SPORADIC CRETINISM

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by

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SPORADIC CRETINISM

Synonyms:

As usual a great many names have been applied to this disease, most of them giving prominence to certain symptoms or set of symptoms, to the exclusion of others less marked, but equally characteristic of the condition.

The following are examples of this class:--

Cretinoid Idiocy

Idiotie avec cachexia Pachydermique or Idiocy with pachydermatous cachexia.

Pachydermie Cretonoide, or Cretonoid Pachydermia.

The name commonly adopted by English writers and which has been used as the title of this essay, Cretinism, makes no special reference to any particular symptoms,but has general reference to the whole set of symptoms which are met with in this disease, and on this account the most simple appellation is to be preferred.

Etymology and History:

Among the great number of etymologies which have/

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have been proposed, the Sardinian commission, whose report was published in 1849, instituted by the late Charles Albert, King of Sardinia, thinks those suggested by Fodere (Turin) and Iphofen as among the most probable, viz. Chretien, from the cretin being incapable of faults and therefore a christian par excellence; or the cretira of the Grison dialect which signifies a poor or stupid creature.

? Crete & Cretons [illegible]

In the different localities wherein they abound a great variety of appellations is bestowed upon them, and as many of these imply contempt (cagot, garaches, etc.) Dr Milligan draws an argument thence in favour of the view popularly entertained that they are the remnants of persecuted races of people holding very much the position of the Indian pariah.

Although describing the disease sporadic cretinism, I find it necessary for the purpose of this essay to draw attention to the difficulty of differentiating between the sporadic and endemic forms of the disease. As will be seen, the disease itself, the history, and the terms applied to it are really descriptions of endemic cretinism and were described years before sporadic cases were discovered.

Although the term sporadic disease means a disease/

disease usually epidemic, attacking only a few in a district and not spreading, there are marked differences between the sporadic and endemic form and one which may for illustration be mentioned, is the entire absence of the thyroid gland in the sporadic, whereas in the endemic the same result on the general condition of the body is attained but the thyroid gland itself is generally very much enlarged, but disorganised and consequently not able to perform its function (Quain's Dictionary of Medicine Vol. I. 1894).

Geological conditions peculiar to special localities such as the valleys of Switzerland, and in this country the dales between Lancashire and Yorkshire also play a most important part in the causation of endemic cretinism, whereas no proof exists of the causation of the sporadic type of the disease by these conditions.

Other differences may exist but the above are sufficient to show the difficulty of differentiation.

History: The first regular mention of cretins occurs in the sixteenth century when on ecclesiastical registration of births and deaths being instituted in Aosta, they were mentioned as innocents or sanctified persons (beats).

Two Swiss physicians, Plater (1500) and Siniler (1574) first described cretinism as it prevailed in the Alps.

Fallopio/

Fallopio (1563) in Venice and Piridus (1612) in Paris described an analagous condition. Although various travellers, as Coxe and Sassure contributed important particulars concerning them, the first complete monograph on the subject was published by Fodère at Turin in 1792 and again at Paris in 1802.

This work excited great attention and since its appearance a large number of treatises has been contributed on the subject.

More recently the disease has been discussed at the meetings of the Swiss and German naturalists, and ~~have~~ commissions have been appointed by these for its investigation. Up till 1802 the contributions relating to cretinism were Continental, and although Dr Reeve of Norwich visited the cretin districts of the Valais in 1805 and published the results of his observations in a short, but excellent paper in the fifth volume of the Edinburgh Medical and Surgical Journal, his views do not appear to have been utilised in connection with the study of the disease. The study of cretinism seems to have been established in England from this time.

Wood, about the year 1810 in "Some Remarks on endemic idiocy and goitre" states that he had seen 19 cretins and a greater number of imbeciles in Derbyshire/

Derbyshire. Read in 1836 in his "History of the cause of Bronchocele" says that Chiselborough in Somerset contained 24 well-defined cretins in its population of 360. In pursuing the history of this disease our attention must again be turned to the Continent.

Until the year 1839 the condition of the cretin was regarded as irremediable; whole villages of them lived and died in Switzerland without the most sanguine physicians of the country ever dreaming that the condition was otherwise than hopeless.

In that year, however, a young physician of Zurich - Dr Guggenbühl - witnessed an incident which convinced him that even a cretin could be taught something; he saw one of these creatures muttering, without understanding it, a prayer beneath a crucifix: the prayer had been taught by the mother, and the words though not the sense had been remembered by the idiot. Convinced that with so much memory the mind could not be utterly extinct Dr Guggenbühl resolved to devote his life to the single purpose of restoring as far as possible the gift of intelligence to these unfortunate beings.

The result of this was the formation in 1841 of the first sanitary establishment (Psychico-physical hospital) on the Abenberg near Interlachen in Switzerland and which was so graphically described/

ed about this time by Dr Forbes in his "Physician's Holiday" (pp.109-111) for cretinism in Europe.

In 1849 the results of the Sardinian commission were published and in the following year Mr Curling described the rare disease in England.

In 1871 Dr Hilton Fagge read a very interesting paper on the subject before the Medico-Chirurgical Society of London. Since the recognition of cretinism as a distinct disease many physicians have been interested in the relationship between it and myxoedema; the chief names associated with this question at first were Dr Hilton Fagge; Mr Curling (both already mentioned); Dr Ord; Sir William Gull; Professor Kocher; Dr Byrom Bramwell, etc.

At the present time though cretinism can be regarded as a comparatively rare disease, reports of cases are of frequent occurrence in our medical journals.

Etiology:

Before discussing the essential cause of sporadic cretinism we might first consider the influence of

Sex,
Age,
Alcoholism.
Phthisis and the allied scrofulous diseases.
Diseased conditions of the central nervous system, and under this head we might include fright or injury to the mother during pregnancy.
Consanguinity.

Sex: In England the disease affects more females than males. Appended is a list collected by different authorities with the proportion of males and females given in tabular form:--

	Males	Females	Total
Fletcher Beach:	43	71	114
Rushton Parker:	34	56	90
Byrom Bramwell:	14	30	44
Langdon Down:	3	9	12
Telford Smith:	1	3	4
	95	169	264

Taking the total of these as different cases it will be seen that there is a preponderance of females in the porportion of 7 to 4. This proportion is equally borne out in the larger lists while the smaller ones, which are necessarily less reliable for statistical purposes, still show the greater frequence of the occurrence of the disease in females. From my own brief experience all the cases seen, four in number, have been females.

Age: There is a great difficulty in determining at what age the symptoms of sporadic cretinism first become manifest.

In the majority of cases it may be assumed that symptoms show themselves during the first two years of extra-uterine life. Some authorities would/

would make the limit even earlier, thus Fletcher Beach quotes 59 cases in 40 of which symptoms appeared under the age of 18 months.

Dr Bourneville is responsible for the statement "that sporadic cretinism generally appears before the age of 18 months and an experienced eye can discover the symptoms from the first year, if not from the first month of life."

Cases are not infrequent in which symptoms are observed for the first time at a much later period of existence; but this may be due to the patients not having been under medical observation till a late stage of the disease, for instance to quote a case from my own experience - that of a female aged 21, when the disease was first recognised. The juvenile appearance of the patient, - she appeared to be only four or five. This goes to prove that the arrest of development which is characteristic of the complaint must have commenced at the age of 4 or 5, and probably much earlier.

On the other hand, at the annual meeting of the British Medical Association held at Carlisle in 1896 Mr Victor Horsley showed a photograph of a well-marked specimen of intra-uterine cretinism and it is quite possible that a fuller and more complete knowledge of the pathology of this disease may show that it is always of embryonic origin.

Alcoholism: This is one of the morbid processes which some would advance as having a strong bearing on the occurrence of this disease.

Foremost among those advocates of this theory Dr Langdon Down states conclusively that out of the twelve cases which came under his care, the majority were directly attributable to the intemperate habits of the parents. Some of the later ones, he says, could not have been procreated under any other circumstances. One must hesitate to accept an assertion of this kind; no doubt procreation constantly occurs while one, or even both parents are under the influence of intoxicants; and again, the common occurrence of a single cretin in a large family of which all the other members are free from the taint would seem to directly contravert this theory.

