CONGENITAL ECTODERMAL DEFECT, WITH REPORT OF A CASE *

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This case is reported not only because of its apparently extreme rarity, but also because it may serve as a stimulus to further investigation of the factors responsible for defective development of the ectoderm. A convenient opportunity is also afforded to discuss certain phases of the function of the skin on which modern physiologists and dermatologists still disagree. Our case is the second of its kind reported in the American literature and the sixth reported in the world's literature. It stands absolutely unique, however, in the fact that the developmental defect occurred in a female; all other cases reported occurred in the male. The significance of the latter fact will be apparent from subsequent discussion. It would seem, however, that the extreme rarity of this type of case is more apparent than real. All evidence in the literature points to the fact that the extremely high grades of ectodermal defect are not excessively rare. A larger number of reported cases could undoubtedly have been classified with the group under consideration had their investigation been sufficiently complete. Nor are these cases of purely academic interest as the experience of the patients and their medical advisers attest.

Cases of ectodermal defects of a partial character, such as congenital aplasia of the teeth, hypotrichosis, or even congenital absence of circumscribed patches of skin, are no longer medical curiosities. Congenital aplasia of the teeth is reported in the very earliest medical writings and numerous references to it are found in the later medical literature. True congenital hypotrichosis is distinctly more rare, and the majority of cases reported as such are undoubtedly of the alopecia totalis type, in which the fetal hair is formed but later drops out and does not grow again. The work of Waelsch,¹ Pinkus² and others, and notably the careful studies of Buschke,³ have familiarized us with true hypotrichosis. A combined absence of the teeth and hair is found in still rarer instances. Circumscribed skin defects of a congenital nature

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1. Waelsch, L.: Ueber Hypotrichosis (Alopecia congenita), *Arch. f. Dermat. u. Syph.* **152**:63, 1910.

2. Pinkus: Ein Fall von Hypotrichosis (Alopecia congenita): Quoted by Waelsch.

3. Buschke, A.: Klinische und experimentelle Beobachtung über Alopecia (Hypotrichosis) congenita, *Arch. f. Dermat. u. Syph.* **108**:27, 1911.

are reported in limited numbers. Abt,⁴ in 1917, collected thirty-four cases in the literature, and added another of his own. Almost any region of the skin was involved in one or the other of these cases, but the defects were most frequently noted in the scalp.

In one group of cases of high grade congenital ectodermal defect reported in the literature there is not only a congenital absence of the teeth and a hypotrichosis, but also a total absence of sweat glands and an almost total absence of sebaceous glands. This combination gives the cases a stamp which places them in a class by themselves. To date there are only five cases of this type on record.[†] The first of these was reported in 1883 by Guilford,⁵ an American dentist (Fig. 1). As he said, he had the pleasure of presenting a very remarkable man. The

Fig. 1.—Guilford's patient. Note the prominent forehead, alopecia, and depressed nasal bridge.

patient was 48 years of age, and in perfect physical health, never having been confined by sickness for a single day. He had been edentulous from birth; he was totally lacking in the sense of smell, and almost devoid of the sense of taste; the surface of his body was destitute of the fine hair that should cover it, and he had never per-

4. Abt, I. A.: Congenital Skin Defects, *Am. J. Dis. Child.* **14**:113, 1917.

[†] Since this article has gone to press I have noted an abstract of an article by J. Strandberg on "Arrested Development of Ectoderm," in *J. A. M. A.* **73**:873 (Sept. 13) 1919, reporting what appears to be a case analogous to those herein described. The original case report appeared in *Nord. med. Arch.* **51**:1 (Oct. 18) 1918, *Int. Med. Sec.*, No. 1. As yet I have not had access to the original article.

