ON

SPORADIC CRETINISM, OCCURRING IN ENGLAND.

BY

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Some years ago Dr. Hugh Norris, of South Petherton in Somersetshire, showed that in a neighbouring village, Chiselborough, there prevailed endemically a disease analogous to the cretinism of Alpine countries, and, like it, frequently associated with goitre and deaf-mutism.[1] From a communication with which I have been favoured by Dr. Norris it appears that "the cretins in Chiselborough have now almost died out. Improved sanitary measures, better food, better education, and greater contact with the outer world, together with fewer intermarriages," appear to have combined in eradicating the disease. Scattered examples of it, however, are still occasionally to be met with in the part

[1] "Notice of a remarkable disease, analogous to Cretinism, existing in a small village in the West of England." 'The Medical Times,' 1847, xvii, p. 257.

of Somersetshire in which Dr. Norris resides. In his letter to me he speaks of having recently had an opportunity of examining a "boy or man of a thoroughly cretinous type, the son of a very intelligent but very goitrous mother, and of a father (a respectable tradesman) of a type tending to cretinism." He is also "cognizant of another case some eight miles from Chiselborough where two children were cretins, the mother being well formed and intelligent but goitrous, the father having made at least more than one step towards the cretin type."

I am not aware that in any other part of Great Britain a similar form of degeneration has been observed within a recent period to prevail endemically. Many years ago Dr. Reid[1] stated that cretinism associated with goitre existed in the Isle of Arran. I have not been able to learn that its presence in that island has since been noted.

Some years ago a boy, æt. 14, was admitted into the York County Hospital who had a large bronchocele, and was of stunted growth and deficient in intelligence.[2] His parents had resided all their lives in York; none of his relations were known to have been idiots, nor to have suffered from goitre.

Now it would seem that this case fairly deserved to be termed one of sporadic cretinism; and as certain foreign writers on the subject have admitted the existence of such a form of the disease, it might appear that there was nothing unusual about the case.

In reality, however, this case is the only one on record (so far as I can ascertain) in which cretinism has occurred sporadically, in association with goitre. The writers to whom I have just referred have contented themselves with a simple mention of sporadic cretinism, and have nowhere given accounts of any cases of the kind.

Thus, I have been in doubt whether these writers have

[1] 'Edinburgh Medical and Surgical Journal,' xlvi, p. 47.

[2] 'Medical Times and Gazette,' 1855, ii, p. 266. An account of this case is also given in the 'Manual of Psychological Medicine,' by Drs. Bucknill and Tuke, 2nd edit., p. 100, 1862.

not rather had in their minds another and apparently a less rare affection, which in some important respects differs from ordinary cretinism, but which has received that name from at least one English observer.

In this affection no enlargement of the thyroid body occurs; and it presents other peculiarities, to which I desire to draw the attention of the Fellows of the Royal Medical and Chirurgical Society.

I also believe that I can advance an hypothesis by which the discrepancies between ordinary cretinism and this peculiar form of the disease can be accounted for. If my hypothesis be accepted, the name of sporadic cretinism will be very applicable to the latter affection. But if this be regarded as essentially of a different nature from endemic cretinism, it ought rather to receive a distinct appellation.

Provisionally, however, I will use the term "sporadic cretinism" for the disease which I am about to describe: and I will now relate as briefly as possible certain cases of it which have recently been under my observation.

CASE 1.—*Idiocy with stunting of body and cretinous type of face (? congenital); no goitre; soft movable tumour on each side of neck outside sterno-mastoid muscle.* (See Plate II, fig. 1.)

(For the notes of this case I am indebted to Mr. ERNEST EVANS and Mr. C. J. OLDHAM, successively House Surgeons to the Evelina Hospital for Sick Children.)

Edward D—, æt. 8, was admitted into the Evelina Hospital for Sick Children under the care of Dr. Fagge, Nov. 26th, 1870, having previously been an outpatient of Dr. E. B. Baxter's.

The boy's father and mother are healthy; they live in a roomy house at Rotherhithe; they are sober and regular in their habits; the most careful inquiry fails to elicit a history of habitual or even occasional intemperance in either parent. Their other children are healthy.

