keratoderma of Palms and Soles and Leukoplakia Oris. Presented by DR. FRED WISE.

S. H., a woman aged 40, registered at the Skin and Cancer Unit on Jan. 23, 1947, presenting various cutaneous lesions. She has a son aged 16 who has similar lesions and who is being presented simultaneously. A daughter aged 19 is said to be normal, and there is no history of a similar disturbance in other members of the immediate family or in distant relatives. From the age of 1 year until she was 7, the patient every year had "skin sores" all over the body; these became blistered and later scabby and crusty. They would last all summer, so that she was obliged to stay in the hospital. At the age of 22 she had an infection of the mouth, and 25 teeth were extracted at one time. Her hands and feet are always cold. Menstruation has been regular. She has been having migraine headaches since the age of 24. Her general health has been good except for difficulty in walking because of the callouses. She is intelligent and alert.

A week or two after birth the nails of the fingers and toes became discolored and thickened, reaching their height within a month or two. They have changed little since that time. A slight injury causes an infection, and the entire nail is cast off. The lesions on the feet first appeared at the age of 18 months, when she began to walk, and became more pronounced and progressively thicker as she grew older, so that at times her mother was obliged to pare them off. They are painful on walking. In 1933 a skin graft was performed on the left heel, with poor results. She has a continuous tired feeling.

At the points of pressure on the soles, heels and toes, symmetrically distributed, are hard, thick, painful, hyperkeratotic, lemon-colored growths, tender to touch. On the right palm at the base of the metacarpals and the interphalangeal joints are similar growths. All the toe nails are dry, dystrophic and blackish in color, with irregular, rough surfaces. The finger nails are long, thickened, curved, discolored and tapering off toward the end, with the nail of the left ring finger partially broken off. On the right buccal mucosa are small, match head–sized, leukoplakic patches. The entire dorsum of the tongue is covered with whitish, adherent patches and with leukoplakic areas which are particularly noticeable on the sides. The scalp is covered with dry, scaly, diffuse, seborrheic patches. There are two hairy, pea-sized, pigmented moles on the chin. The hands and feet are cool to touch. The integument is dry.

The Mazzini reaction was negative. The urine was normal. The hemogram revealed a normocytic anemia, with 3,710,000 erythrocytes, a hemoglobin content of 12.2 Gm. per hundred cubic millimeters (73 per cent). The color index was 98. The leukocyte count showed 8,000 cells per cubic millimeter, with 50 polymorphonuclear neutrophils, 41 lymphocytes and 5 monocytes.

An ophthalmologic examination including the eyegrounds and a slit lamp examination of the cornea revealed nothing abnormal.

The total carotinoid content of the blood was 170 units per hundred cubic centimeters (normal, 50 to 100). The vitamin A content was normal.

Vitamin A, 75,000 units daily for a period of six weeks, was taken until three weeks ago. Since February 19 the patient was advised to continue taking 50,000 units of vitamin A and 10 mg. of vitamin E three times a day and ¼ grain (0.015 Gm.) of thyroid extract. Salicylic acid salve, 20 per cent, was prescribed, to be applied to the callouses three times a day.

Pachyonychia Congenita with Keratoderma of Palms and Soles and Leukoplakia Oris. Presented by Dr. Fred Wise.

Lupus Miliaris Disseminatus Faciei. Presented by Dr. Jack Wolf.

Dermatitis Venenata of the Feet and Popliteal Areas (Nylon). Presented by Dr. Jack Wolf.

A Case for Diagnosis (Angioma Serpiginosum?). Presented by Dr. Fred Wise.

M. L., a woman aged 38, registered at the Skin and Cancer Unit on Nov. 21, 1946, complaining of lesions of about fifteen years' duration. Her first and only pregnancy resulted in a stillbirth in October 1945. The Rh factor of both the patient's and her husband's blood was normal. The basal metabolic rate in April 1946 was said to be on the "low side," and she took thyroid extract tablets for two months. She gives no history of previous cutaneous disorders.

The eruption first came out on the chest. During pregnancy it began to appear on the neck and has been "creeping up and down." The eruption is worse during the warm weather.

There is a well defined, fairly symmetric, rose red eruption on the sides of the neck, lower third of the face and front of the chest. It is most pronounced on the right side, where it involves two thirds of the right clavicle, arranged in a V shape. The lesions consist of pinpoint to pinhead to match head–sized spots and irregular, diffuse patches. Most of the spots are light brownish, while the patches are rose red. The lesions on the neck consist mostly of isolated pinpoint