Phthisis: The advocates of phthisis as a predisposing cause, like those of alcoholism, rely solely upon statistics to support their arguments; thus they say that in 43 per cent of cases the parents suffer from phthisis and in other 8 per cent. the disease was manifest in more remote ancestors. Considering the frequency of the occurrence of phthisis in this country we cannot consider these figures of much value as a proof of the relation between phthisis in the parents and sporadic cretinism/

inism in their offspring.

Other proof seems similarly deficient.

Diseased Conditions of the Central Nervous System: With regard to such we are again driven to statistics, and we find that Dr Fletcher Beach from inquiries into over 100 cases, discovered that in no less than 21 per cent. there was evidence of functional disease of the nervous system occurring in the direct line of parentage, and in others, minor functional disturbances were present.

Dr Telford Smith also lays great stress upon the direct connection between the condition of mental depression and worry on the part of the mother during pregnancy.

Concerning the influence of fright or injury which the mother may have sustained while pregnant: Proof of this depends entirely on the statement of the mother and I would merely suggest that this evidence must not be taken too seriously, it being a habit of a great many women to associate any abnormality in her child with some occurrence of this nature; for instance, how commonly does a woman attribute the presence of a naevus on her child to some injury in a similar region which she herself received whilst "carrying the child".

Consanguinity/

Consanguinity: Excepting so far as this conduces to imperfect development of the system and more especially the nervous system, I have not been able to find any proof that this may be regarded as one of the predisposing causes of sporadic cretinism.

The Essential Cause or Causation:--

All pathologists are agreed in associating this condition with the Thyroid Gland. In the majority of cases in which an autopsy has been obtained the gland has been found to be congenitally absent, when present there has generally been some gross pathological change of the gland noted; thus out of 27 post mortem examinations, records of which have come within the notice of authorities, there was complete absence of the gland in 24, and in the remaining three, although the gland was present, a bronchocele was observed in each case. Thus in every case the gland was either absent or had undergone gross morbid changes.

Examination of patients during life goes to further confirm these conclusions: thus out of 86 cases reported by Fletcher Beach no gland could be felt in 73, in 11 a gland could be felt, and in 2 it was markedly enlarged. Some observers would ascribe/

ascribe the onset of symptoms to be due in some cases to absence of, or non-development of the sexual organs, for instance, M.Buillanger showed a case of sporadic cretinism before the Société Medico-Psychologique of Paris, which he described as a very remarkable one - "It is a girl", he said, "born at Melum, of healthy and well formed parents; the first dentition was completed at 3 years of age, and it was then that general development became arrested; the girl is now 27 and has the intelligence and tastes of a child of 4 or 5; she plays with a doll and has no sentiments of modesty."

After describing the characteristic symptoms of the case, he goes on to say that the body is fat, the limbs thick, short and sufficiently regular; the second dentition only commenced at 18 and has not yet terminated; the pubis is smooth; the mammary glands rudimentary; menstruation has not yet occurred, nor has there ever been any sexual sensation.

M. Buillanger then goes on to suggest that the condition of this case and of another which he had seen in Paris arose from inactivity of the generative organs.

Having regard to the fact that in all cases in which the gland has been absent or diseased we would naturally incline to the theory that the generative organs/

organs in these cases merely submit to the same arrest of development that affects the other organs and tissues of the body, that in fact we have to deal here with symptoms of the disease rather than a cause.

In support of the thyroid theory on the other hand we have the experience of Kocher, Victor Horsley, Byrom Bramwell, and others.

Professor Kocher at the twelfth congress of German surgeons held at Berlin in April 1883, described 18 cases of complete extirpation of the thyroid gland; in one a small accessory thyroid gland became hypertrophied and in another a recurrence of the goitre occurred and these two cases showed no symptoms of cretinism. In the whole of the remaining 16 cases there occurred changes, more marked in the oldest cases than in those more recently operated upon, pointing to their distinctly progressive character, which may be briefly summarised as follows:--

Trophic changes resulting in loss of tone and vigour, both bodily and mentally; swellings of face and body occurring first in the intra-ocular regions and eyelids, later affecting the face, hands, feet and trunk; dryness and harshness of the skin; Falling/

Falling hair and marked anaemic symptoms; and, in those cases in which the patients at the time of operation had not attained full growth, development became most markedly affected.

Professor Kocher concluded by pointing out the undeniable resemblance of these symptoms to those of cretinism.

Mr Victor Horsley in his Brown lectures reported in The Lancet of January 3rd, 1885, describes the results observed by him in monkeys after extirpation of the thyroid gland - a few days after the operation he noticed tremors followed by paresis and hebetude, oligaemia and leucocytosis; mucin appeared in the blood, in the subcutaneous tissues which became swollen, jelly-like, and sticky, and in the salivary glands which became enormously hypertrophied owing to the extreme production of mucin, the parotid even secreted this substance; puffiness of the face and eyelids, and subnormal temperature, ending in death in 5 or 6 weeks.

It will be seen that the effects which in man require years for their development appeared in a few weeks in monkeys. Nevertheless the similarity of these symptoms to those occurring in Kocher's patients is evident.

The occurrence of these symptoms after removal of/

of the thyroid gland and especially the arrest of development observed in the more juvenile of Kocher's patients seems sufficient to justify the assumption that the similar symptoms occurring in sporadic cretinism are due to absence of thyroid function.

Pathology and Morbid Anatomy:

As already stated in discussing the causation of this disease, the thyroid gland is absent in the vast majority of the cases, being represented merely by a few fatty granules. In other cases the gland has undergone cystic or fibro-cystic degeneration.

Dr Rushton Parker divides cretinism into three pathological varieties. The first, which he describes as embryological, is due to non-development or partial development of the thyroid body and analagous to any other body malformation from deficiency such as absence of, or perverted development of the uterus, ovaries, testicles, etc. The second variety, he states, is due to atrophy of the parenchyma of the thyroid gland occurring occasionally after some serious illness in childhood and analagous to the atrophy of the testicles, which is a common sequel to mumps. The third variety is due to goitrous degeneration of the thyroid body. He goes on to say that though the aetiology and pathology of the varieties are quite distinct, the symptoms appear to

be/

be identical, and to depend solely upon the degree to which the function of the thyroid body has been lost and the youthfulness of the patient in whom the loss of function occurs.

Coming to the morbid anatomy, we observe thickening of the cranial bones with diminished diploea. Virchow has described premature closure of the spheno-basilic suture.

The brain is diminutive and there is an excess of fluid contained in the ventricles and sub-arachnoid space.

Shortening of the long bones, except the clavicle, occurs, and there is frequently a curious cupping of the extremities, giving rise to an appearance of epiphyseal enlargement, such as is characteristic of rickets.

The subcutaneous tissue, more especially of the hands, feet and eyelids, is swollen and infiltrated with a solid oedema which does not pit on pressure. The supra-clavicular swellings which will presently be described when we come to consider the physical signs, are attributed by some to deposits of non-encapsulated fat, while others hold that they are collections of mucin. Dr Robinson of Sunderland informs me that in one case of his, though these swellings were well marked during life it was found at/

at the post mortem that they had completely disappeared. From this he infers that mucin and not fat was the nature of the deposit.

On the other hand, Byrom Bramwell arguing that they are fatty in nature says, "they may disappear before death in consequence of emaciation and marasmus, which not infrequently occur during the terminal stages of the case."

There are no visceral lesions. It would be interesting to note the nature of these supra-clavicular swellings in a case of sudden death where emaciation had not taken place.

Symptoms:

In the great majority of cases which have been recorded the first indications of the disease were noticed during the first three or four years of life. In many instances the symptoms were apparent before the end of the first year.

It is not unlikely as Bourneville has suggested, that in many cases in which the disease appears to be developed later, e.g. about the third or fourth year, a skilled observer might have detected some indications during the first year of life.

The clinical picture which typical and fully developed cases of the disease present is a very striking/

striking one.

