



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.

the condition was present at the patient could remember in as so in Engman's case. If here, this would be more evidence, this would be more evidence of dystrophy or atrophy of the neck and upper part of the face. In Engman's cases, a dyskeratotic atrophy of the extremities and membranes of the mouth. "The process is dystrophic." The process is like the ichthyoses or the keratosis similes (bullosis mechanica similes) fit in the Darier's comedone-Brooke. The symptoms like those of acanthosis nigricans, but in that disease were free of the dystrophic character of the disease. The process corresponds closest to that described by Lewandowsky, eighteen cases described. Because of the difference would separate them as still distinct entities.

These dyskeratoses may be is a group. Richards would say that the process is ectoderm. Consanguinity cases, as has been brought out from Jadassohn's clinic. The dominant, and ichthyosiforme characteristic. Bettmann (p. 715) thinks that the anomaly of the feet is a disease of inheritance and are to be considered as a dominant inheritance.

There may be two similar ones in the skin. The process is probably congenital and dystrophic, pigmentation, dys-

trophic changes in the nail and by leukokeratosis of the mucous membrane of the mouth.

3. The process has some characteristics in common with the dystrophic type of epidermolysis bullosa (bullosis mechanica dystrophica), with Darier's disease and with the juvenile type of acanthosis nigricans. It probably approaches closest, however, to the syndrome described by Jadassohn and Lewandowsky under the term pachyonychia congenita.

4. We believe the characteristics of the latter syndrome to have been well enough defined and described in eighteen cases and one questionable case, which we have culled from the literature, to entitle it to the designation of a distinct disease.

5. In the three cases mentioned, the condition appears to be most like pachyonychia congenita, but because of the dystrophic effect we would provisionally suggest the name of dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris.

In conclusion we wish to express our thanks to Dr. Maurice L. Richardson and to Dr. Otto Saphir of the Pathology Department of the Cleveland City Hospital for their help in the preparation of pathologic specimens and in the making of photomicrographs.

ABSTRACT OF DISCUSSION

DR. MOOK, St. Louis: I think unquestionably the cases shown by Dr. Engman and Dr. Cole belong to the same group. In our case the boy was 12 years of age when we first saw him. At that time he had ulcers only on both sides of the tongue. The skin, hair and nails were normal. An attempt was made to diagnose the condition tuberculosis, but he had no visceral lesions of tuberculosis. We next saw him ten years later, and he had then developed the universal condition of the skin. His hair was thin, the eyelashes and eyebrows had disappeared, the nails were completely destroyed and he had pigmentation of the mucous membrane with considerable atrophy of the mucous surfaces of the cheeks. At the same time a younger brother was developing a similar condition on the tongue, and we believe that in time he will develop the same condition. In this family, at any rate, the condition is familial and probably congenital.

There was one feature about our case that was suggestive that it might belong to the nevus group, in that on the abdomen there were scroll-like lesions similar to those in nevus unius lateralis, without the keratosis. In addition, there was a line running down from the sternum to the pubis and a cross line at about the tenth ribs; so that his trunk was divided into quadrants, the upper left and the lower right being much more pigmented than the upper right and the lower left. The general health of the patient was unaffected, but the process is progressive.

DR. HOWARD FOX, New York: I was much interested to hear of this case of Dr. Cole's. It did not seem to be precisely like the one reported in the *Ikonographia* by Jadassohn and Lewandowsky as pachyonychia congenita. As Dr. Cole mentioned a case of mine and put it in the doubtful class, it might be well to discuss it. Dr. Andrews and Dr. Strumwasser who reported a case from New York considered my case almost identical with their own. The nails were thickened rather than atrophied and were precisely like the picture published in the *Ikonographia*. The

patient was a boy, about 4 years of age, who had wartlike lesions on the extremities and buttocks and a few flaccid bullae on the feet underneath callosities. He did not have leukokeratosis of the tongue, nor did he have lesions suggesting granuloma rubra nasi, but it seemed to me that it was unquestionably a case of the anomaly described by Jadassohn and Lewandowsky.

PROF. JOSEF JADASSOHN, Breslau: Dr. Cole was kind enough to show me his interesting case when I was in Cleveland. I agree with him that it is to be classified as one of the congenital anomalies, and would like to add only a few remarks.

As Dr. Cole noted, Lenglet was the first to enumerate the different symptoms which are to be found in this group. Dr. Cole and I completed this series. I want to call attention to a further symptom, namely, hyperhidrosis of the fingertips while the palms are dry. I saw this symptom in different forms of this group, even in common ichthyosis.