5. Guilford, H. S.: A Dental Anomaly, *Dental Cosmos* **25**:113, 1883.

spired. Tendlau,⁶ in 1902, reported a case presenting very much the same characteristics as that of Guilford (Fig. 2). He concerned himself chiefly, in his discussion, with the absence of sweat glands in the skin and erroneously interpreted the lack of development of the skin as an atrophy. In fact he heads his article: "Concerning Congenital and Acquired Atrophia Cutis Idiopathica," and discusses elaborately several cases of acquired atrophy of the skin. Wechselmann and Loewy,⁷ in 1911, subjected Tendlau's case and two others (Figs. 3 and 4) to a most careful study. Their investigation was devoted chiefly to the mechanism of production of insensible perspiration. Christ,⁸ in 1913, added a further case and discussed the relationship of the various ectodermal structures to one another in the animal kingdom. To this classic group I wish to add our own case.

Fig. 2.—Tendlau's patient. Note the features characteristic of congenital ectodermal defect. Mustache and beard well developed as in all other male patients.

REPORT OF CASE

Case 240873, I. C., a woman, aged 21, born in England, stenographer, came to the clinic primarily to have some plastic work done on her nose, which was rather flat in the bridge.

Examination.—Examination revealed a person of a rather frail build, but on the whole presenting a fair average physical development, excepting only that of the skin and its appendages. The skin was exceptionally fair, smooth, thin, pli-

6. Tendlau, B.: Ueber angeborene und erworbene Atrophia cutis idiopathica. *Virchows Arch. f. path. Anat.* **167**:465, 1902.

7. Wechselmann, W., and Loewy, A.: Untersuchungen an drei blutsverwandten Personen mit ektodermalen Hemmungsbildungen, besonders des Hautdrüsensystems, *Berl. klin. Wchnschr.* **2**:1369, 1911.

8. Christ, J.: Ueber die kongenit. ektodermalen Defekte und ihre Beziehungen zu einander; viktisierende Pigment für Haarbildung, *Arch. f. Dermat. u. Syph.* **116**:685, 1913.

able and dry, and showed the superficial veins plainly. There was a notable absence of the lanugo hair on all parts of the skin, and only a few straggling hairs were seen on the scalp. The genitalia and axillae were practically hairless. About the labionasal folds and on both upper and lower lids was a number of soft split-pea sized papules strongly suggesting small sebaceous cysts or milia. On the back were a few papules and pustules and the scar of an intramuscular

Fig. 3.—Wechselsmann's patient. Note the characteristic profile of patient practically duplicating that of our patient in Figure 6.

Fig. 4.—Another patient of Wechselsmann.

arsphenamin slough. The nails of the hands and toes presented uniform fine longitudinal furrows. The vermillion border of the lips was poorly defined. On physical examination it was found that both jaws were devoid of visible teeth. The roentgen-ray report, however, stated that several "fragments" of teeth could be seen in the right upper jaw. The patient gave a history of having had several defective teeth removed from her upper jaw; she had never to her

knowledge had any teeth in the lower jaw. The rhinolaryngologist reported an atrophic rhinitis, and a destruction of the turbinates on both sides with much pus and crusting. No adenopathy could be determined. The forehead was unusually prominent and the bridge of the nose flat (Figs. 5 and 6). Aside from these two features, there were no osseous changes which even remotely suggested any of the osseous stigmata of lues hereditaria. The nervous mechanism was apparently normal, only a slight hyperactivity of the knee jerks being noted. The mentality was fair, probably somewhat above the average. The viscera were normal.

Fig. 5.—(240873.) Author's patient. The scalp is covered by a wig, which hides the almost total alopecia.

History.—The patient gave a history of two aunts and one uncle having died of tuberculosis; one aunt suffered from epileptic attacks and died insane. The father was said to have had syphilis. There was no history of any other member of the family having been afflicted as was our patient. One sister died at 1½ years of age; one brother and one sister were living and perfectly well.

The patient began to have trouble with her nose when she was 9 months old. She had never perspired even in the hottest weather. She had suffered

from a variety of ailments all her life, but a definite diagnosis of her condition had not been made. For many years she had been taking medicine by mouth regularly, but had never known exactly for what purpose. She had had two salvarsan injections, evidently on the supposition that she was afflicted with heredo syphilis.