The body is markedly stunted - heavy-looking and podgy. At 20 or 30 years of age the patient may only measure three feet in height or even less. The mental development is more or less and in many cases almost entirely arrested: the patient though in years an adult, is in respect of stature, sexual development, and mental condition, a mere child. The facial appearance is very characteristic and often extremely ugly. The expression is heavy and apathetic. The skin of the face is almost invariably of a dingy gray or earthy colour and in certain parts, e.g. the eyelids, wax-like and translucent. The face is moon-shaped, the eyes set wide apart; the forehead low and narrow: the cheeks fat, pendulous, and baggy (gowl-like);the nose flat and peg shaped; the eyelids swollen, chronic inflammation of the lids - ciliary blepharitis - is often present. The mouth is large and usually open: the lower lip and sometimes both lips thick, swollen and often everted. The tongue is usually very large, thick, and often is seen protruding between the teeth.

The head is usually large in proportion to the size of the body, dolico-cephalic in shape,(as shown in photo No.4) narrow in front, broad behind: the anterior fontanelle with rare exceptions remains unclosed/

unclosed even after the patient has attained to adult age.

The ears are usually large, pale and swollen-looking. The hair, which in most cases is fairly abundant, more especially on the back and sides of the head, is straight, dry and coarse, like horse-hair. In the cases observed by Bourneville the hair of the head was almost always of a reddish or chestnut colour. This peculiarity has not been observed in England or Australia, indeed in the remarkable series of cases reported by Dr E. C. Stirling, only one red-haired member of the family was affected, and this family contained eleven members, five of whom were affected with the disease. The hair of the members was of two distinct types as regards colour; in the one it was dark brown, in the other auburn inclined to red; four of the brown-haired members were affected and only one red-haired member. When contrasted with Dr Bourneville's experience, this point is worthy of note.

During infancy a scaly or eczematous eruption is very generally present on the scalp and this condition may persist throughout the whole life of the patient; but in many cases the scalp though dry, rough and scaly, is not distinctly eczematous in later life.

The teeth are usually irregular, worn down, or carious./

carious. In many cases some of the milk teeth which are usually very late in being cut, persist even after the age of manhood is reached.

The neck is usually short and thick. A depression can often be felt in the position of the thyroid gland, and in many cases it is obvious even in life from the ease with which the lower rings of the trachea can be felt, that the thyroid gland is wanting.

Elastic swellings, which in many cases attain to such a large size as to be actual deformities and which I have already alluded to under the head of pathology and morbid anatomy, are almost invariably present in the supra-clavicular regions and often in the axillae or elsewhere. These deposits are probably almost always present at some stage of the case. A subcutaneous thickening which is probably due to deposits of fat is in many cases to be seen and felt in the upper dorsal region between the scapulae.

A growth of fine hair is also often seen in this situation, a remarkable occurrence considering the absence of hair in other parts of the body.

These supraclavicular deposits and this interscapular growth of fine hair were noted by Curling and/

and it is interesting to note that he attributed the fatty deposits and the defective cerebral development to the abolition of the function of the thyroid gland. In fact, so far back as the year 1850 Mr Curling correctly indicated the true pathology of the disease. He stated, "I am not acquainted with any case on record in which a deficiency of the thyroid gland has been observed, in the human subject."

Apart from the interest which must attach to the cases just related from their great variety, the development of adipose tissue forming symmetrical swellings in the neck, cannot fail to add to their importance, for it is highly probable that this abnormal secretion of fat is dependent on the absence of these changes which result from the action of the thyroid, or from some imperfection in the assimilating processes consequent on the want of this gland.

The facts detailed may not be without significance in directing the researches of future inquirers into the use of this body.

In countries where cretinism and bronchocele prevail it has long been supposed that there was some connection between the defective condition of the brain and the hypertrophy of the thyroid.

Pathologists/

Pathologists have recently been inclined to view the coincidence of these two affections as accidental and as having no direct relationship. Enlarged veins are often present over the upper part of the front of the chest, and venous mottling of the limbs is not uncommon.

The abdomen is always enlarged, often enormously so. Herniae, usually umbilical, but sometimes inguinal, are often present. In many cases the belly is protruding and pendulous, while the back is arched. (lordosis). The limbs are short and broad. The lower limbs often curved and apparently rickety.

In some of those cases in which the long bones are curved their ends may not be enlarged; there may be no beading of the ribs and the skull may present none of the characteristic features of rickets.

The hands and feet are broad, thick and swollen looking - spade like: they are cold and often somewhat purple coloured.

The skin of the face, neck and abdomen may be fine and wax-like. Over the limbs and back it is usually rough,harsh and dry.

The secretion of sweat in all cases is greatly diminished or entirely absent. A fine, furfuraceous desquamation/

desquamation is often seen on the skin of the back and over the lower extremities. Eczematous eruptions on the skin frequently occur; ichthyosis - like patches are not uncommon.

The presence of moles, warts and naevi has in some cases been noted. The voice is rough, hoarse, harsh or squeaky.

The internal (rectal) temperature is usually subnormal. The surface temperature is almost invariably subnormal.

Patients affected with sporadic cretinism are extremely susceptible to cold, they like to bask in the sun or toast themselves before the fire. They are usually much more active in warm than in cold weather. The gait is clumsy and waddling. In some instances the patient is unable to walk alone. It is only in slightly developed cases that the patient is able to run and even in these cases the movements are stiff and clumsy.

The subjects of sporadic cretinism show a singular repugnance to exertion both of body and mind. In many instances they will sit perfectly still in one position for hours together without speaking or apparently taking any interest in their surroundings, and yet perfectly happy and contented. The degree of mental development to which sporadic cretins attain/

attain differs considerably in different cases. Some are little better than idiots, unable to speak intelligently or to dress or feed themselves. Others though 25 to 30 years of age resemble, as regards their mental development, a dull child of 4 or 5. In others the mental qualities are more advanced.

In one of the cases reported by Fagge, "the patient was very intelligent and fond of reading novels." This case seems to have been altogether exceptional. It can hardly be regarded as a typical case of sporadic cretinism, for the symptoms did not begin to develop till 8 years of age; the limbs were small and well formed; the manner as far advanced as is usual at her age (16½ when she was examined). Menstruation had appeared at the age of 15 and had up till the date of examination been regular. As has been stated the mental development was for a sporadic cretin exceptionally advanced. Furthermore, after death, the thyroid gland though diseased, for there was a tumour in the right lobe, was not absent as has been the case in every instance of sporadic cretinism in which the condition of the gland has been ascertained after death.

The subjects of this disease are usually of a placid/

placid and affectionate disposition. Like patients affected with myxoedema, sporadic cretins are usually very unemotional.

Dr Langdon Down states as one of their characteristics that they are extremely placid, unruffled in temper, that they seldom cry and never shed tears.

They are generally cheerful and easily amused: they are usually fond of playing with children and in some cases they possess the faculty of humour and imitation.

In Dr Robinson's case although speech was rudimentary and the patient was unable to do more than partially pronounce a few words "octor" for Doctor, and so on, he could imitate the mewing of a cat: he also imitated the same animal when asked to mimic any other beast. Patients affected with the disease are generally cleanly in their habits, in many cases modest in their demeanour, and, considering the low state of mental development, singularly careful of allowing themselves to be exposed. Bourneville had great difficulty in persuading one of his patients to strip herself for the purpose of being examined. The same patient when dressing or undressing at home took infinite pains to prevent any exposure of her person even although her sister alone was present.

Patients with sporadic cretinism usually sleep/

sleep well.

A notable and characteristic feature of the disease in typical and well-marked cases is the arrested development of the sexual organs. Even at 25 or 30 years of age there is no hair on the pubes or axillae. In one case of my own, the patient, though 47 years of age, had no hair in these regions.

The testicles, ovaries, and uterus are in most cases entirely undeveloped. As a rule the menstrual function is not established, or if it does occur the flow ceases after one or two periods. The pelvis is narrow and contracted in comparison with the abdomen and upper part of the trunk.

The appetite is in most cases moderate or capricious; the breath is often foul.

