Pachyonychia Congenita Project

15 March 2005

Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

> Sec. 107. - Limitations on exclusive rights: Fair use
> Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, **teaching (including multiple copies for classroom use), scholarship, or research**, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

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.

2386 East Heritage Way, Suite B, Salt Lake City, Utah 84109 USA
Phone +1-877-628-7300 • Email—Info@PC-Project.org
www.PC-Project.org

VOLUME 107
NUMBER 1

As in the first biopsy, the marrow was completely cellular. The microscopic appearance is illustrated in figures 3 and 4. However, there was a very marked reduction in the number of immature red cells (megaloblasts), which were present only in small numbers in a few isolated areas. Some of these stained faintly pink. There were many more mature erythrocytes, and the number of normal nucleated red cells (normoblasts) was greater. The myelocytic elements were markedly increased above the number in the previous biopsy. There were many eosinophilic myelocytes and leukocytes and a great abundance of neutrophilic myelocytes and leukocytes. The megakaryocytes appeared to be about the same in number as before. The constituents were normal but the marrow was hyperplastic.

COMMENT

The material from the sternal marrow in this case indicates that in the macrocytic anemia of pregnancy the change is fundamentally the same as that in true pernicious anemia and in the macrocytic anemia of sprue.

During relapse, both in pernicious anemia and in sprue, the marrow is characterized by extensive cellularity, chiefly owing to megaloblastic proliferation with a diminution of normoblastic and myelocytic elements.[7] On the administration of liver extract this megaloblastic proliferation ceases and the normoblastic and myelocytic cells return to their normal ratios. The changes are somewhat proportional to the degree of the anemia; i. e., the more severe the anemia, the greater the megaloblastic hyperplasia. The sternal marrow in this case showed a similar megaloblastic hyperplasia with reduction in the normoblastic and granular elements and after therapy with liver extract showed a similar return to normal.

Strauss [4] has pointed out that in the macrocytic anemia of pregnancy there is a failure in the formation

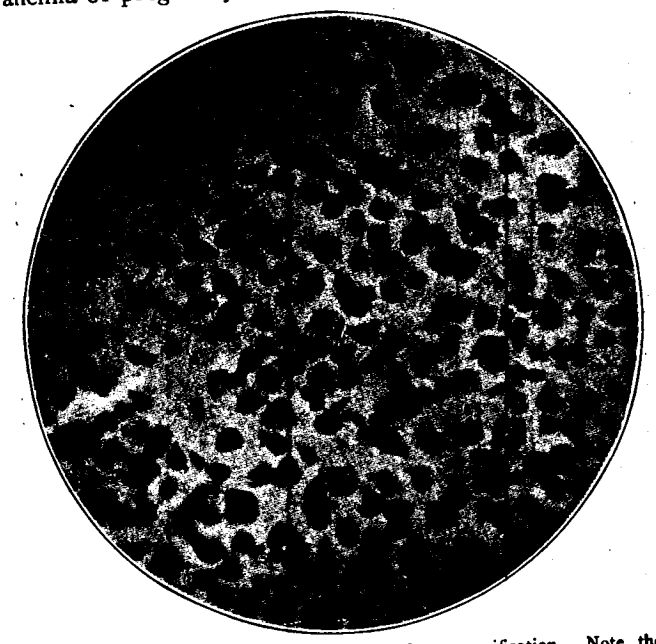

Fig. 4.—Field from figure 3 under higher magnification. Note the cluster of normoblasts just above the center. This is surrounded by myelocytes and young polymorphonuclear cells.

of antianemic factors similar to that in true pernicious anemia except that this failure is only temporary. This deficiency of antianemic factor may be the result of a lack either of the extrinsic factor in the diet or of the intrinsic factor in the gastric secretions. The diet in the case reported was deficient. Free hydrochloric acid was found both in the vomitus and in the gastric juice. Wintrobe and Shumacker [8] have recently suggested that the fetus in the development of its blood-forming tissues utilizes the antianemic factors elaborated by the mother. There is a definite macrocytosis of the red cells early in fetal life and, as term is reached, the cells approach normal size. If the amount of antianemic factor formed is too small (owing to insufficient amounts of either intrinsic or extrinsic factors as shown by Strauss), and this is all utilized by the fetus, the maternal blood forming tissues are inadequately supplied. The soil for the development of macrocytic anemia in the pregnant mother is thus prepared.

