HEREDITARY ECTODERMAL DYSPLASIA

BY

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Of late years, a considerable amount of attention has been given in America and Germany to that class of case which has been variously termed 'combined defects of hair and teeth,' 'malformations simultanées du système dentaire et du système pileux,' and 'dystrophy of the hair and nails.' More recently the more comprehensive term 'congenital ectodermal defect' has been used, and still more recently, Weech²⁵, to whom I am indebted for much of the bibliography of this subject, has suggested 'hereditary ectodermal dysplasia' as the most suitable title.

The following is the case report of a child who shews what Goeckermann⁹ and Mackee and Andrews¹⁷ term 'high grade' or 'major' defect, or as Weech prefers to call it, the 'anhidrotic type of hereditary ectodermal dysplasia.'

CASE REPORT.

A.B., aged 5 years 9 months, was first seen at an examination prior to attending school. He is the first of four children; two other children, aged three years and eight months respectively, are alive and well; one child died of pneumonia. Both mother and father are healthy and there is no evidence of similar anomalies in any other member of the family, excepting one aunt on the mother's side, who is said to have had a double set of teeth when a child, but it has not been possible to confirm this statement.

He was born at full term and, apart from an absence of hair on the scalp, was thought to be a normal child. He cut his first teeth, which occupy the normal position of the upper incisors, at 18 months; the four temporary first molars between two and three years, and the four permanent molars during the past three months. He began to walk at one year and ten months.

When three months old he was admitted to hospital suffering from convulsions of unknown origin. Apart from the high temperature (107.8°) nothing abnormal could be found in what appeared to be an otherwise healthy infant. After sponging with iced water the temperature fell to normal and the child was discharged well on the sixth day (18.7.23). He has had nine fits at intervals of a few weeks during the past summer; they all occurred between the months of May and October. He has also had attacks of epistaxis. The boy is said to have had no hair on the scalp for several months after birth, but a fine growth of hair can be seen in the photograph taken at the age of six months (Fig. 1). He never perspires even in the hottest weather, when he suffers extreme discomfort. During the summer months he frequently sleeps on the bare floor divested of all his clothes, and in the daytime often lets the cold water tap run over his head, from which procedure he appears to derive much benefit. In spite of the very imperfect dental development, he has little difficulty in mastication excepting with hard foods such as crusts.

Physical examination (17.12.28).—The patient was a slightly built child measuring 40 in. and weighing 33 lb. (normal for age 42 in. and 44 lb.). The most striking feature was the curious facies. The scalp, the skin of which was very dry and glossy, was covered with a sparse growth of very fair, floss-like hair. Large blue veins, which contrasted strikingly with the white, thin skin, coursed over the scalp but were especially well-marked in the fronto-parietal region, as shewn in Fig. 2. The eyebrows and eyelashes, although present were extremely scanty and there was no hair whatever to be seen on the rest of the body. The auricles were well formed and equal in size but stood out obliquely from the sides of the head. The bridge of the nose was considerably broadened and depressed and the skin here, being stretched to a greater extent than elsewhere on the face, had a bluish appearance. The nares were well developed and although there was some excoriation of the nostrils, there was no atrophic rhinitis or ozæna. The mucous membrane

of the mouth and pharynx appeared normal and there was an average amount of salivary secretion. The voice was not unduly shrill but the absence of teeth interfered considerably with articulation. The lips were thick and everted but the vermilion border was well defined. Both jaws were under-developed so that the face had a triangular appearance, having the forehead as base and the chin as apex. There were ten teeth present; six in the upper and four in the lower jaw; two occupied the position of the upper incisors; there was one first temporary molar on either side in both jaws and posterior to each of these temporary teeth there was one, first permanent tooth. Using letters in alphabetical order for the temporary set and numbers for the permanent set his dental formula may be represented as follows:—

$$\begin{array}{ccc} 6D & 1\ 1 & D6 \\ \hline 6D & & D6 \end{array}$$

The molars were well formed and enamelled. The teeth in the incisor region, however were cone-shaped, the free end being sharply pointed, and were yellow in colour. The gums

Fig. 1.—A.B., aged six months. There is a growth of fine hair on the scalp; the thick, everted lips and the projecting ears are already noticeable.

where the teeth were absent were retracted and had a sharp narrow edge. The thin, parchment-like character of the skin, the large superficial scalp veins, the sunken nasal bridge and the under-developed jaws combined to give the child an old-mannish look. The bony prominences of the skull were well-marked, especially the supra-orbital ridges but the fontanelle was closed. The nipples were present but no glandular substance could be felt. Examination of the chest and abdomen revealed no abnormality. The child was forward mentally but seemed of an unduly sensitive nature. The genitalia were normal, and the nails on both hands and feet were well developed and smooth. There was no deficiency of lachrymal secretion but on no occasion was any sensible perspiration seen.