In many cases there is constipation, in others intestinal derangements and diarrhoea are readily induced by slight dietetic errors.

The liver and spleen are usually normal. A slight degree of enlargement of the lymphatic glands in the neck and axillae is not uncommon. As a rule the circulatory, respiratory and urinary functions are not deranged in any notable degree. The pulse is usually small and feeble and in some cases somewhat quicker than normal.

Shortness/

Shortness of breath on exertion has in some cases been noted, but since most sporadic cretins are neither able nor willing to exert themselves actively, this symptom may not be observed. The duration of the disease is variable. Some patients die in infancy and childhood; not a few attain to adult age.

Several cases are on record in which the patients have attained the age of 30 or more.

I myself have a case at the present time of a female aged 47 and the little creature is apparently as well as ever she has been.

Diagnosis:

The diagnosis in typical and well-marked cases presents no difficulty. The arrest in the physical and mental development; the peculiar and most striking facial physiognomy; the earthy colour of the complexion; the shortness and thickness of the neck; the depression in the position of the thyroid gland; the supraclavicular fatty swellings; the large swollen abdomen; the presence very often of an umbilical or inguinal hernia; the thickness and shortness of the limbs; the short, broad, swollen hands, and the broad swollen feet; the rough, harsh, or squeaky character of the/

the voice; the dry, harsh condition of the skin; the absence of sweating; the lowered temperature; the marked susceptibility to cold and the love of warmth; the dislike to exertion; the marked torpidity of the body and mind; the arrested sexual development; and the solid subcutaneous oedema which does not pit on pressure: all of which symptoms are usually present in typical and well-developed cases form a clinical picture which is not easily mistaken.

Prognosis:

In itself the disease is not a fatal one, death as a rule being due to some intercurrent affection such as convulsions, erysipelas, pneumonia, bronchitis, oedema of the lungs, etc. Once a case has come under treatment, the prognosis is favourable in proportion to the age of the patient.

Treatment:

The statements which were to be found in the text-books and indeed in the best and most extended monographs a few years ago with regard to the treatment of cretinism were of the most meagre and unsatisfactory character.

On consulting the "Theory and Practice of Medicine by Frederick T. Roberts" eighth edition, 1890/

1890, I find that this disease is not even mentioned. Since that time a revolution has taken place in the treatment of the disease, with the result that at the present day the treatment of cretinism by thyroid extract or freshly prepared sheep's thyroid gland is one of the greatest triumphs of modern therapeutics.

From a consideration of the etiology, pathology, and clinical history of cretinism, it would appear that the main objects of treatment should be firstly, to protect the patient against everything likely to aggravate the symptoms or accelerate the progress of the condition.

Secondly, to endeavour, as Victor Horsley has suggested, to supplement the function of the atrophied and degenerated or entirely absent gland, as the case may be, by grafting new and healthy thyroid gland tissue into the body of the patient , or by supplying the necessary want by feeding them on thyroid gland.

In carrying out the first indication, the protection of the patient from cold is by far the most important point and is insisted upon by all authorities.

As has already been seen in the study of the clinical history of the disease, cretinoid patients almost/

almost without exception are greatly affected by cold and are worse in cold and better in hot weather.

Horsley's experiments have conclusively shown that exposure to cold materially aggravates the symptoms of the disease produced in lower animals.

At the present day the treatment of cretinism resolves itself into the administration of extract of thyroid gland and the marked and rapid improvement in these cases during this treatment has been vividly brought before us within the last few years.

The same remarkable results I have been able to verify, viz., a great and rapid diminution of bulk, due to absorption of myxoedematous deposits, seen especially in the collapse of the protruding abdomen, in the spontaneous reduction of umbilical herniae, in the recession of the previously swollen tongue behind the teeth, in the disappearance of baggy swellings under the skin, above the collar bones, outside the nipples and elsewhere; in the thinning of the dropsy-like puffiness of the face, limbs, and other parts of the body.

A great and rapid increase in physical development shown especially by a rapid growth of several inches in height even in cretins from 20 to 30 years of age, whose stature had been nearly or quite stationary for many years previously; also by/

by the replacement of the coarse, sparse hair by a finer and more abundant growth; by the eruption in quick succession of teeth which had been long overdue.

A striking diminution of several deformities especially lordosis in the lumbar spine, of the bulky head, of the ugly sinking of the bridge of the nose and sometimes of the rickety curvature of the legs. A rapid and very striking increase of intelligence occurs, as can be well seen by comparing the dull, stupid, heavy, listless, often idiotic countenance before treatment, with the bright, cheery, pleasing expression which soon takes its place.

In commencing treatment it is well to begin with a small dose, say 3 grains a day, and to increase it gradually to five or more grains according to the effect produced. If a large initial dose be given, symptoms of great depression may show themselves - vomiting, headache, cold sweats, fall of temperature followed by a rise to 103°F. or higher, and signs of heart failure.

Judging from the different degrees in improvement in cases that have been published, I think the conclusion that "in cases of equal degree, the younger we can commence the treatment, the quicker will be the improvement," is borne out.

I am interested in the case (photo I) which/

which is at present under observation and speculate whether, in this case of sporadic cretinism detected while the child is in its infancy and before its mental and physical constitution has become impressed with, one might almost hope the not entirely eradicable stamp of the disease, where treatment has been commenced so early and the physiological defect supplied by thyroid administration, the child might not grow up in an almost normal condition.

This stage of cretinism certainly appears to be the one which offers the most hope of improvement from early and continuous medical treatment.

Transplantation of the thyroid has been practised by Dr Lanelongue (reported in Lancet, March 22nd, 1890) in a case of sporadic cretinism.

Several German surgeons, notably Boccher, and in this country Victor Horsley, Clutton and others had tried the implantation, either in the abdominal cavity or beneath the skin, of portions of thyroid glands of sheep, or of parenchymatous bronchoceles from human subjects. The effect though favourable, was transitory and disappeared with the absorption of the implanted tissue.

In 1892 Dr Murray showed that subcutaneous injection of an extract of the thyroid gland caused alleviation of symptoms, and later Dr Mackenzie showed/

showed that feeding by the mouth was equally efficacious.

As a point in the practical treatment of these cases, Dr Telford Smith, Royal Albert Asylum, Lancaster, has found that during thyroid treatment the rapid growth of the skeleton leads to a softened condition of the bones, resulting in a yielding and bending of those which have to bear weight, and as cretins under treatment become much more active and inclined to run about, this tendency to bending has to be guarded against. (Lancet, October 2nd, 1897, page 853)

Case I.

Family History:

The child's father, who is 27 years of age, has always been healthy. He is temperate and fairly intelligent. He was born and always has lived in Sunderland. The mother is 24 years of age; she has always been healthy, before and since marriage. She was also born in Sunderland. She had always an easy time during labour. There is no history of a fright or accident during pregnancy.

There is no history of phthisis in either family; the father and mother of the child were not related, and there is no history of consanguineous marriages in their families. The child has two sisters, both healthy children; no near relation is afflicted with cretinism or myxoedema.

History of Patient:

Patient was born on January 7th, 1899 and was a fine healthy child at birth. No instruments were used. Has had no convulsions nor febrile attacks; was never sick; was often constipated and needed castor oil from birth. As a rule she was a good child; was sometimes cross when constipated. She was on the breast which she took very well till admission/

admission to hospital. The symptoms were first observed at the age of 12 months, when it was thought that the child was suffering from weakness of the back,and which it was thought by the parents would disappear as the child got older.

At this time the child always lay still on her back and did not offer to move - hence the above conclusion come to by the parents.

She was admitted to the Monkwearmouth Hospital, Sunderland, on May 1st, 1900, under my care at the age of one year and four months, on account of her inability to swallow, which was caused by a great swelling of the tongue, which not only entirely filled the mouth, but protruded between the lips, presenting a most unique pathological condition.

The enlargement of the tongue in this disease is of course not an unexpected occurrence, but the presence of such an abnormal enlargement which almost necessitated surgical interference must, I think, class this as an interesting example of an interesting disease.