CONCLUSION

The bone marrow in a case of severe macrocytic anemia of pregnancy was studied. It was found, first, that the bone marrow resembles that seen in true pernicious anemia and in the macrocytic anemia of sprue during relapse and, secondly, that under the influence of specific therapy the cell constituents return to normal, as has been found when pernicious anemia and the macrocytic anemia of sprue are treated in a similar manner.

40 North Street.

7. Rhoads and Castle.[3] Peabody, F. W.: The Pathology of the Bone Marrow in Pernicious Anemia, Am. J. Path. 3: 179-202 (May) 1927.

8. Wintrobe, M. M., and Shumacker, H. B., Jr.: Comparison of Hematopoiesis in the Fetus and During Recovery from Pernicious Anemia, Together with a Consideration of the Relationship of Fetal Hematopoiesis to Macrocytic Anemia of Pregnancy and Anemia in Infants, J. Clin. Investigation 14: 837-852 (Nov.) 1935.

Clinical Notes, Suggestions and New Instruments

PACHYONYCHIA CONGENITA

ELMORE B. TAUBER, M.D.; LEON GOLDMAN, M.D., AND HARRY CLAASSEN, M.D., CINCINNATI

Pachyonychia congenita is a rare and uncommon condition. It is quite possible, however that many cases are overlooked, and they may be diagnosed as a simple congenital deformity of the nails. Most of the cases reported have been in children, for the reason that this condition is a particular form of a congenital abnormality.

This particular syndrome includes more than deformities of the finger nails. The title of Jadassohn and Lewandowsky's paper in 1906 "Keratosis Disseminata Circumscripta Tylomata and Keratosis Linguae" shows the essential features of this condition. Pachyonychia seems to be the constant symptom in all the cases reported. The others include bullae, palmar and plantar keratoses, leukoplakia, anomalies of the hair, follicular keratoses of an acneform type, especially on the knees and elbows, and dyskeratosis of the cornea. Heller [1] includes these changes under the term congenital dyskeratoses. Most of the cases seem to have been reported in males, chiefly Jews of Slavonic origin. Kumer and Loos [2] recently have studied a family of five generations and found thirty-four cases. From their cases and from a survey of the literature, they believe that pachyonychia congenita can be divided into three classes:

1. Type I: Symmetrical keratoses of hands and feet with follicular keratoses of body.
2. Type II: Symmetrical keratoses of hands and feet, follicular keratoses of body and leukokeratoses oris; this is the commonest type (typus Riehl).
3. Type III: Symmetrical keratoses, follicular keratoses of the body and corneal changes.

The recent reports in the American literature of Diasio,[3] Sohrweide [4] and Andrews [5] give excellent summaries of the

1. Heller, Julius: Die Krankheiten der Nagel, Handbuch der Haut- u. Geschlechtskrankeiten, Berlin, Julius Springer, 1926, vol. 13.
2. Kumer, L., and Loos, H. O.: Ueber Pachyonychia congenita (Typus Riehl), Wien. klin. Wchnschr. 6: 174 (Feb. 8) 1935.
3. Diasio, F. A.: Pachyonychia Congenita Jadassohn, Arch. Dermat. & Syph. 30: 218 (Aug.) 1934.
4. Sohrweide, A. W.: Pachyonychia Congenita, Arch. Dermat. & Syph. 32: 370 (Sept.) 1935.
5. Andrews, G. C.: Pachyonychia Congenita, Arch. Dermat. & Syph. 33: 183 (Jan.) 1936.

literature and add three new American cases. The case reported here combines all the features of pachyonychia congenita. This case was overlooked for a long time, being considered an atypical type of epidermolysis bullosa presenting nail deformities, chiefly because of the prominence of the bullae on the extremities.

REPORT OF CASE

The diagnosis in this case was made at the Mississippi Valley Central States Dermatological Conference in Cincinnati, Nov.