Investigations.—The Wassermann reaction was negative. The blood pressure, systolic 80, diastolic 45 mm. Hg. (normal 92.6 and 67.3). The colour index was rather low but examination of the blood revealed no gross abnormality. The sugar tolerance, urine and urea concentration tests were all normal, as were also the fundus and fields of vision and the skiagrams of the sella turcica and the long bones: no unerupted teeth were to be seen in the skiagrams. A piece of skin an inch long and a quarter of an inch wide was excised from the anterior chest wall in the anterior axillary line at about the level of the nipple. A series of a large number of sections was examined but no sweat or sebaceous glands or hair follicles could be found.

REVIEW OF THE LITERATURE.

It is surprising that although the first case of hereditary ectodermal dysplasia was reported in England in 1842 by Thurnam²³, the subject has received most attention elsewhere. It is also remarkable that although the anomaly is a congenital one it has but seldom been described during childhood. The literature abounds with cases of isolated ectodermal defects but there is one type of patient who deserves to be set in a class apart, for as Goeckermann says, 'In one group of cases of high grade congenital ectodermal defect . . . there is not only a congenital absence of teeth and a hypotrichosis, but also a total absence of sebaceous glands. This combination gives the cases a stamp which places them in a class by themselves.' The following cases come into this class and have already been reported in the literature :—

TABLE OF PREVIOUSLY REPORTED CASES.

Author.	Date.	Sex.	Age.	Race.
1. Thurnam ²³	1842	male	58 years	English.
2. Thurnam	1842	male	13 ..	English.
3. Williams ²⁸	1842	female	15 ..	English.
4. Guilford ¹⁰	1883	male	48 ..	American.
5. Ascher ¹	1898	male	15 ..	German.
6. Ascher	1898	male	21 ..	German.
7. Tendlau ²¹	1902	male	42 ..	German.
8. Loewy & Wechselmann ¹⁶	1911	male	—	German.
9. Christ ³	1913	male	13 ..	German.
10. Gibbs ⁷	1915	male	7½ ..	English.
11. Gibbs	1915	male	6½ ..	English.
12. Strandberg ²³	1918	male	39 ..	Swede.
13. Goeckermann ⁹	1920	female	21 ..	English.
14. MacKee & Andrews ¹⁷ ...	1924	male	14 ..	Jew.
15. Weech ²⁵	1929	male	14 ..	Jew.

The case which I have reported is therefore the sixteenth to be described, although it is probable that there are others which belong to this group but which, for lack of sufficient information cannot be included in this list. Hutchinson¹² shewed a boy aged three years at the Royal Medical and Chirurgical Society in 1886 whose description conforms very closely with those above, but as no note was made on the absence of perspiration, this patient has been omitted.

As stated previously, instances of various kinds of ectodermal dysplasia occur very frequently in the literature, and reports of dental anomalies of number, shape and date of eruption are especially common.

These dental variations may or may not be associated with abnormalities in the other ectodermal structures. Wieting²⁷ described a boy of twelve years who had been completely edentulous from birth, but who shewed no other peculiarity. Hopson¹¹ also has reported a similar case. In others the teeth and hair are affected simultaneously as was the case in Rushton's¹⁸ patient,

an Armenian boy of sixteen, who had never had any teeth and whose hair was very soft and white, resembling wool. Darwin⁵ also noted this tendency both in human beings and in animals. He speaks of Julia Pastrana, a Spanish dancer, who 'was a remarkably fine woman, but she had a thick masculine beard and a hairy forehead' and 'she had in both upper and lower jaw an irregular double set of teeth one row being placed within the other.' He also mentions that three hairless Egyptian dogs had many deficient teeth, and 'that the two orders of mammals namely the Edentata and Cetacea which are the most abnormal on their dermal covering are likewise the most abnormal either by deficiency or redundancy of teeth.'