Histologic Report.—The patient, after some persuasion, submitted to biopsy. The tissue was taken from the outer aspect of the left arm; a piece at least three-fourths inch in length was excised from deep in the subcutaneous fat. While we realize that in theory the total absence of hair, sebaceous glands, and

Fig. 6.—Lateral view of author's patient.

sweat glands can be demonstrated only by an examination of a number of pieces of tissue from different parts of the body, such an examination in this case was out of the question.

For purposes of comparison an exactly similar piece of tissue was taken from the skin of the outer side of the arm of a normal adult female (surgical amputation (Fig. 7). In order to identify and compare the anlagen of the pilosebaceous and sudoriferous systems in the human embryo, a specimen was taken from the outer sides of the left arms of human embryos measuring 5 cm., 7 cm., 10 cm. and 25 cm., and from a fetus at 7 months.

The epidermis and cutis of the patient were absolutely normal as compared with a section of normal skin, except for the total absence of hair follicles, sebaceous glands and sweat glands. The cutis in the case of ectodermal defect displayed a normal vascularity, made artificially conspicuous by the absence of the usual association with pilosebaceous and sudoriferous structures. There were no increase or diminution in the collagenous or elastic tissue of the cutis, no signs of an inflammatory process, no abnormal elongation of the occasional rete pegs, and no cellular infiltration of any type. The sections showed the tissue far into the subcutaneous fat. There were no cell inclusions, either solid or tubular, of the type generally recognized as anlagen of sweat glands in the familiar picture of syringocystoma (Fig. 8).

The comparison of the skin of the fetal arm with that of our patient indicated that in the site from which the specimen was excised there are in late fetal life and immediately after birth many lanugo hairs with their associated sebaceous glands, and numerous sudoriferous glands which have not quite reached their final development by the seventh month of fetal life (Fig. 9). The various structures representing the anlagen of these glands, as illustrated in Figures 10 and 11, were carefully searched for in the material from our patient, but as seen in the photomicrograph (Fig. 8) they were totally absent.

In the histologic material, therefore, we find that the pathologic process in such a patient is neither an atrophy nor a simple rudimentary development, but consists in a total absence of pilosebaceous and sudoriferous systems and their anlagen, at least in adult life. Whether at some stage in the embryonic development in our patient such anlagen appeared only to undergo regression or to be destroyed, it is of course impossible to state with positiveness from an examination of a specimen removed from the adult.

DISCUSSION

Wechselmann believes that congenital ectodermal defects of the type presented by our case are familial and inherited, and are analogous to the type of hereditary transmission seen in hemophilia and hemeralopia. The analogy may be allowed in so far as these diseases are transmitted through the female, but the defect need not always be transmitted to the male, as our case plainly shows. In other words, these congenital defects are not always recessive in the female. Whether or not hemophilia and hemeralopia are always recessive in the female is still a problem, despite the careful studies of Bulloch and Fildes.⁹ The familial transmission of the ectodermal defects is well shown by a family tree compiled by Wechselmann for his cases. From these findings he wishes to prove the recessive character of the ectodermal defect in the female. Our own case evidently disproves this supposition. Christ's case apparently also bears out the theory that this condition is familial. A familial history was not elicited in our case, possibly because of the insufficiency of the data obtainable. Most of the reported cases of ectodermal defect, like those of hemophilia

9. Bulloch and Fildes, quoted by Osler, W., and McCrae, T.: *Modern Medicine*, Philadelphia, Lea and Febiger 4:715, 1915.

Fig. 7.—Section from the arm of a healthy woman. Note the presence of sweat glands and the hair follicle.