The child was at first fed by rectum and nasal tube, but on account of the ensuing diarrhoea rectal feeding had to be discontinued a week after commencement. She was then fed by nasal tube alone, but soreness of the nose followed and feeding/

ing by the oesophageal tube had to be substituted.

At this time the swelling of the tongue had to a great extent abated and feeding in small quantities by the mouth was persevered in until eventually the oesophageal tube was abandoned.

Description of the patient on admission to Monkwearmouth Hospital:

Date, May 1st, 1900.

The expression of the child was dull and heavy. The face was large for the body and the head relatively to the face was small; the face was swollen, pale and wax-like. The forehead was low and narrow and the eyes wide apart. The eyebrows were sufficiently abundant, the eyelids swollen; the eyes grey in colour, and the pupils dilated. The nose was broad and flat. The mouth was very large (as will be seen in photo). The ears were well formed and normal in size and position. The tongue was greatly enlarged as mentioned above. The hair of the head was fairly abundant and was of a light brown colour. The anterior fontanelle was not closed. The skin of the scalp was scaly.

The circumference of the head at the middle of the forehead was 18 inches. From the root of the nose to the external occipital protruberance - tape-measurement was 11 inches. From one external auditory/

auditory meatus to the other was 12½ inches. The neck was short and thick and measured 10 inches in circumference. The head had in fact the appearance of being fitted on to the shoulders without any neck intervening between it and the body. The lower rings of the trachea could be felt. The thyroid gland was completely wanting. A small elastic swelling could be made out in its situation on each side of the neck above the clavicle in the posterior triangle.

The abdomen was large and broad; an umbilical hernia was present. The tape measurement at the level of the umbilicus was 18 inches. The chest girth was 17 inches. The whole of the chest wall appeared to be covered with a thick pad of fat. The back was slightly curved. The limbs were remarkably short, stout, firm and round. The hands were characteristically spade-like. The feet were long and narrow. The hands and feet were always very cold.

There was evident cyanosis, on which account she was ordered brandy. She lay on her back and moved her hands and feet but never attempted to walk. The knee jerks were slightly exaggerated but equal. The plantar reflex was normal.

The skin was of a yellowish colour and in texture, not only over the face and ears, but over the entire/

entire body was fine. The skin felt cold. For her age she did not understand. She slept well. Her bowels were constipated. The urine was free from albumen. The circulatory and respiratory systems were normal, as were the liver and spleen. There was no enlargement of the lymphatic glands.

Treatment: On admission the child was sixteen months old and weighed 17 pounds. As has already been noted under "History of Patient", the child was fed by rectum and nasal tube and by this means thyroid extract in one grain doses was administered daily, - occasionally by the rectum but more frequently by the nasal tube. On account of the ensuing diarrhoea already mentioned, the nasal tube had to be entirely depended upon for the administration of the drug. For the first week, little or no improvement was noticed, then the swelling of the tongue began gradually to subside, and at the end of fifteen days the enlargement was no greater than might be expected in an ordinary case of cretinism.

Feeding by the mouth was then resorted to. For the next three or four months there was no appreciable change in the condition, with the exception of loss of body weight. After this signs of/

of improvement were noticed, viz. decrease in bulk; child seemed brighter; the skin felt warmer to the touch; the hands had slightly improved in shape; the abdomen had diminished in size, and the movements of the limbs were more active.

The dose of the extract was now increased to 1½ grains daily. From this time onward the improvement steadily continued. The umbilical hernia gradually disappeared, as will, I think, be demonstrated by comparing photographs Nos.1 & 3.

The diminished bulk in the size of the abdomen can also be made out by comparing photographs No.2 and No.3. As has been remarked, the body weight gradually decreased during the first few months of improvement, but with continued treatment the child now began to gain in weight.

She was now able to sit up in bed when placed in the sitting posture. A month later she could get up with a small amount of assistance, such as getting hold of either arm, steadying herself by getting hold of the side of the bed with her free arm, and also supporting herself ~~and also supporting herself~~ in rising from the sitting position. She had now been undergoing thyroid treatment for nine months. At the end of the tenth month she could stand, leaning on the bed or supported by the wall behind her.

The/

The characteristic supra-clavicular swellings had now entirely disappeared and, in fact, to all appearances the child's body had assumed a normal condition.

The following will give some idea of the times of eruption of the teeth. Four months after admission, i.e. at the age of one year and eight months, the two lower central incisors came through the gum; two weeks later the right upper incisor appeared, and one week after this the right upper molar was visible. At the age of two years and two months she had sixteen teeth.

She was under my care for ten months, at the end of which time she was discharged from the hospital, as will be seen from the above description in a greatly improved condition.

Most unfortunately, the parents soon after removed from the district and I am unable, as I intended, to further supplement the notes on the improvement which I expect would have taken place with continued treatment.

The accompanying charts will show at a glance the variations in temperature observed during the first few months of treatment by thyroid extract, followed later by a subnormal temperature which, in this case, remained as such.

Case 2.

Margaret Etherington, age 48: single. The most remarkable feature in this case is perhaps the age, for in no records on the subject of sporadic cretinism can I find a patient who has approached this time of life. In the present instance the little creature is 48 and apparently as well as ever she was.

The family history is good and presents nothing abnormal; all the other members are strong and healthy men and women.

She is 3 feet 9 inches in height. Her gait is slow and waddling, like that of a child of three or four years. The hair of the head is dry and coarse, but fairly abundant; the scalp is scurfy and the hair is of an auburn colour. Her skin is harsh and rough; her voice squeaky; she seldom speaks and when she does it is usually in monosyllables, e.g. yes and no, and these she pronounces in a slow, thick, childish voice. Her teeth are short but sound. The mammae are very slightly developed. Pubic hairs are absent. She has never menstruated. Perspiration has never been noticed. There are no herniae present. The thyroid gland is completely wanting. The supraclavicular swellings are present.

Urea is diminished; there is no albumen in the/

the urine. She is placid in temper till provoked, when she gets quite sullen. Her favourite posture is sitting on a little chair in the kitchen with her back to the door of the oven. She is very sensitive to cold. She dislikes exertion. In other respects her description tallies with that of a typical cretin.

Her relatives, who are of the mining class, have always regarded her as something "not canny", and would not even permit medical treatment in her case.

DISEASE.

Retinitis

Notes of Case.