Kjaer¹⁵ described a man in whom the deciduous teeth had developed very imperfectly and the permanent teeth had never erupted, who was born without finger or toe nails. Eisenstadt⁶ had three brothers suffering from a dystrophy of the hair and nails with no other ectodermal defect. Sufficient has been said to show that any combination of ectodermal defects may occur, but at the same time it seems desirable to differentiate from the others what Goeckermann calls 'high grade defect' and Weech 'the anhidrotic form of hereditary ectodermal dysplasia.'

NOSOLOGY.

Hitherto most of these cases have been described as examples of 'congenital ectodermal defect,' but as Weech says this is neither the most suitable nor the most comprehensive title. As this author points out 'hereditary' is to be preferred to 'congenital' for whereas the latter may connote either something existing at or dating from birth, 'hereditary' can only be applied to a condition which is capable of being transmitted from parent to offspring, and is therefore at the same time more comprehensive and precise. 'Dysplasia' is preferable to 'defect' for similar reasons. Weech's suggestion that this condition should be called 'the anhidrotic form of hereditary ectodermal dysplasia' has therefore been adopted throughout this article.

SYMPTOMATOLOGY.

In reviewing the signs and symptoms attention should be drawn to the fact that although those recorded by Gibbs and the one reported here are the only examples of the condition in young children, several were seen at or about puberty, when the hypotrichosis which manifests itself later in the absence of beard, whiskers, pubic and axillary hair is not the prominent feature it is in adult life. This may partly account for their not being recognized during childhood.

Skin.—In all the cases described, the thin, smooth glossy skin has been a uniform feature and in all except the one described by Weech the skin has been fair, whereas in his boy 'the skin was everywhere rather dark.'

Histological examination of the skin was carried out by Thurnam, Tendlau, Goeckermann and in the present case, and although the skin was excised from such widely scattered areas as the arm, axilla, chest wall, epigastrium and pubis, the reports conform generally to the description given by Erasmus

Wilson of Thurnam's patient :—' The peculiarities of the portions of skin . . . are their whiteness, thinness and softness, and the tenuity of, and absence of pigment in the epiderma . . . The chief peculiarity, however, is . . . a state of extreme laxity and apparent atrophy of the derma, with a total absence of fat and the usual filling up material.' He goes on to say that ' the sudoriferous glands are not wanting but their ducts are unusually delicate and deficient

Fig. 2.—A.B., aged 5½ years. The dry, brittle, sparse hair, the large veins, the thick lips, the projecting ears and frontal eminences are well shown

in structure.' This last opinion was not confirmed by Bowman, who also examined the skin from the same patient and ' did not obtain satisfactory evidence of sweat or sebaceous glands or hair follicles.' Goeckermann reported a ' total absence of pilo-sebaceous and sudoriferous systems and their *anlagen*' in his sections. In the case recorded here none of these structures could be seen in the sections. From the patient's point of view this absence of sweat glands is of the utmost importance, since it is responsible for his extreme discomfort in hot weather.

Guilford¹⁰ describes how, in order to keep cool in hot weather, his patient had to employ a boy to pour water over his clothing as soon as it became dry,

and if there happened to be any delay in the arrival of the water he became weak and would be nearly thrown into a spasm by the intense internal heat. Tendlau's own description is :—' Wurde die Hitze zu stark, so stellte sich heftigen Blutandrang nach dem Kopfe ein, und er musste dann die Arbeit aussetzen, ja oftmals brach er fast bewusstlos bei derselben zusammen.' I have described how my patient when only three months old had an attack of hyperpyrexia accompanied by fits and that during the summer he still has the fits. Tendlau carried out certain experiments on the excretory function of the skin in his patient and found that pilocarpine had no sudorific effect. He also noted that five minutes after drinking hot milk, the patient's temperature rose from 36.9° to 37.1° C, and that on standing in the sun for 20 minutes when the air temperature was 32° C., the patient's temperature rose from 36.7° to 38.8° C., the rise being accompanied by severe headache and discomfort. Loewy and Wechselmann made observations on the evaporation and excretion of water from the skin in their patients.

Fig. 3.—A.B. Showing the sharply pointed teeth in the upper incisor position.