Fig. 8.—Section from the arm of our patient, showing total absence of the sudoriferous and pilosebaceous systems.

and hemeralopia, are in the families of exceptionally fertile women. In Guilford's case the mother of the patient had married at 16 and died at 40, and during that time had given birth to twenty-one children. The mother of Tendlau's patient gave birth to fifteen children in two marriages, and the mother of Wechselmann's patients also bore fifteen children. This fecundity of the mothers of patients is not apparent in Christ's case nor in our own, each of these mothers having given birth to four children.

Syphilis as a causative factor deserves special attention. Familiarity with this striking group of cases, which present practically identical features, would hardly warrant an assumption of activity of the *Spirochaeta pallida* as it appears in the ordinary case of syphilis hereditaria. The activity of the spirochete in ectodermal defect could be explained only by assuming that the injurious influence had been suddenly exerted at the time of the third or fourth month of embryonal existence and had then rapidly subsided, which would seem rather unlikely. Christ reports that his patient was without doubt infected with syphilis. He does not say on what grounds he bases this assumption; the Wassermann reaction on the patient's blood was negative on one occasion. In discussing his case, he admits that he was at first inclined to believe that the developmental defect was of syphilitic origin. On discovering the familial tendencies, however, and on careful consideration of our present day conception of the genesis of congenital syphilis, he later came to the conclusion that syphilis is not a causative factor. In our case the history is rather suggestive of a syphilitic factor. The patient's mother told her that the father had acquired syphilis while serving in the British navy. The patient herself had been treated for syphilis, but evidently chiefly on the basis of the changes found in the nose, and probably by some one ignorant of the existence of the type of congenital defect under consideration. There were no other signs suggestive of syphilis hereditaria except those equally characteristic of this group of defects. The Wassermann test of the blood was negative. If syphilis was present in Christ's case or in our own, it was probably only incidental.

Guilford does not discuss the pathology of the skin of his patient. Tendlau, in a description of the skin of his patient, speaks of an atrophy, and in his report elaborately discusses other cases of cutaneous atrophy. In one paragraph he carefully notes, however, that nowhere does he see any regressive changes such as atrophy. Moreover, he concedes that there are no rudimentary nor partially developed sweat glands to suggest that the anlagen of these structures had ever existed. Wechselmann insists that there is no sign of atrophy in the skin but that it resembles the embryonal type. In our case there were no signs of regressive changes in the skin. There was an entire

Fig. 9.—Section from the arm of a 7-months-old fetus. The sweat glands and hair follicles are not yet fully developed, but they show plainly.

Fig. 10.—Section from the arm of a 10 cm. fetus. Note the epithelial buds.

absence of such cell inclusions as might warrant an assumption that embryonal vestiges of lanugo hair and sweat glands had ever existed. A striking feature in this group of ectodermal defects is the hypotrichosis described by all observers. Our patient's scalp contained only a few tufts of straggling, dry hair. The genitalia were also practically devoid of hair, and the lanugo hair was absent over the entire body. The aplasia of the teeth has been noted in all cases, and such teeth or dental "emboli" as there are, occur in about the same part of the upper jaw as in our case, probably replacing the incisors and canines. In the lower jaw in all cases studied by roentgen ray, teeth and the anlagen of teeth were completely absent. Wechselmann's article is illustrated by photographs of plaster models of the jaws, and copies of his illustrations are here reproduced (Fig. 12). Such teeth as there are, are broad at the base and pointed at the tip, showing a close resemblance to the canine teeth. They are widely separated. Our patient had never had a tooth in her lower jaw, and the upper teeth were imperfect and had been removed. Roentgen-ray examination showed "fragments" of teeth in the right upper jaw.

The gross characteristics of the skin are in all instances very similarly described by the various authors. The skin is said to be thin, pliable, dry, unusually white and transparent, with marked prominence of the superficial veins. The mouths of the follicles are distinctly visible in some locations, but not in others.