At birth the child was large, and forceps were employed in his mother's delivery.

He began to cut his teeth at two years, and first attempted to walk when three years and a half old. During early infancy it was not noticed that anything was wrong with the child; but after a time his mother observed that he would sit down whenever he could, and often remained silent in one position for hours together. He also ceased to grow; his mother does not think that he has grown since he was two years and a half old.

At the present time, although eight years of age, he seems more like a child two or three years old. He is, however, particularly quiet, sitting still in whatever place he may be put, and rarely moving of his own accord. An air of torpid contentment generally characterises him. Sometimes his face will light up with a slow smile. Now that he has become accustomed to the hospital ward he always smiles when notice is taken of him, and he will at any time smile stupidly when told to do so. He says very little, and appears not to know many words; but he will name correctly things shown to him. He is said to be passionate, but such explosions of temper occur but rarely; and he is a great favourite with the nurses and attendants. He is clean in his habits.

The boy's height is now 2 feet $7\frac{3}{4}$ inches; he weighs 25 lbs.

His appearance will probably be indicated by no description so well as by the plate taken from a photograph which accompanies this paper (see Plate II, fig. 1). The head is large and round; the face is broad; the eyes are wide apart, being separated by the broad flat root of the nose; the tip of the nose is flattened and upturned, and the openings of the nostrils are rounded; the mouth is large and generally open, but there is little or no flow of saliva; the lips are thick; he has all his first set of teeth; the tongue is of natural size.

On either side of the neck, just above the clavicle, is a soft, movable, inelastic swelling. This can be drawn downwards across the clavicle to some extent, and appears to be connected with the subcutaneous rather than with the deeper tissues.

No prolongation of it can be discovered in the axilla. It has been thought that kneading causes it to disappear for a time. On application of the stethoscope a very distinct respiratory murmur is heard over each swelling.

There is no goitre, and no indication of the presence of a thyroid body can be felt in front of the trachea.

The chest is well formed; the limbs are short and thick; the tibiæ are somewhat curved, but there is no rickety enlargement of the epiphyses; the hands and fingers are very broad, short, and thick, as are also the feet and toes.

The skin all over the body, but not that of the face, is harsh, and presents scattered, small, hard scales of a light brownish-grey colour. The hair and eyelashes are long, dark, and abundant.

CASE 2.—*Stunting of body, with change of features a cretinous type, dating only from an attack of measles (?) at eight years of age; no goitre; soft tumours in both supraclavicular fossæ; mental faculties unimpaired; age of patient* 16¾ *years* (see Plate II, fig. 2).

(Notes given to Mr. J. LACEY MORLEY by patient and her elder sister.)

Kate —, æt. 16¾, came as an out-patient to Guy's Hospital under the care of Dr. Fagge, and was subsequently admitted into Mary ward under Dr. Wilks.

Her parents are in good circumstances, her father being station-master at an important railway station about twenty miles from London. She has three brothers and one sister, all of whom are well grown.

Until she was eight years old she was a good-looking child, with a large quantity of black hair. She was lively and good tempered, and played like other children. She went to school at four years of age, and made satisfactory progress.

When six years old she had "a slight attack of measles," but did not keep her bed. About the same time she had hooping cough. Two years afterwards she had "a second

attack of measles." She then kept her bed for two weeks; she was not insensible. She is further stated to have had erysipelas at that time. Sores followed on the head, and she lost her hair in patches. She suffered from severe diarrhœa and also from shortness of breath.

Whatever may have been the precise nature of this illness, it appears beyond doubt to have been the starting point of a remarkable alteration in her physical development. Her relations and friends concur in stating that she has not grown in height since that time, and that her features have undergone a complete change. Two or three weeks afterwards the hair grew dry and crisp; and whereas it was black, it acquired in places a golden colour. From that time it has remained extremely scanty and short.

Her present appearance is perhaps indicated by the accompanying photographs better than by any description. (See Plate II, fig. 2.) She measures four feet one inch in height.