Skin eruptions.—Papular lesions on the face have been described by several writers. Loewy and Wechselmann and Goeckermann diagnosed them as milium; Christ thought it similar to xeroderma pigmentosa; Strandberg thought it was of pilo-sebaceous origin, while MacKee and Andrews, who examined them histologically, state that 'the lesions appeared to be the result of large, superficially placed, degenerated sebaceous glands, together with hyperkeratosis, dilated follicular orifices, and surrounding acanthosis.' Epidermolysis bullosa has been noted in association with certain congenital defects, but not as yet in the anhidrotic type.

Teeth.—Abnormal dentition is a constant feature in these patients and affects both temporary and permanent sets, in number, form and date of eruption. Some are edentulous as was Guilford's patient, but more usually a few of each set appear. In several, delayed eruption has been noted and deformed teeth are common, the ones so affected being those which occupy the upper incisor position, although it is difficult to tell whether they are incisors or canines, as they occupy the position of the former, while resembling the latter more closely in shape. These teeth are often yellow and cone shaped; sometimes the free end is sharply pointed and tusk-like, or as MacKee and Andrews say, 'gimlet-like.' The molars, though often deficient in number, are normal in form.

Hair.—The hair on the scalp is fine, fair, dry and sparse. Some patients had no hair for several months after birth; others had a feeble growth which did not improve as the child grew older. The sparseness is very marked in all cases, so that the dry, glossy skin shines through between the hairs. It is interesting to note that no case of alopecia areata has been recorded among these patients, although the mother of Hutchinson's patient, who as stated elsewhere should probably come into this group, had been compelled from the age of six to wear a wig on account of a complete alopecia areata.

The lanugo hair on the body is generally absent, but the axillary and the pubic hair varies; some patients have no growth whereas in others it is normal. MacKee and Andrews and Strandberg noted that the pubic hair grew in the female distribution. Eyelashes and eyebrows, especially the outer third of the latter, are either absent or scanty. It is curious, as Weech points out, that even when there has been no beard or whiskers, the patient has usually grown a moustache.

Nails.—Dysplasia of the nails is nothing like so frequent as are abnormalities in the hair and teeth. Goeckermann mentions that the nails had longitudinal furrows; MacKee and Andrews that they had free lateral margins, which were flush with the skin instead of being imbedded, and Hutchinson's patient's were spoon-shaped, but in most cases they are normal.

Mammary glands.—Absence of the nipples and mammary glands were noted by Tendlau. Ascher's patient had flat nipples and breasts in which no glandular substance could be felt. Hutchinson wrote that his boy had no nipples, their sites being occupied by little patches of scar tissue and nothing like a mammary gland could be traced.

Lachrymal glands.—Thurnam's two patients are the only ones on record in which the lachrymal secretion was absent. His first patient had never been known to shed tears and 'the painful emotions generally relieved in this way had in him an unusually distressing appearance.' The lachrymal buds are of ectodermal origin and form at the third month of intrauterine life, *i.e.*, at the same time as the teeth are laid down, and it is strange that absence or diminution of this secretion has not been noticed in any other patient.

Nose, mouth and throat.—One of the main features, and one which plays an important part in the characteristic facies of these patients, is the broad, sunken nasal bridge or saddle nose, which is often so marked as to suggest a luetic origin. It is probable, however, that although it is not a syphilitic stigma, its mode of production is similar, and that it is due to an osteo-perichondritis, since there is almost always a history of nasal trouble in early life. Goeckermann's patient had had trouble since he was nine months old, and Weech states that his first patient had 'dry snuffles' at four months. In adult life there is frequently an atrophic rhinitis. The depression of the nasal bridge is accentuated by the projection of the frontal bones and more especially the supra-orbital ridges. Atrophic changes in the pharyngeal and laryngeal mucous membranes were marked in Tendlau's patient, whose voice was shrill and hoarse. The mucous membrane of the mouth and the salivary secretion are usually normal. MacKee and Andrews and Goeckermann described poor definition of the vermilion border of the lips, but this is not constant. Although Guilford's man had no sense of smell and that of taste was poor, the special senses are often unaffected, and when they are, it probably is the result of changes in the mucosæ.