Wechselmann noted a few milia about the nose in his cases. These general characteristics of the skin were apparent in our patient, and we also noted the milia about the nose. Christ describes a condition in the nasal and orbital region of his patient, which reminded him of a xeroderma pigmentosum; Touton, who saw this patient, was also reminded of the same disease, but only in a mild form. This condition was not described in any of the other patients, nor was it seen in our own.

The nails of Tendlau's patient were well developed. Wechselmann notes that in one of his patients the nails were normal. A definite trophic deformity of the nails was noted in our patient; its etiologic basis is unexplained.

Atrophic rhinitis was present in the patients described by Tendlau, Wechselmann and Loewy, and Christ. Guilford does not describe this condition, but he specially mentions the fact that his patient had lost the sense of smell and largely that of taste. In our patient a well marked atrophic rhinitis, with atrophy of the mucous membrane of the pharynx and larynx, was noted, the latter producing a distinct hoarseness. In addition to this a nasal examination revealed destruction of the turbinates on both sides with much crusting, probably a result of secondary infection.

The vermillion border of the lips in our case was not so sharply defined as in the normal person. The detail is not specifically noted in any of the other cases.

Prominent frontal bosses and a depressed nasal bridge were the chief bony changes, as all the pictures well show. Our patient came

Fig. 11.—Section from the arm of a 25 cm. fetus. The immature sweat glands and hair follicles are clearly shown.

Fig. 12.—An illustration of Wechselmann's plaster model of the jaws and dental "emboli" of one of his patients.

for treatment primarily to have the shape of her nose improved. A plastic operation was attended by a considerable degree of success.

The feature which distinguishes this group of cases of ectodermal defect from all others, is the total absence of sweat glands and the

almost complete absence of sebaceous glands. On this defect are based the symptoms which give them their special practical and theoretic interest. All the patients state that they do not perspire even in the hottest weather, and to this our case is no exception. Guilford's patient was a cobbler by trade and lived in a farming community. During the summer months he worked with various farmers as a field hand. Whenever so employed it was always necessary to engage a boy to carry water from some neighboring brook or well to pour over him to keep his clothing wet. If the arrival of the water happened to be delayed, he became weak and collapsed from the heat. Tendlau's patient wore a wet shirt during the hot months. This he kept continually moist under a pump. When the heat became too great he frequently collapsed. This patient from childhood had been in the habit of eating only cold meals during the hot weather, because he had noticed that he became feverish on the ingestion of hot foods. A similar story is told by the other patients, and all of them were forced to change their original occupation because they could not tolerate heat. Because of these peculiarities one of Wechselsmann's patients was temporarily regarded as a malingerer and the other as suffering from phthisis. Our patient, while she could not perspire, suffered less hardship probably because as a woman she was less subject to exposure and hard physical work.

The mental condition of Guilford's patient was reported as normal; the patients of Tendlau, Wechselsmann and Loewy, and Christ were of a distinctly inferior mentality; our patient was of average mental acuity, if not somewhat above it.

Congenital ectodermal defect apparently does not interfere with the longevity of the patient. Guilford's patient was 48 when his case was reported. Wechselsmann studied the patient originally described by Tendlau when the patient was 57 and in good general health.

Much interesting experimental work has been done on these cases and much light thrown on the physiology of the skin. Vierordt¹⁰ estimates that the relative values of different modes of heat loss are about as follows:

VALUES OF DIFFERENT MODES OF HEAT LOSS

Loss by	Per Cent.
Urine and feces.....	1.8
Expired air; warming of air.....	3.5
Vaporization of water from the lungs.....	7.2
Evaporation from skin.....	14.5
Radiation and conduction from skin.....	73.0

Of course, these percentages vary somewhat with the surroundings, but they tend to emphasize the importance of the skin as a heat regu-

10. Vierordt: Quoted by Howell, W. H.: *A Textbook of Physiology*, Saunders, 1913, Ed. 5, p. 933.

lating organ. While it is true that the lung normally dissipates the greater part of the heat in some of the lower animals, and does so vicariously very effectually in others, in man this function of the skin is apparently vicariously taken up by the lung in a very unsatisfactory manner. The cases of ectodermal defect previously reported have shown the relationship that exists between the lungs and the skin as heat regulating organs, and have offered ideal conditions for experimentation. Tendlau and Loewy have definitely shown the importance of the skin as a heat regulator and the inability of the lungs vicariously to assume this function in man.