Name { Catherine Richardson

Age 16 months

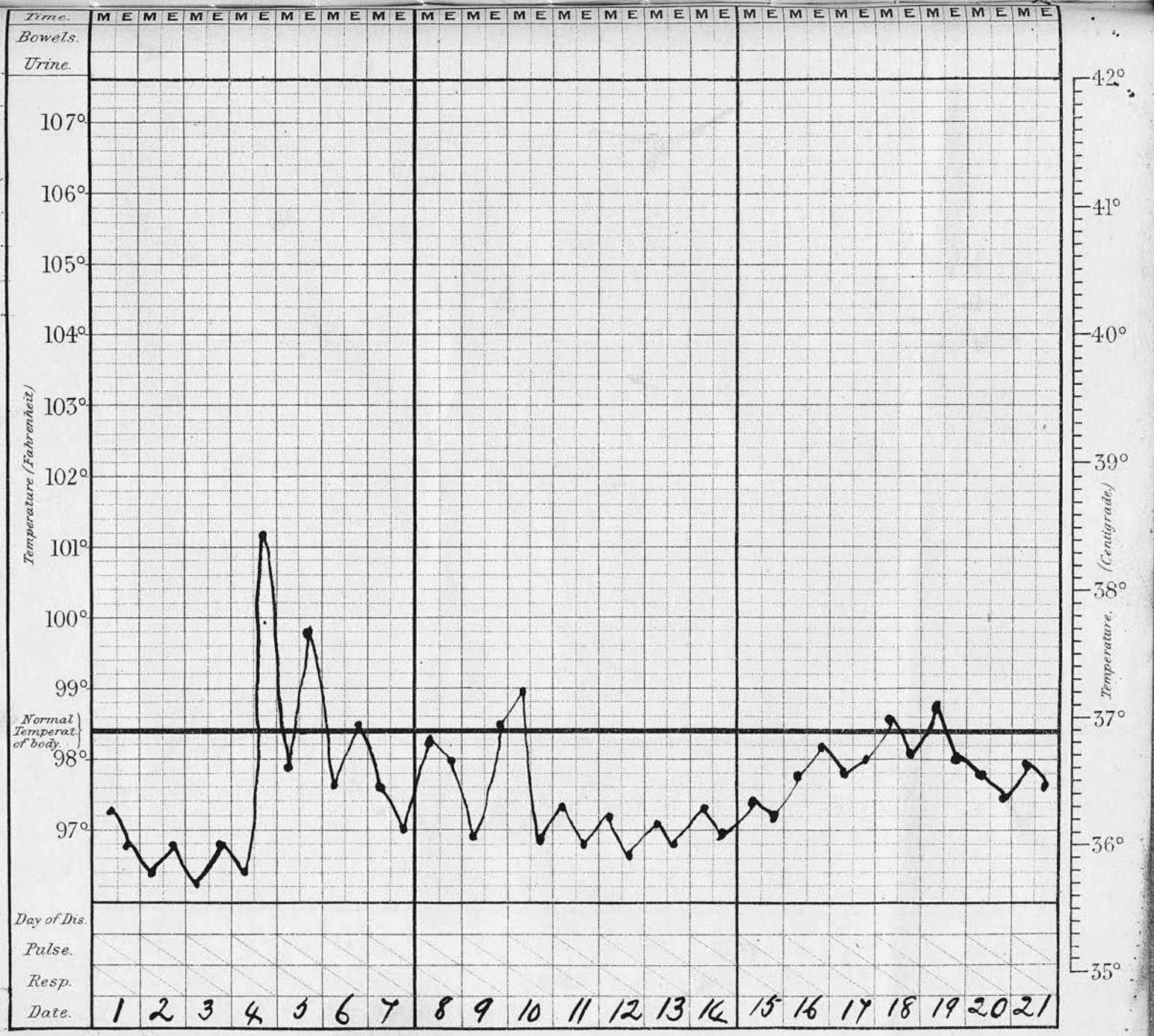
Diet

Case Book No.

Date of admission.

May 1st. 1900.

Result



Entered at Stationers Hall. Printed and Published by Wodderspoon & Co. 7 Serle Street Lincoln's Inn Gould's Clinical Chart.

DISEASE.

Cretinism

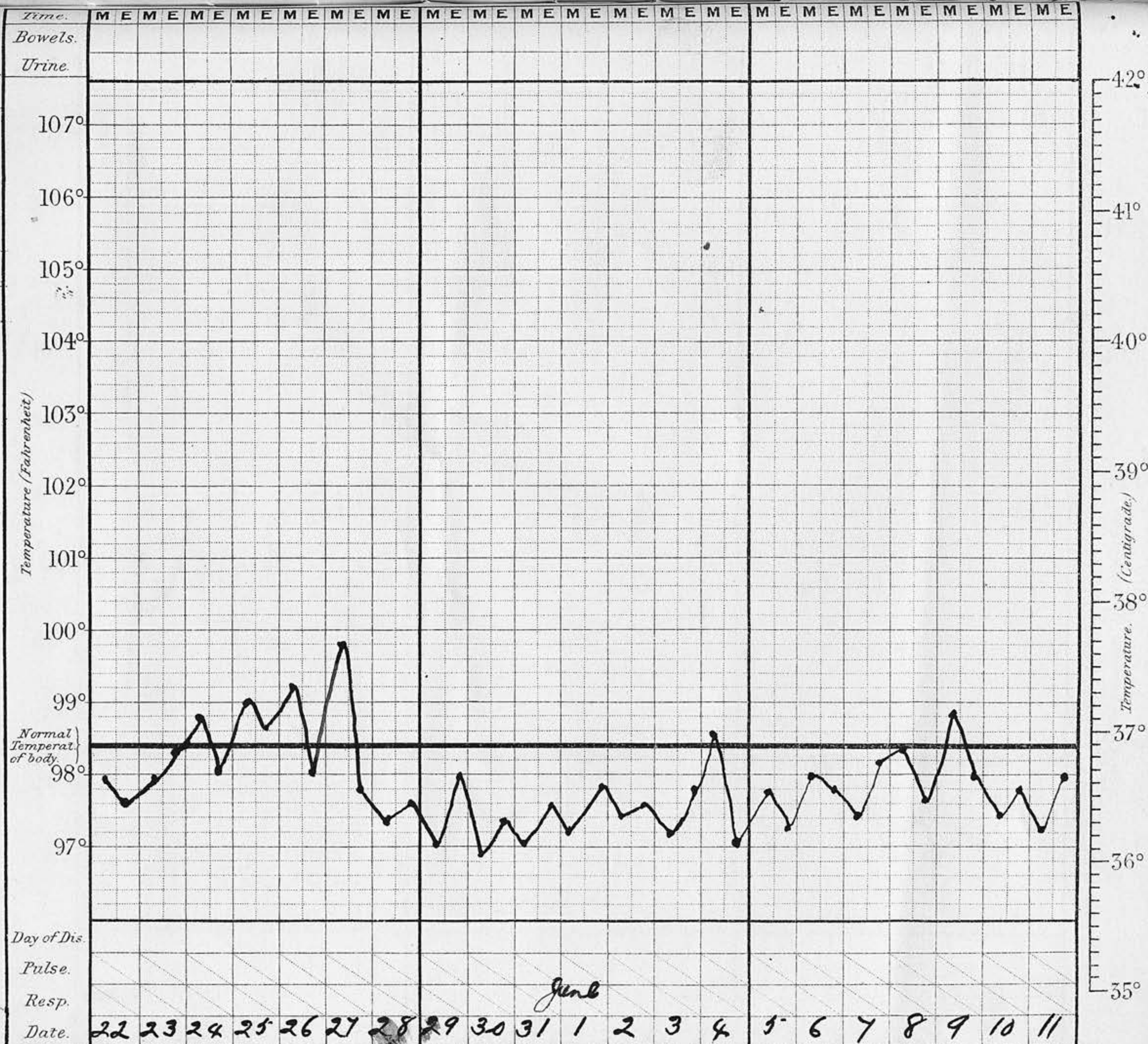
Notes of Case.

Name: Catherine Richardson

Age: 16 months

Diet

Case Book No.



Date of admission.
May 1st 1900

Result

Entered at Stationers Hall.

Printed and Published by Wodderspoon & Co. 7 Serle Street Lincoln's Inn.

Gould's Clinical Chart.

DISEASE.

Cretinism

Notes of Case.