The lips are everted, thick and muscular. This has been attributed to their excessive use in mastication rendered necessary by the absence of teeth, but in the photograph of my patient (Fig. 1) taken at the age of six months, this eversion is already well marked. Weech says that the epithelium on his patient's lips was thickened at nine weeks, and MacKee and Andrews noticed that the vertical lines on the lips were more pronounced than normally.

Ears and eyes.—The auricles of Ascher's first patient stood out obliquely from the sides of the head, and were unequal in size, the right being smaller than the left, while MacKee and Andrews noticed the lobes of the ears were small and attached to the cheeks.

Darwin asserts that the organs of sight and hearing are generally admitted to be homologous both with each other and with the various dermal appendages; hence these parts are liable to be abnormally affected in conjunction. Moreover, White Cowper¹ mentions that in all cases of double microphthalmia brought to his notice, he met at the same time with defective development of the dental system. On the other hand in all the subjects of the anhidrotic form of ectodermal dysplasia where a note of the special senses has been made, hearing and vision appear to be normal.

Nervous system.—It is interesting that although the nervous system is ectodermal in origin, gross malformations of this system have not been recorded. Tendlau, Loewy and Wechselmann and Christ mention that their patients were of feeble intelligence; in other instances, there were insane members in the family (Thurnam), but the greater number have been of average intelligence. My patient, indeed, although he has never attended school, reads better than many of the same age and is quick at mental arithmetic. Reflexes, sensation and motor power were normal in all those who have been examined.

Osseous system.—Although the osseous system is of mesodermal and not of ectodermal origin, certain peculiarities of the skull bones have been constantly present. Thurnam's patient, the only one on whom an autopsy has been held, shewed the dura mater firmly adherent to the calvarium which was very thick. All the writers mention the prominent supraorbital ridges and occipital and other eminences of the skull. There was no increase in the density of the skeletal bones or any epiphyseal changes in the boy here described, but the abnormally high blood calcium may have some, as yet unknown, significance.

ÆTIOLOGY.

Congenital Syphilis.—The facial aspect has often led to a tentative diagnosis of congenital syphilis, and indeed the pallor, large head, the eminences of which are very pronounced, the large superficial veins, saddle shaped nose, and sparse dry hair may easily suggest this diagnosis, which in some patients would appear to be still further supported by a history of papular eruption and rhagades on the face, a desquamation of the palms and soles, and of snuffles. The Wassermann reaction has never been found positive and the teeth, although malformed, bear no resemblance to Hutchinson's teeth. It has been pointed out that the ectodermal structures are developing during the third month of intra-uterine life, and, as Goeckermann says, it is very unlikely that if the spirochæta pallida were the causative agent its injurious influence would be exerted particularly at that time and then be suddenly removed; nor is it likely that it would at that early date have a selective action on ectodermal structures.

Endocrine dyscrasia.—Our knowledge of the role of the hormones during intra-uterine life is as yet very imperfect, but some aspects of these cases suggest an endocrine dyscrasia, and this hypothesis cannot therefore be wholly disregarded. Thurnam's patient had a bronchocele; the basal metabolic rate in Weech's patient was 10 per cent. below the normal and was associated with a small sella turcica; MacKee and Andrews and Strandberg noticed that the pubic hair grew in the female distribution. Barrett² found an increased sugar tolerance in a patient suffering from a dystrophy of the hair and nails, and Wheelon²⁶ described four patients, three of whom were females, in whom the upper lateral incisors were deformed or absent: the females shewed marked signs of pituitary insufficiency such as obesity, low basal metabolic rate and amenorrhœa, while the male exhibited signs of hyperfunction of the gonads. Moreover, Keith¹⁴ maintains that 'we have evidence that the growth of the canine teeth can be regulated by internal secretions or hormones,' and that 'the growth of the jaws is also influenced, during normal development, by secretions or substances thrown into the blood by such glands as the pituitary and thyroid.' Vichot²⁴ also thinks that an abnormal condition of the glands of internal secretions is one of the primary factors in dental regression and adds that 'les facteurs primaires créent les variations; l'hérédité semble les fixer.' That deficiency of the thyroid secretion leads to excessive dryness of the skin and hair is well known and that many of the glands affect the deposition of fat, the absence of which is such a marked feature in these subjects, is also

recognized, so that although it is difficult to attribute hereditary ectodermal dysplasia entirely to an endocrine dyscrasia, it is possible that it may play some part in its production.