The head is round and well formed, appearing small rather than large in proportion to the size of the body; the forehead is not projecting; the eyes are small, the eyelashes short; the pupils dilated and sluggish; the root of the nose is much flattened; the tip of the nose is wide and upturned; the alæ are thick; the mouth is large; the lips are thick; the cheeks are plump and firm; the complexion is pale and unhealthy looking; the ears are small, even for her size, but very well shapen.

The neck is rather short, but not thick. On each side, outside the sterno-mastoid muscle, is a doughy soft swelling. These swellings are movable to a considerable extent, they can be drawn down over the clavicles, and seem, as it were, to disappear when kneaded. They are not very definitely circumscribed, and yet they seem to be lobulated. On auscultation a respiratory murmur can be heard over them. It has, therefore, been supposed that they contain the apices of the lungs; but it is by no means certain that the respiratory murmur is more distinct than it normally is in the same regions.

These swellings were first noticed four years ago: first that on the right side, and two or three weeks later the left one.

They are about the size of hen's eggs, the right one rather the larger. They overlie the sternal half of the clavicle on both sides.

There is no goitre, nor can any part of the thyroid gland be felt in front of the trachea.

When she first came to Dr. Fagge it was on account of the extremely scurfy state of the head, amounting, indeed, to a condition of pityriasis. The hair was then sparse, of a light colour, dry and short. The appearance of the scalp was, in fact, very like that which is sometimes left by favus after its cure, quite unlike that left permanently by any ordinary eczematous or scaly eruption. Under appropriate treatment, however, the hair has become quite thick and of fair length.

There was also a scaly eruption scattered over the shoulders and back, resembling an eczema squamosum rather than a psoriasis. This also has been greatly improved by local treatment.

The limbs are small, but perfectly well formed. The bones are straight, and their extremities are of natural size. They present no indication of a past rachitic state. The muscles of the limbs and of the body generally are well-developed and very firm. The hands and feet are of about the size of those of a child six or seven years old; kid gloves of No. 4 size are rather too large for her hands.

Her mental faculties are very good. She appears to be very intelligent; she is not nervous or shy, and converses freely. She is, however, said to be of a reserved character; she is very fond of reading all kinds of books, but has a weakness for novels. She does not care for music.

Her voice is like that of a child, but is squeaky, and rather disagreeable.

The catamenia first appeared when she was fifteen years old, and have been regular ever since. The mammæ appear to be as much developed as is usual in girls of her age.

She does not enjoy good general health; she is subject to colds. Her breath is often offensive. Her extremities are apt to be cold, and she is fond of sitting over the fire with a book.

The heart-sounds are normal; the pulse is 73, small and feeble.

CASE 3.—A. B—, æt. 20 years (see Plate II, fig. 3), has been in an idiot asylum ten years. His height, when he was admitted, was 2 feet 4 inches; it is now 2 feet $7\frac{3}{4}$ inches. His weight with his clothes on was 1st. $9\frac{3}{4}$lbs.; it is now 2st. 5lbs.

He is affectionate, placid, and quiet. He sleeps well. He has not the power of speech.

His senses are natural. He can walk only by clinging to the furniture.

His countenance is pallid, and his features are cretin-like in an extreme degree. There is no goitre, and no indication of the thyroid body can be felt in front of the trachea. The swellings above the clavicles are present, but are not of any great size. He is constantly dirty and wet, and makes no signs as to his wants. The skin of his hands and legs looks as if too large for him.

A sister is said to be affected in the same way as he is.

CASE 4.—C. D—, æt. 12, recently admitted into an idiot asylum (see Plate II, fig. 4).

Her mother died of hepatitis; her father is healthy. They were not connected by consanguinity. No other child is affected.

The girl's condition is congenital, and is attributed to the mother having been frightened, when pregnant, by the sudden death of a neighbour.

Her height is 3 feet $10\frac{1}{2}$ inches; her weight is 4st. 3lbs. Her complexion is sallow; her features are cretinous. The tumours above the clavicles are well marked; the width of the forehead is 4·1 inches; the hands are short and broad. The skin is harsh and dry.

She is deaf and dumb, but is very affectionate, cheerful, and happy. She can say *a*, *b*; she can drill, and she helps to dress the other children; she is afraid of dogs; she is not mischievous, nor noisy, nor spiteful; she has a good memory;

she sleeps well; she has had no epileptic fits; she uses a knife and fork a little at her meals.