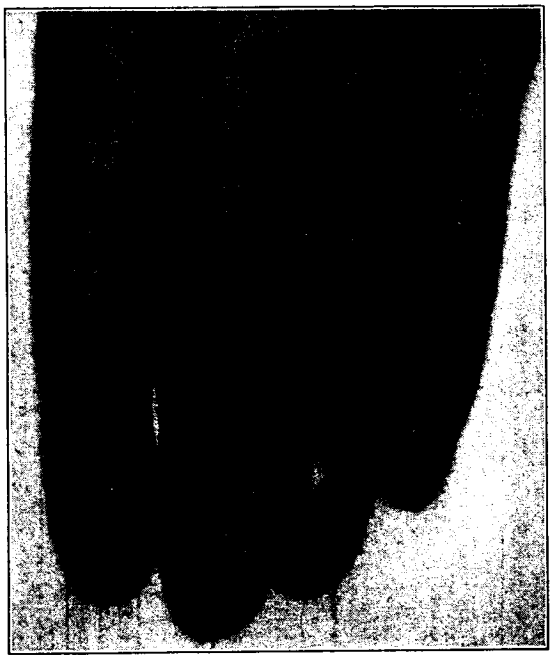

Fig. 1.—Deformity of nails and thickening of derma.

16, 1935. A Jewish girl, aged 20, had had since birth thickening and wrinkling of the nails of the hands and feet. Shortly after birth also some dryness of the skin was noted. In 1930, painful blisters appeared spontaneously over the soles of the feet and

Fig. 2.—Nails are deformed, thickened and wrinkled.

about the toe nails. About this time also the patient complained of some slight burning of the tongue. No blisters appeared on any other surface of the body or about the hands at any time. In 1932 the large toe nail on the left foot was removed surgically. Since 1930 the lesions on the feet have shown some slight improvement. Some of the large bullae have healed completely. The therapy has consisted of various mild local ointments, calcium and viosterol. The recent addition of the x-rays to the therapy, one erythema dose in divided doses, gave the patient great relief for a time.

Examination of the patient shows that she is well developed, of the brunette type. The skin in general, especially over the extremities, is dry and scaly. There is a mild papulopustular acne of the face. The finger nails show wrinkling and thickening and, for the most part, are deformed completely. The toe nails exhibit the same changes, perhaps a litte more exaggerated. There is also a small amount of deformity visible on the great toe on the left. Scattered over the soles of both feet are large, oval, oozing areas covered with a red, smooth, moist base. These lesions have a foul odor. No inflammatory reaction has been noted about these lesions. There are similar lesions over both heels. On the lateral aspect of the right foot, and in the mesial aspect of the left foot, are thin, bluish-like scars representing healed areas. Sweating of the hands and feet is present, though not marked. Dermographism is not present. Nikolsky's sign, even on the plantar surfaces of the feet, is negative. The mouth is normal except for the tongue, which shows definite atrophy and whitish leukoplakial areas along the lateral portion. The buccal mucosa is normal. Palpation shows no thickening. Vaginal examination was not done. The remainder of the physical examination was negative. Repeated examinations were negative for fungi on the nails of the hands and feet. Smears of the oozing skin lesions show staphylococci.

Fig. 3.—Scars on side of right foot.

The patient has been under observation for some time and the nails show no definite improvement, but there has been some spontaneous healing of many of the small bullous lesions about the toes and of several of the larger lesions on the plantar surfaces of the feet. The patient believes that she got the greatest relief from roentgen therapy. At the present time she is wearing a special shoe and a protective cotton boot.

COMMENT

This case, then, showed pachyonychia, palmar and plantar keratoses, mucous membrane changes, and a generalized degree of mild ichthyosis. Our patient was disturbed, outside of cosmetic disfigurement, by the presence of the large, painful bullae, which so frequently became secondarily infected. During the period of observation of the patient we could watch the spontaneous healing of some of the bullae. From the pathogenesis of this condition, repeated trauma and friction seem to be a predisposing factor for the development of these bullae. With the institution of bed rest, elevation of the feet, and protection of the feet with cotton boots, and special shoes when walking, the patient secures some relief. Because of the presence of areas of leukoplakia in her mouth, the patient was cautioned about the importance of good dental care, the avoidance of tobacco, and any irritating articles in her diet. In the reports in the literature, no cases of malignancy superimposed on this condition have as yet been described.

19 West Seventh Street.