Heredity.—The majority of these cases have shewn marked hereditary tendencies. The most complete family history illustrating the transmission of hereditary ectodermal dysplasia is that of the Kitzings, of whom two members were described by Ascher and one other by Loewy and Wechselmann. From the data given by these authors Weech has constructed the following pedigree chart. This chart shews that eight members of the family were affected, seven of whom were males and the sex of the other is not stated. Thurnam's

second patient was a cousin-german of the first, and Thurnam adds that the maternal grandmother had very delicate skin and a very limited amount of perspiration, but no peculiarity of the hair or teeth. Guilford's patient illustrates the tendency to transmit associated dysplasia of the hair and teeth although the patient himself was the only member of his family who did not perspire; his maternal grandmother had no teeth or hair; her daughter, the mother of the patient, was normal but her brother was edentulous from birth; the patient was one of twenty-one children, one of the later ones, and although he was the only one who was completely edentulous, some of his brothers had never erupted certain teeth; the patient was the father of eight children, the two youngest of whom, aged fourteen and sixteen years respectively, had many teeth absent; it is not stated whether the patient's uncle, who was hairless and edentulous too, perspired or not.

A similar history was given by the mother of Gibb's two boys. The father was normal and none of his relatives had any dental anomalies; the mother, however, was the second of eight children, and although her parents were normal, two brothers had very pointed teeth, and several of her sister's children (sex not stated) shewed abnormal dentition. Christ's patient had a younger brother and a first cousin on his mother's side who were similarly affected. On the other hand no pertinent family histories could be obtained by Strandberg, MacKee and Andrews, Goeckermann, Weech (first case) or myself.

One of the most striking facts which emerges from a study of these data is that, excepting the patients recorded by Williams and Guilford, all have been males. Moreover, there is "a report by Thadani²² of a type of man

occurring in the Hindu Amil community of Hyderabad Sind, a town in India, who is 'characterized by an absence of teeth, a bald head and by extreme sensitiveness to heat.' Although meagre, this description is probably sufficient to identify them as persons afflicted with the anhidrotic form of ectodermal dysplasia. Thadani's publication was in the nature of a preliminary report. Actual pedigrees were not given, but the author stated that the inheritance has been strictly sex-linked'' (Weech). The chart of the Kitzing family shews especially clearly that the anomaly was here also transmitted by the female and inherited by the male.

It is interesting to contrast with these facts Jacobsen's conclusions on the inheritance of dystrophy of the hair and nails of which he traced sixty-four members in five generations of one family. From his study he decided that the transmission was Mendelian and not sex-linked.

It is impossible with the scanty material available to make generalizations and it is especially difficult to explain the occurrence of what would appear to be a sex-linked character in the female. Weech believes that this condition must have its origin in a genetic mutation, and that this mutation may in the beginning occur in either sex, but thereafter the transmission would be sex-linked and the anomalies would appear only in the male. In support of this hypothesis he points out that in neither of the female patients described in the literature is there a record of antecedents affected, and that the history in Guilford's case, of an affected woman transmitting the characteristics through a non-affected daughter to an affected son, may be explained in the same way.

DIAGNOSIS.

In discussing the symptomatology it has been mentioned that these patients bear many superficial resemblances to the subjects of congenital syphilis. The absence of visceral involvement, eye lesions, bone or joint affections, the consistently negative Wassermann reaction and the non-luetic family history, should be sufficient to prevent the erroneous diagnosis of this disease.

Another condition which may suggest itself is progeria. Weech mentions that two competent observers made this diagnosis of his patient and the same suggestion was made with regard to my patient¹⁹, but there are several features which distinguish it from that extraordinary condition described by Hastings Gilford⁸, indeed the similarity is entirely based on the aged appearance produced by the sparse white hair and the under-development of the edentulous jaws. There are no signs of arteriosclerosis or renal disease, and although the skin is dry it has not the wrinkled appearance of advanced senility found in progeria. The prognosis in the two conditions is very different, for whereas the subject of progeria usually dies about puberty, those with hereditary ectodermal dysplasia often lead active lives to a good age.