In proof that the peculiar type of conformation exhibited in these four cases is not very common, I may state that the Asylum at Earlswood contained only two examples of it when I recently had an opportunity of visiting that institution. The cases in question were kindly shown to me by Dr. Grabham, and form two of the four cases on which this paper is based. Each of them presented the peculiar cretinous configuration in a marked degree. It may be added that, according to the officials and nurses, of whom I made particular inquiry in reference to this point, no other children with similar features had recently been inmates of the asylum. No conditions intermediate between that of the two patients above described and ordinary idiocy were to be found. So far as I could learn, common idiots never present the peculiar supra-clavicular swellings, the existence of which appears to be a constant feature in sporadic cretinism.

Another case, very similar in all its characters to those above related, was exhibited to the Pathological Society, in the year 1869, by Dr. Langdon Down.[1] The subject of it was a female child, æt. 5, who measured only 22 inches in height, could only stand with the help of a chair, and gave utterance merely to a few monosyllabic sounds. The hair was sparse and coarse; the tongue was large; and there was a "venous tumour" on each side of the neck above the clavicle.

Dr. Down brought forward this case as an example of a group of cases in which arrested development had been due (as he believed) to intoxication of one or both of the progenitors at the time of the procreative act. In each instance the "venous tumours" in the neck had been observed.

Two very remarkable cases of sporadic cretinism were described by Mr. Curling in a paper read before this Society

[1] 'Pathological Transactions,' xx, p. 419.

in the year 1850.[1] They are especially important, as being the only cases in which an opportunity has as yet been afforded of studying the anatomy of this morbid state.

Mr. Curling's first case was that of a child, æt. 10, a native of Lancashire, who was an inmate of the Idiot Asylum at Highgate, and was regarded by Dr. Little as a cretin. She measured 2 feet 6 inches in height. Her expression was idiotic; the mouth large, and the tongue thick and protuberant. She could not talk, and could only manage to walk from chair to chair with assistance. At the outer sides of the neck, external to the sterno-cleído-mastoid muscles, were two doughy inelastic swellings; similar swellings were also observed in front of the axillæ. She died of erysipelas, and Mr. Curling found that the peculiar swellings were composed simply of fat, which dipped down behind the clavicles and filled the axillæ. The fat was not encapsulated. There was not the slightest trace of a thyroid body.

Mr. Curling's second case was that of an infant, æt. 6 months, which was sent to him for examination on account of the existence of similar tumours. The infant had a marked idiotic expression; the tongue was large and protruding from the mouth. The child died, and the swellings were found to consist of superficial collections of fat tissue, without any investing envelope. No trace of the thyroid gland could be discovered.

The reports of these seven cases appear to show that the subjects of them presented a remarkable uniformity, both in their physical configuration and (in general) in their mental condition. The characteristic features of the disease may be summed up as follows:—

I. The body is stunted, the height scarcely exceeding four, three, or even two feet, in different cases. The head is round; the face is broad; the eyes are widely separated by the flat root of the nose; the alæ nasi are thick; the nostrils are

[1] "Two Cases of Absence of the Thyroid Body, and symmetrical swellings of fat tissue at the sides of the neck, connected with defective cerebral development." 'Med.-Chir. Trans.,' xxxiii, p. 303.

rounded; the mouth is very large and generally widely open; the lips are thick; the hands and feet, as well as the fingers and toes, are short and broad.

II. When "sporadic cretinism" is congenital it is also attended with deficiency in the mental powers, varying in degree, but of a character very like that which belongs to the "endemic" form of the disease. The child is free from the mischievous tendencies displayed by so many idiots. It is good humoured, but torpid, often sitting for a long time quiet in one place. Sometimes it can walk only with the assistance of a chair. It is not rarely deaf and dumb.

III. Sporadic cretinism, instead of being associated (like endemic cretinism) with goitre, appears to be attended with a wasting or absence of the thyroid body. The discovery of this fact by Mr. Curling has already been mentioned, and I am able to confirm it to this extent, that I can feel no trace of the thyroid in the neck of any of the four patients whom I have had an opportunity of examining.