Name { Catherine Richardson

Age 17 months

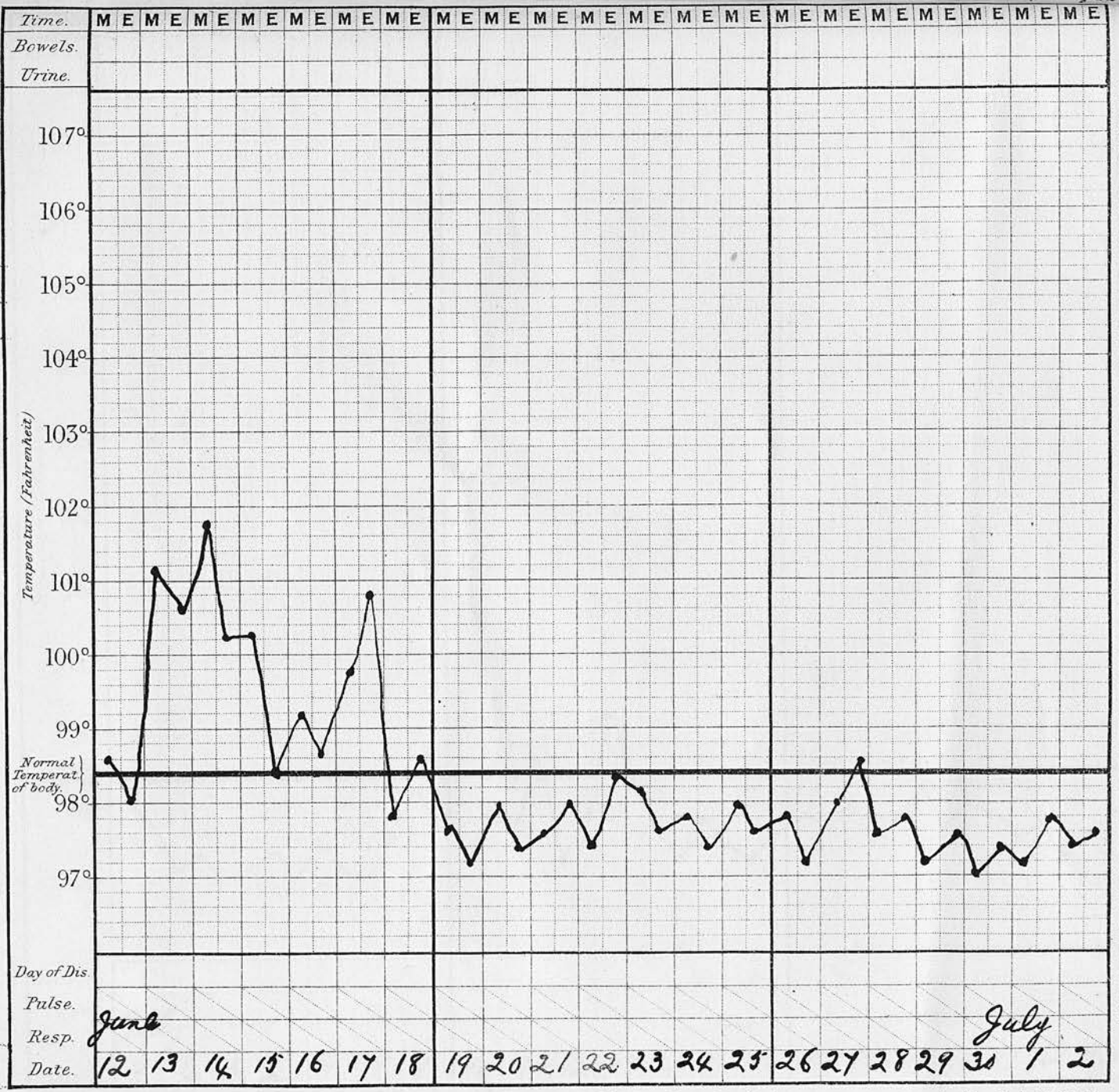
Diet

Case Book No.

Date of admission.

May 1st. 1901

Result



Entered at Stationers Hall. Printed and Published by Wodderspoon & Co. 7 Serle Street Lincoln's Inn. Gould's Clinical Chart

DISEASE.

Cretinism

Notes of Case.

Name { Catherine Richardson

Age 18 months

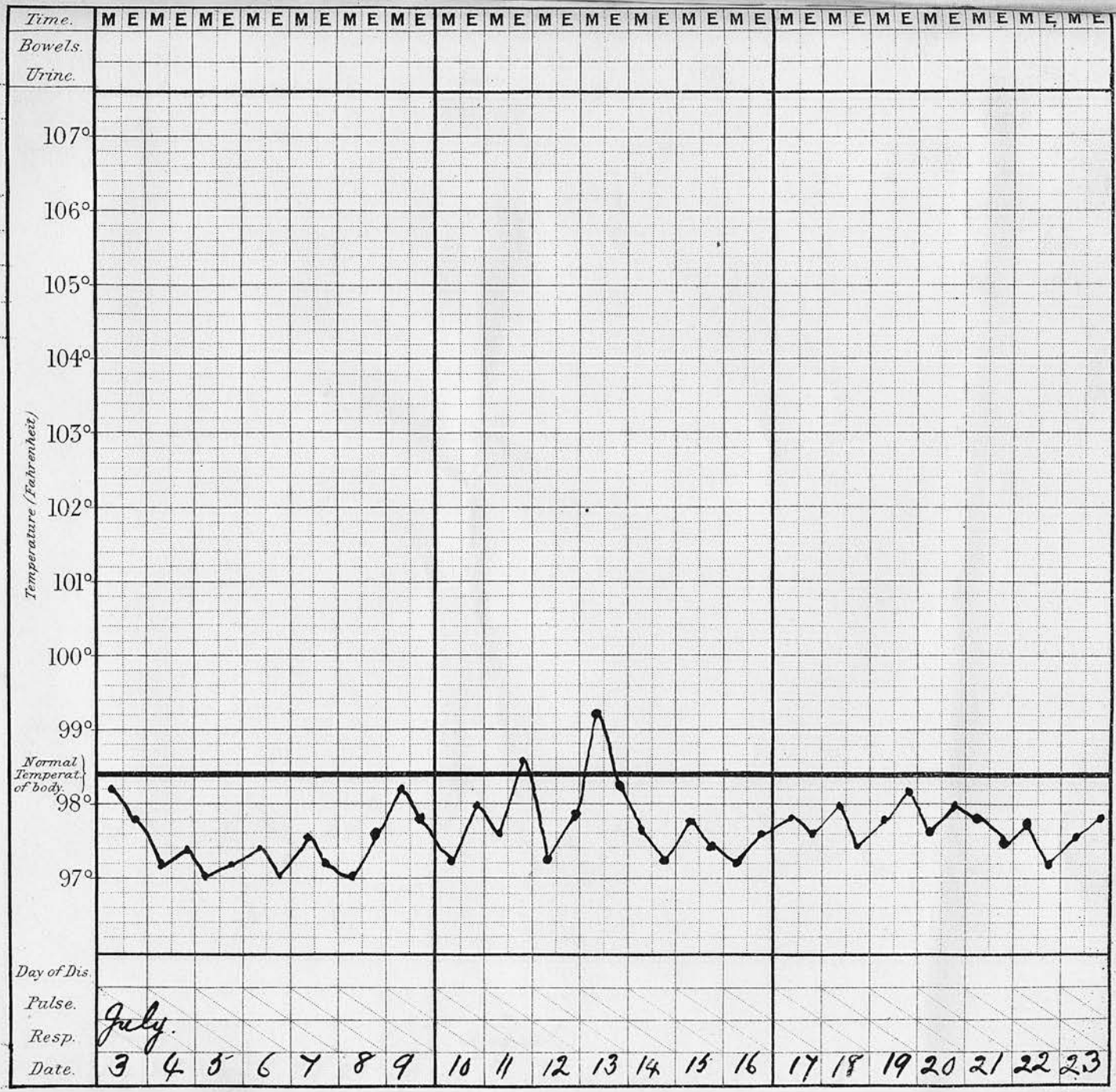
Diet

Case Book N°

Date of admission.

May 1st.

Result



Entered at Stationers Hall. Printed and Published by Wodderspoon & Co. 7 Serle Street Lincoln's Inn Gould's Clinical Chart.

DISEASE.

