Pachyonychia Congenita Project

15 March 2005

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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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similar to those occurring in other parts of the body. This opinion was made without any suggestion from the dermatologic department. There is no history or evidence that the telangiectasia is congenital or familial. It was not present at birth and was never noticed until the patient was 40 years old.

Pachyonychia Congenita. Presented by DR. A. W. GRACE.

J. P., a schoolboy aged 9 years, was first seen in December 1936. At 2 years of age he had had measles, complicated by pneumonia. During convalescence an eruption arose all over the body in the form of small pustules, occurring at first singly and then in crops, and finally coalescing to form thin-walled bullae filled with seropurulent material. At the same time the finger-nails and toe-nails became markedly thickened. During the past seven years the condition has persisted, with the bullous lesions healing in places and arising in new sites; the nails have been frequently shed. In October 1936 hyperkeratotic lesions were first noted on the buttocks, the extremities and the back. The family history was noncontributory.

On examination the upper lip, just below the nares, shows a small crusted superficial lesion resembling impetigo contagiosa. On the dorsum of the right foot is a collapsed bulla about 2 cm. in diameter, with a base and a small surrounding zone of erythema. All the nails are greatly thickened, and some show perionychial inflammation. Practically the whole surface of the skin, with the exception of the scalp, the palms and the soles, is the site of follicular hyperkeratosis. On the borders of the natal cleft and on the tips of the elbows the hyperkeratosis is particularly well marked, forming warty masses, some as large as a match head. The larger lesions are slightly hemorrhagic as a result of scratching. The tongue is covered on its dorsal and lateral aspects with a whitish "fur" which cannot be scraped off, and similar elevated areas are present on the hard palate and the buccal mucosa, opposite the dental line on both sides of the mouth. The patient is pale and appears chronically ill.

Histologic examination of a hyperkeratotic area of skin showed follicular dipping down of the horny layer, which was much thickened. Between the keratin plugs the skin was somewhat atrophic and the cornification not so marked. In a few places at the bottom of such plugs there was a degenerating layer of cells belonging to the stratum granulosum, the granules of which were swollen and deep staining and lay about loose in the degenerating tissue, resembling the "grains" of Darier's disease. *Corps ronds* were not seen. Serial sections were not examined.

The results of complete physical examination and of the Kline test, a blood count, urinalysis, determinations of cholesterol and urea nitrogen in the blood, measurement of the basal metabolic rate, culture of stools, and mycologic examination of lesions on the feet were noncontributory. Treatment consisted of application of ammoniated mercury ointment to the bullous lesions.

DISCUSSION

DR. E. B. TAUBER, Cincinnati (by invitation): I agree with the diagnosis. The patient shows the characteristic symptoms originally reported by Jadassohn and described in a later case under different terminology by Cole and Dower. He shows the condition of the tongue, which has a thrushlike appearance, the cutaneous lesions and the lesions of the nails. I have a patient, 20 years old, who has a similar condition, and in a case of such long duration the nails are usually completely absent. The condition in this type of case will pass at times as possible epidermolysis bullosa and at times as some other congenital condition. There will have been some roentgen therapy and sometimes application of ointments, possibly with improvement at times. The cutaneous lesions, curiously, seem at times to improve spontaneously. There is an apparent loss of immunity to pyodermic conditions. Patients with this condition complain that every little trauma irritates them. They are practically in the condition of persons with epidermolysis bullosa.

Proceeding along lines suggested by these observations, empirically and inductively, I discovered that there was absence of cystine. Dr. Louis A. Brunsting, of the Mayo Clinic, has been using cystine in the treatment of other conditions. I used injections of cystine for my patient, and complete amelioration of the cutaneous manifestations followed. The nails were gone, but the cutaneous condition cleared up, and the mucous membranes are now smoother and less aggravated than they have been in the past ten or twelve years.

DR. A. W. GRACE: I shall carry out what Dr. Tauber suggested in regard to therapy in this case. It is probable that therapy by replacement of sulfur was suggested by the work done on pellagra some time ago. It was found that patients with pellagra lost a good deal of sulfur through shedding of the nails. Since then the giving of sulfur has been found to be of some benefit in treating pellagra. It is possible that this boy is losing a good deal of sulfur from his economy by the falling off of these large nails from time to time.

Dyshormonal Dermatosis (Werner's Syndrome). (Scleroderma, Poikiloderma, Bilateral Juvenile Cataract, Precocious Graying of the Hair, Pluriglandular Dysfunction.) Presented by DR. MARION B. SULZBERGER.

D. G., a man aged 44, born in the United States, is the brother of the patient A. G., to be presented here tonight.

The illness began at the age of 6 years with the discovery of high grade myopia. At the age of 12 years graying of the hair appeared. At 18 the patient suffered from gonorrhea, and at 22 there was completely gray hair and progressive alopecia. At 28 years bilateral periarterial sympathectomy was performed on both femoral arteries (no details of which are available). At 29 extensive cellulitis developed on the left foot, with subsequent development of numerous ulcerations on both feet. At 36 blindness suddenly developed within six months, and vision was restored after successful removal of cataracts. The patient has indulged freely in alcohol and tobacco. The libido and potency are claimed to be normal.

Family History.—Both the grandparents and the parents were first cousins. The patient's father died at the age of 83 of diabetic gangrene. The mother had graying hair at an early age and has bilateral cataracts. There is an extensive family history of premature graying of the hair. The patient's brother has a similar condition. Eight other siblings died during childhood.

The patient is mentally rather subnormal and has a defective memory. The chest and the trunk are well developed; the extremities are small; the bony prominences are exaggerated. There is advanced baldness, with graying of the remaining hair. The eyes have both been operated on for cataracts. The teeth are in fair condition. The patient's voice is remarkably highly pitched. His blood pressure is 134 systolic and 95 diastolic. The external genitalia and testes are small. There are sclerodermatous changes on the distal parts of all the extremities and poikilodermatous changes over the knees. There is definite hyperkeratosis of the soles, and numerous scars from old ulcerations are visible on the feet. There are other patches of hyperkeratosis over the olecranal processes. The skin of the face is thickened and roughened.

Laboratory Data.—The blood count was normal except for slight lymphocytosis. The urinalysis showed no abnormality. Chemical analysis of the blood showed 13 mg. of urea nitrogen and 89 mg. of dextrose per hundred cubic centimeters. The dextrose tolerance test showed results of 85, 94, 95, 95 and 88 mg. per hundred cubic centimeters, and the calcium determination, 10.5, 10.5, 11, 10.5 and 10.2 mg. per hundred cubic centimeters. The calcium balance was within normal limits. The phosphorus determination gave results of 4.1, 3.5, 3.8 and 4 mg. per hundred cubic centimeters. The basal metabolic rate ranged between +4 and —15 per cent.

Histologic examination revealed changes characteristic of scleroderma and poikiloderma. Roentgenograms demonstrated linear calcifications in the soft parts of the calves. Several phalanges and parts of the metatarsal bones were missing.