On the other hand, sporadic cretinism seems to be invariably accompanied by the presence of symmetrical fatty tumours, one of which lies beneath the skin of the neck on each side, just external to the sterno-mastoid muscle.

IV. Sporadic cretinism is not necessarily congenital. It may arise as late as the eighth year in a subject previously healthy and well developed.

V. It is not related either to rickets or scrofula. It is not inherited syphilis.

VI. Not only is this form of cretinism sporadic, but it does not arise by the intensification of a morbid influence, of which earlier manifestations can be traced in the parents of those affected by it. It springs up, generally without apparent cause, in the offspring of a healthy father and mother.

VII. It has been supposed in certain cases to have resulted from one or both of the parents having been intoxicated at the time of procreation. But it does not seem that this explanation holds good for all cases, even for all those which are congenital. Nor does it appear that it can be attributed

to the employment of instruments in the delivery of the mother, a view which was at one time urged in reference to the endemic form of the disease.

The subject of sporadic cretinism appears hitherto to have attracted very little attention. I am not aware that anything has been written concerning it, with the exception of the papers which have already been quoted. In Virchow's great work on tumours I have failed to find any reference to it, either in the chapter on goitre (in which "endemic cretinism" is discussed at some length) or in that on fatty tumours.

It is, therefore, especially incumbent on me to state that, in the course of his clinical teaching at Guy's Hospital, Dr. Gull some years ago made me acquainted with many of the principal features exhibited by these cases. So far as I remember, the characters on which he laid most stress were the broad face, the flat nose and thick lips, the broad hands and feet, and the mild, tranquil disposition, so different from the mischievous tendencies of the idiots with whom these children are so generally associated. I do not think that Dr. Gull's attention had at that time been drawn to the presence of the peculiar tumours above the clavicles. He called the disease *cretinism*.

In the majority of cases *sporadic* cretinism, like *endemic* cretinism, is congenital. But Case 2 appears, beyond doubt, to be an example of the development of the same physical state as late as the eighth year in a child previously healthy. I believe that no instance of a similar kind has hitherto been recorded. It will be observed that in this case the peculiar physical configuration was alone manifested, or, at any rate, that any change in the mental powers was doubtful. It may, therefore, be interesting to speculate as to what characters would be present, should the disease (if that be possible) arise still later in the course of adult life. The peculiarities in the form of the cranial and facial bones, and in the bony framework generally, would then probably be absent, the development of the skeleton being unalterable when once completed. And I think we must conclude that

the most marked features in such a case would be a coarseness and thickness of the soft parts of the face (especially the lips), and perhaps of the subcutaneous tissues of the hands and feet, besides the presence of the supra-clavicular fatty tumours, and possibly a wasting of the thyroid body, if that should prove to be a constant character of the disease. I have heard that symmetrical fatty growths are not very uncommonly developed in adults in the supraclavicular regions, but I am not aware that their presence has hitherto been observed to be associated with an impairment of the general health.

The fact that in Case 2 the cretinous conformation of body began to manifest itself only when infancy had been passed is of interest from another point of view. It affords a broad ground of distinction between sporadic and endemic cretinism. For, so far as I can ascertain, such an occurrence has never been observed in those countries where cretinism is endemic. It is true that various opinions have been expressed as to whether it can be determined at the time of birth, or even within the first two years, that a child will become a cretin. But with advancing development the cretinous configuration appears always to manifest itself. In the Report of the Sardinian Commission[1] it is stated that "according to information received from medical men practising in infected districts and according to all those who have written on this degeneration, there is no example in which, after the seventh year, a healthy child has become a cretin." And the Commission further quote with approval the statement of Maffei (who practised for a long time where cretinism was endemic, and who, therefore, had good opportunities of observing), "that the period within which cretinism may commence is limited by the fourth year of life."

It must, indeed, be mentioned that Rösch[2] has recorded two cases in which the disease is said to have begun

[1] 'Rapport de la Commission créée par S. M. le Roi de Sardaigne pour étudier le Crétinisme,' Turin, 1848, p. 11.