The well known types of congenital diffuse anomalies of the skin are characterized by typical combinations of some of these single symptoms. If this combination is a different one, then we have the atypical cases, as, for instance, Dr. Cole's case. It seems to me important to describe these cases exactly as Dr. Cole did, because they may not be as rare as our literature seems to show, and if we have no exact description, we cannot bring together the corresponding cases. This is necessary not only from a diagnostic standpoint but also for studies about the heredity of single diseases. It seems important, especially in these atypical cases, to find out whether the same atypical combination is to be seen in different members of the same family. We must always try to examine as many relatives as possible, to establish whether or not there is consanguinity of the parents, and so forth.

I want to emphasize that there are in this whole group many abortive forms ("formes frustes"). They are well known in the common ichthyosis, but as far as I can see they are not recognized, e. g., in the cases which belong to the ichthyosis paratypica of Besnier or the hyperepidermotrophie of Brocq. I see, not infrequently, stripes of hyperkeratosis on the flexor sides of the knees and the elbows, on the front side of the axillae and on the navel. I have seen that anomaly sometimes even in several members of the same family. It seems to be not without interest that many of the single symptoms of these congenital anomalies are only exaggerations of qualities of the skin which we generally do not call anomalies. I mention hyperhidrosis, keratosis pilaris, dry and somewhat hyperkeratotic skin, etc. In this connection I must mention the dilatation and light hyperkeratosis, and later on, atrophy of the follicles of the neck and chest, and the form of the toe nails, which resembles the pachyonychia, by their strong convex (but not hyperkeratotic) forms. Perhaps it will be possible by studying these formes frustes in families to find more material for the study of heredity in skin diseases.

Finally, I want to say that the name pachyonychia described by Lewandowsky and myself is incomplete. We wanted to publish in the *Ikönographia* the results of studies on the nail form, but it will be necessary to complete this name, and we can do this only by a short enumeration of the principal symptoms. In this manner it will be necessary both to give to the new cases the right place in the group of congenital anomalies, and to individualize it in such manner that it will not be lost in the future.

DR. WILLIAMS, New York: I am much interested in this presentation for several reasons. In the first place, I am interested in the diagram showing groups of circles and straight lines, which I suppose are intended to show some sort of relationship. It is a vivid picture, satisfying to the eye, but misleading for that reason. It seems to clarify our knowledge, but really does nothing of the kind, because there is no definition of the exact relationship which is intended.

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In our group many abortive forms of ichthyosis are common, but as far as ichthyosis which belong to the ichthyosis of Brocq. I see, not infrequently of the knees and the elbows, I have seen that anomaly sometimes. It seems to be not without congenital anomalies are only generally do not call anomalies. In somewhat hyperkeratotic skin, and light hyperkeratosis, and the form of the toe nails, convex (but not hyperkeratotic) the formes frustes in families to diseases.

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presented in this presentation for the diagram showing groups intended to show some sort of eye, but misleading for that only does nothing of the kind, which is intended.

In the second place, we have a good deal of confusion because, as Professor Jadassohn has brought out, we have the occurrence in a family of a disorder with definite but varied symptoms. All the cases have certain points of resemblance, but there are many individual differences. The only conclusion, I think, is that there is some underlying fault, some dysfunction of the elements that control development.

I wish to emphasize further what Professor Jadassohn said, that if such cases are to be studied, all members of the family must be studied, and all the symptoms recorded.

The deformity of one of the nails looked, as Dr. Cole said, exactly as if it had been caused by trichophytosis. It certainly did. I have seen many of these, in which I was unable to decide whether the deformity was due to dystrophy or to an infection caused by *Trichophyton*. I believe that in many cases dystrophy, whether produced by injury, by malnutrition or by some other causes, is responsible for the greater part of the visible deformity, and furnishes a suitable ground for the growth of various organisms, including the trichophytons.

DR. COLE, Cleveland: In reply to Dr. Williams' question, I may say that at one time we wondered about the possibility of this being a parasitic process. We thought we cultivated a thrushlike organism from the tongue, and looked for it zealously in the nails. I think it is possible for a parasitic organism to get in and yet mean nothing.

I did not emphasize, possibly, as much as I should the fact that this is undoubtedly a familial disease. In the cases reported by Bettmann the father and three sons had the condition. In the report from the Jadassohn clinic there was a marked influence of consanguinity when cousins married, the flare-up in the next generation illustrating this. In our case we found no family history in this way as far back as we could go. I have not seen the case mentioned by Dr. Fox. I saw the reprint but was not sure about putting it in this group, or whether it was a type of epidermolysis bullosa. Of course, Dr. Fox saw the patient and knows far more than I could get out of the report.

As far as the circles in one of our illustrations are concerned, any one who reads anything from Brocq's clinic knows that the clinicians there are fond of circles, and this illustration was from his clinic.