On one occasion Tendlau's patient, on drinking a pint of milk at 104 F., was observed to undergo a rise of temperature of 0.4 degrees within five minutes, followed by a further rise of 0.9 within the next fifteen minutes. The temperature then gradually dropped to its first level. Very noticeable were also the rises in temperature following exposure to the sun. On a perfectly quiet day the temperature rose at the rate of 1.8 F. every ten to fifteen minutes. This rate was increased by physical exertion, extreme degrees of heat, and an increased humidity. On one occasion the patient was exposed at 12 o'clock noon while the air temperature was 89.6 F. After an exposure of twenty minutes to the sun the body temperature rose 3.7 F. Simultaneously mild, subjective symptoms appeared, such as are seen in a febrile person; that is, slight restlessness, sensation of heat, and headache. Similar experiments were repeated frequently, always with the same results. The highest temperature observed on this patient was accidentally obtained on a hot day. It rose on this occasion to 105.4 F.

Tendlau, by observations on his patient, was reminded of the behavior of poikilothermic animals. He, therefore, determined if possible to obtain the temperature changes in the opposite direction by applying cooling procedures. Prolonged cold baths were used, but no abnormal reduction in temperature was observed. Similar experiments were carried out by Loewy¹¹ on his patients with similar results. Both observers carefully watched the respiratory function in the persons in whom an increased amount of heat dissipation was made necessary by the experiments. Tendlau's patient was carefully examined by Zuntz relative to his respiratory behavior under these conditions. Zuntz found an increased respiratory volume and rate at all times, analogous to the results obtained in nonsweating animals. Loewy confirmed these findings by further experiments.

11. Loewy, A., and Wechselmann, W.: *Zur Physiologie und Pathologie des Wasserwechsels und der Wärmeregulation seitens des Hautorganes*, Virchows Arch. f. path Anat. **206**:79, 1911.

In an endeavor to determine the absorbing power of the skin of such a patient Tendlau employed two methods. In both instances he used a potassium iodid ointment, which in all probability does away with the objection that some of the drug was absorbed by inhalation. When this ointment was applied loosely under a bandage, no absorption of the potassium iodid occurred. On the other hand, when the ointment was rubbed into the arm and the arm bandaged, potassium iodid could be demonstrated in the urine in about six hours, which is no marked variation from the normal. Tendlau's first experiment tends to show that a certain amount of friction is necessary to produce any absorption. His experiments did not determine whether or not a certain amount of trauma is necessary to produce absorption, because it is impossible to say after thorough friction that the skin has not been sufficiently traumatized to produce a histologic break.

The mercurial inunction experiments performed by Wile and Elliot¹² on patients, and on rabbits by Schamberg,¹³ apparently definitely settle the fact that there is absorption through the skin when ointments are rubbed into it. Many other observers believe that oily preparations are quite readily absorbed through the skin. Because of the total absence of sweat glands and the probable total absence of sebaceous glands on the arm of Tendlau's patient, the findings in his inunction experiments constitute experimental evidence that the glands of the skin are certainly not essential and in all probability not necessary to the absorption of drugs in the normal person.

Merely as an illustration of the wide divergence of opinion on the mode of absorption of substances through the skin, Landois,¹⁴ on the one hand, believes that the mercury globules pass into the hair follicles and ducts of the glands, where they are affected by the secretion of the glands, and transformed into a compound capable of absorption. This view is supported by Neumann,¹⁵ and Auspitz,¹⁶ and concurred in by Duhring,¹⁷ Robinson,¹⁸ Stelwagon¹⁹ and others. On the other

12. Wile, U. J., and Elliot, J. A.: Mode of Absorption of Mercury in the Inunction Treatment of Syphilis. Preliminary Report, *J. A. M. A.* **68**:1024, 1917.