[2] 'Untersuchungen über den Kretinismus in Würtemberg,' Erlangen, 1844, pp. 179, 183.

respectively at five years of age and between seventeen and eighteen years; but this writer avowedly includes under the name of cretinism a variety of forms of mental disturbance, occurring in subjects of widely different habits of body. Indeed, he expressly asserts that all idiots are cretins. The first of the cases above referred to was one of ordinary dementia, starting from convulsions, as is so often the case during childhood. Living where goitre was endemic, the patient had goitre; but there is no reason to suppose that this was more than a coincidence. The other case seems simply to have been one of inherited insanity.

Should further researches show that an atrophy of the thyroid body is a constant feature in sporadic cretinism, it may be interesting to speculate whether this can possibly be the cause of the other changes which make up that morbid state. Such appears to have been the conclusion to which Mr. Curling inclined when he published his observations on the subject. And this view acquires great interest from the fact of the association of endemic cretinism with the opposite condition of goitre.

The discrepancy is, indeed, so striking that I have sought for an hypothesis by which (as I think) it may be explained.

In the first place it must be borne in mind that the relation between goitre and endemic cretinism is by no means a very simple one. Goitre prevails endemically in many parts of England where endemic cretinism is unknown. Goitre is the earlier effect of the endemic influence; cretinism shows itself when the action of that influence is intensified by operating on more than one generation. From these facts it would be expected that the individuals most severely affected with cretinism would invariably have very large goitres; but observation shows that the exact contrary is the case. In the worst cretins the thyroid body is often no larger than natural; while persons with enormous tumours are frequently well developed, both physically and mentally, although they and their ancestors have long resided in the same neighbourhood, exposed to the morbific agency.

These considerations have led some observers to doubt

whether cretinism and goitre depend upon a common cause, and even to suppose that their association is a mere accident.

It appears to me, however, that the correct inference from the facts above stated is rather that a certain antagonism exists between goitre and cretinism. I have thought that when a large goitre exists, it may possibly have the power of protecting against the more severe effects of the endemic influence; and thus that cretinism is associated with an enormous bronchocele only when the exciting cause is present in a very intense degree.

It is well known that the most careful investigation has failed to show, either in the air, in the water, or in the soil of Alpine valleys where cretinism is endemic, the constant presence of any element, which is uniformly absent where the disease does not prevail. There is, therefore, nothing inconsistent with the facts in the supposition that the cause of cretinism may be much more widely diffused than the disease itself, although, doubtless, with an intensity varying in different localities. It is only needful that some counteracting tendency should be discovered to account for the limitation of the disease.

It is at this point, as I think, that the occurrence of sporadic cretinism, in association with an absence of the thyroid body, may be brought to bear upon the theory of the subject. We have but to suppose that the healthy thyroid body is capable of exerting such a counteracting influence, and that in most parts of England the cause of cretinism acts only with a low degree of power; and we can then at once see why a form of cretinism should show itself when the thyroid body is atrophied.

I have already suggested that in those regions where cretinism prevails endemically, the bronchocele, which may be regarded as an hypertrophied thyroid body, exerts a similar action in protecting against the more powerful operation of the same cause; and this view appears to me to afford a satisfactory explanation of those relations between goitre and endemic cretinism which have hitherto appeared so difficult of comprehension.

DESCRIPTION OF PLATE II.

Cases of Sporadic Cretinism occurring in England.

FIG. 1. Case 1.—E. D—, idiot, with stunting of body and cretinous type of face. Æt. 8 years, 2 feet 7¾ inches in height (see page 157).

FIG. 2.—Case 2.—Kate T—, stunting of body, with change of feature to a cretinous type, from attack of measles at 8 years of age. Æt. 16¾ years, 4 feet 1 inch in height (see page 159).

FIG. 3. Case 3.—A. B—, idiot, with peculiar cretinous conformation. Æt. 20 years, 2 feet 4 inches in height (see page 162).

FIG. 4. Case 4.—C. D—, idiot, deaf and dumb, features cretinous. Æt. 12 years, 3 feet 10½ inches in height (see page 162).

Fig 1.

Fig 2.

Fig 4.

Fig 3.