Yet another condition from which this must be differentiated is xeroderma. These patients certainly do suffer from a xeroderma using the word in its etymological sense, but it differs from the more limited conception of the dermatologist in that it shews a sex-linked inheritance, an absence of sweat

glands and an association with dental and other ectodermal anomalies. Renal and other forms of infantilism may suggest themselves but a careful general examination should render their exclusion simple, for although these patients are slightly built and often considerably under the normal weight owing to the paucity of subcutaneous fat, there is no delay in the onset of puberty, and sexual function is normal, nor is there any evidence of renal disease.

SUMMARY.

A case of hereditary ectodermal dysplasia is described and a review of the literature, in which fifteen other cases have been recorded, has been made.

Numerous instances of various forms of ectodermal dysplasia have been hitherto reported but the cases described in this communication form a clearly defined class which is best styled 'the anhidrotic form of hereditary ectodermal dysplasia.' Reasons for preferring this title, which was suggested by Weech, to the older one of 'congenital ectodermal defect' are given.

The symptomatology is described and the diagnosis from congenital syphilis, progeria, xeroderma, renal and other forms of infantilism discussed. Syphilis is not an aetiological factor and although endocrine dyscrasia may play some part in creating these, the most striking feature is the inheritance which appears to be sex-linked.

I am greatly indebted to Dr. C. K. J. Hamilton, Dr. Montague Maizels and Mr. W. D. Southern for their help and advice in the preparation of this paper.

REFERENCES.

1. Ascher, B., *Verhandlung d. Berl. Gesellsch. f. Anthrop.*, Berlin, 1898, 114.
2. Barrett, A. M., *Arch. Neur. & Psychiat.*, Chicago, 1919, II, 628.
3. Christ, J., *Arch. f. Dermat. u. Syph.*, Berlin, 1913, CXVI, 685.
4. Cowper, W., quoted by Darwin, C., *v. infra.*, 321.
5. Darwin, C., *Variations of Animals & Plants*, Lond., 1868, II, 319 & 321.
6. Eisenstadt, J. S., *J. Am. Med. Ass.*, Chicago, 1913, LX, 27.
7. Gibbs, J. H., *Dental Rec.*, Lond., 1915, XXXV, 669.
8. Gilford, H., *Disorders of Postnatal Growth and Development*, Lond., 1911.
9. Goeckermann, W. H., *Arch. Dermat. & Syph.*, Chicago, 1920, I, 396.
10. Guilford, S. H., *Dent. Cosmos*, Philad., 1883, XXV, 113.
11. Hopson, M., *Proc. R. Soc. Med.*, Lond., 1920, XIV, (Odont. Sect.), 23.
12. Hutchinson, J., *Med. Chir. Trans.*, Lond., 1886, LXIX, 473.
13. Jacobsen, A. W., *J. Am. Med. Ass.*, Chicago, 1928, XC, 686.
14. Keith, A., *Antiquity of Man*, Lond., 1925, II, 675.
15. Kjaer, T., *Dent. Cosmos*, Philad., 1907, XLIX, 924.
16. Loewy, A. & Wechselmann, W., *Virchow's Arch. Path. Anat.*, Berlin, 1911, CCVI, 79.
17. Mackee, G. M. & Andrews, G. C., *Arch. Dermat. & Syph.*, Chicago, 1924, X, 673.
18. Rushton, W., *Trans. Odont. Soc.*, Lond., 1904, XXXVI, 199.
19. Smith, J., *Proc. R. Soc. Med.*, Lond., 1929, XXII, (Sec. Dis. Child.), 54.
20. Strandberg, J., *Nord. Med. Ark.*, Stockholm, 1918, LI, 1.
21. Tendlaw, B., *Virchow's Arch. Path. Anat.*, Berlin, 1902, CLXVII, 465.
22. Thadani, K. I., *J. Hered.*, Washington, 1921, XII, 87.
23. Thurnam, J., *Med. Chir. Trans.*, Lond., 1848, XXXI, 71.
24. Vichot, J., *Des anomalies dentaires*, Lyon, 1911, 85.
25. Weech, A. A., *Am. J. Dis. Child.*, Chicago, 1929, XXXVII, 766.
26. Wheelon, H., *Endocrinology*, Los Angeles, 1925, IX, 35.
27. Wieting, ., *Deutsche Med. Wchnschr.*, Leipsic, 1911, XXXVII, 504.
28. Williams, C. J. B., *Lond. Med. Gaz.*, Lond., 1848, XLI, 336.