13. Schamberg, J. F., Kolmer, J. A., Raizess, G. W., and Gavron, J. L.: Experimental Studies of the Mode of Absorption of Mercury When Applied by Inunction, *J. A. M. A.* **70**:142, 1918.

14. Landois, L., and Stirling, W.: *A Textbook of Human Physiology*, Ed. 4, Philadelphia, Blakiston, 1891, p. 571.

15. Neumann, quoted by Stelwagon: *Diseases of the Skin*, Philadelphia.

16. Auspitz: *Ibid.*

17. Duhring, L. A.: *Ibid.*

18. Robinson: *Ibid.*

19. Stelwagon, H. W.: *Diseases of the Skin*, Philadelphia, Saunders, 1914, 1250 pp.

hand, Rindfleisch,²⁰ Fleischer,²¹ Voit²² and others believe that the particles of mercury pass directly through the epidermis. The latter view appears to be most strikingly supported by the observation of Voit, who found globules of mercury between the layers of the epidermis and in the corium of an executed criminal, into whose skin mercurial ointment had been rubbed previously.

With reference to the question of the mechanism of perspiration, on which these cases throw some light, physiologists are well agreed that sensible perspiration is a true secretion, a product of the sweat glands. On the production of insensible perspiration, however, they have disagreed ever since the discovery of the sweat glands by Malpighi and Stenson. The arguments pro and con brought forth by various observers in support of their views need not be discussed here. Suffice it to say, that in a considerable number of modern textbooks on physiology and dermatology a discussion on the mode of production of the insensible perspiration is either entirely omitted or no decided stand is taken with regard to it.

The experiments undertaken by Loewy on Wechselmann's patients with a total absence of sweat glands tend to substantiate the marked importance of the physical process of transudation as a factor in the loss of moisture by the body under ordinary conditions. By carefully controlled experiments Loewy found that under conditions in which only insensible perspiration could be given off, the two patients with ectodermal defect eliminated a minimum of 123 gm. and 242 gm., and a maximum of 436 gm. and 600 gm., respectively, as compared with an average minimum of 112 gm., and an average maximum of 700 gm., in five healthy persons. This result substantiates the fact that so long as the water is given off in an insensible manner, the quantity of water given off by the healthy person is no greater than that given off by those without sweat glands. The sweat glands, therefore, are not perpetually active, but only become so under special stress. Diffusion and not secretion is, therefore, the primary method of water elimination through the skin. Loewy further demonstrated on his case, by the method of Aubert, that there is a point at which perspiration is still insensible but at which the sweat gland takes some part in its production. This point varies in different persons and also on different parts of the body in the same person. It was impossible to carry on physiologic experiments on our patient, largely because of her indifference.

20. Rindfleisch: *Ibid.*

21. Fleischer: *Ibid.*

22. Voit, V., quoted by Landois, L., and Stirling, W.: *A Textbook of Human Physiology.*

CONCLUSIONS

1. There exists a group of typical cases of high grade congenital ectodermal defect, the extreme rarity of which, suggested by the scanty literature, is probably more apparent than real.

2. The patients in this group all present a facies very closely resembling that of heredosyphilis.

3. The influence of syphilis in the production of these congenital defects is probably nil.

4. The reported cases of this group of ectodermal defects have exhibited a total absence of sweat glands, an almost total absence of sebaceous glands, a hypotrichosis with absence of lanugo hair, and a dental aplasia.

5. Such patients suffer from a disturbance of the heat regulating mechanism, dependent on the inability of their skins to eliminate the necessary amount of water to keep the temperature level constant under varying external conditions.

6. Valuable contributions to our conceptions of certain phases of the physiology of the skin have been made by experiments on patients with this type of congenital defect.

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