Cretinism

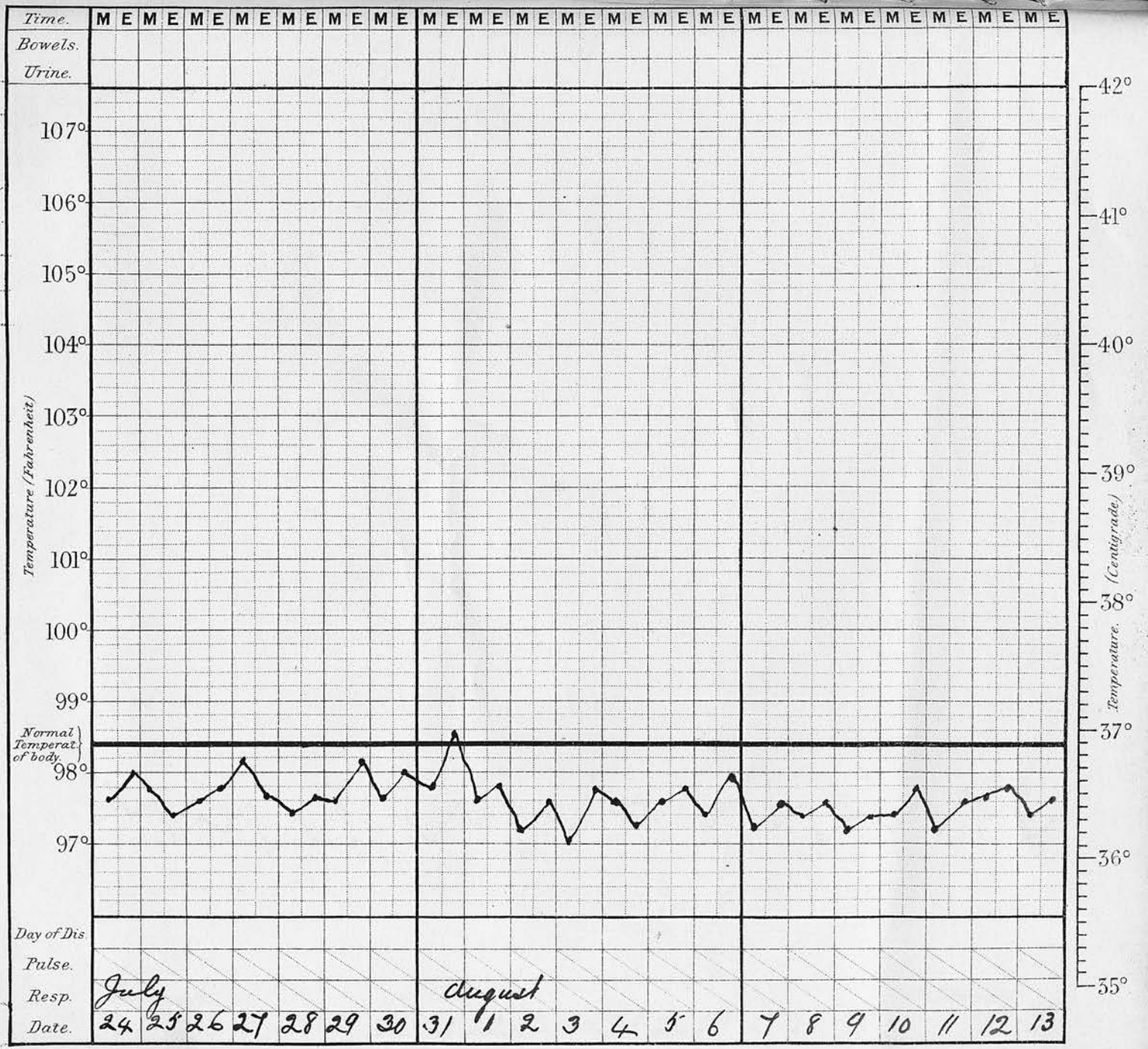
Notes of Case.

Name { Catherine Richardson

Age 18 months

Diet

Case Book No.



Date of admission.

May 1st 1900

Result

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DISEASE.

Cretinism

Notes of Case.

Name { Catherine Richardson

Age 19 months

Diet

Case Book N°

Date of admission.

May 1st 1900

Result

Time. M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E M E

Bowels.

Urine.

Temperature (Fahrenheit)

107° 106° 105° 104° 103° 102° 101° 100° 99°

Normal Temperat. of body.

98° 97°

Day of Dis.

Pulse.

Resp.

Date. 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30 31 1 2 3

August September

42° 41° 40° 39° 38° 37° 36° 35°

Temperature (Centigrade)

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DISEASE.

Cretinism

Notes of Case.

Name { Catherine Richardson

Age 20 months

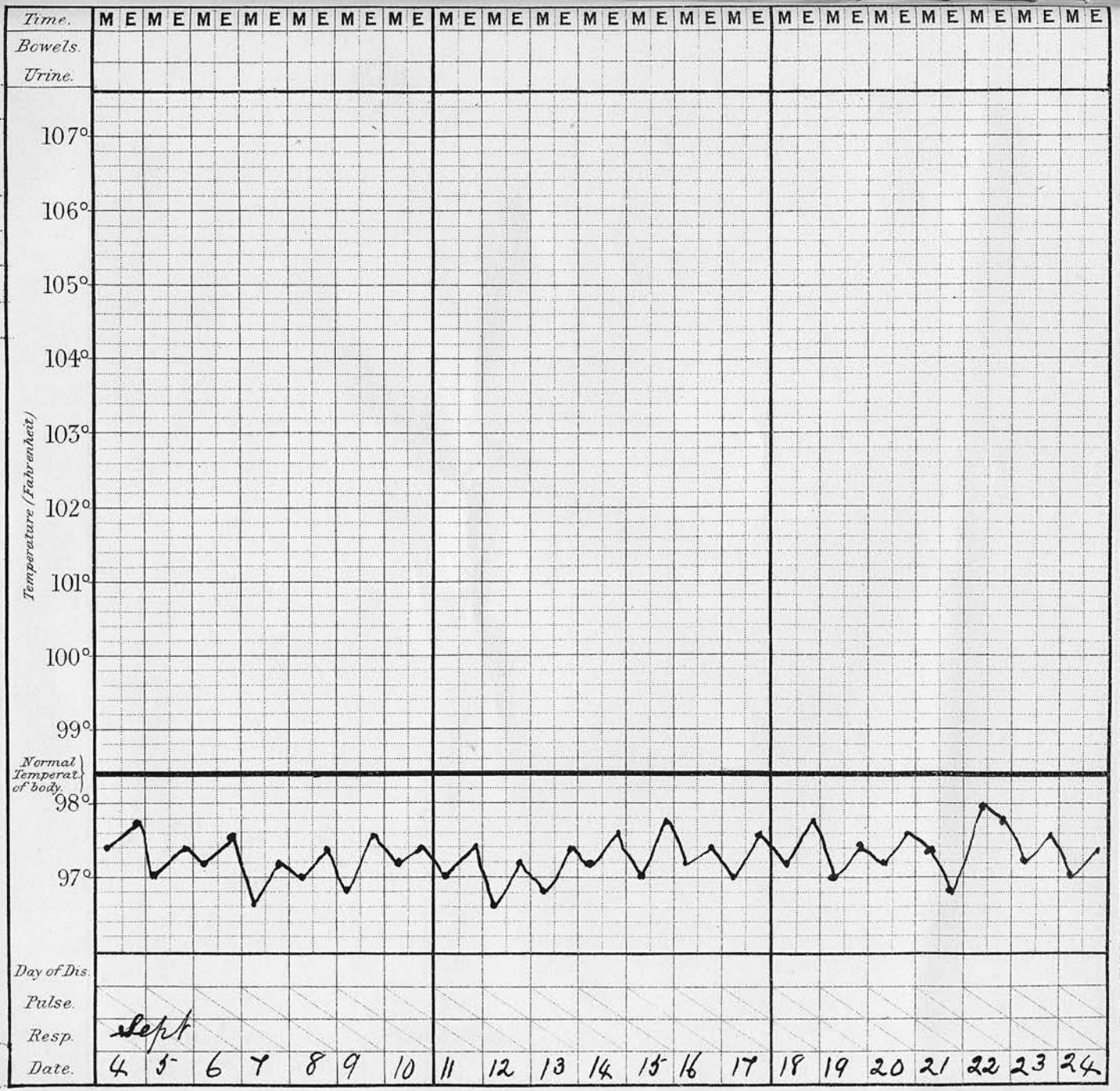
Diet

Case Book N°

Date of admission.

May 1st 1901.

Result



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DISEASE.

Cretinism

Notes of Case.

Name { Catherine Richardson

Age

Diet

Case Book N°

Date of admission.

May 1st 1901.

Result

Time.	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E	M E
Bowels.																														
Urine.																														

Temperature (Fahrenheit)

107° 106° 105° 104° 103° 102° 101° 100° 99°

Normal Temperat of body. 98° 97°

Temperature (Centigrade.)

42° 41° 40° 39° 38° 37° 36° 35°

Day of Dis.								
Pulse.								
Resp.	September						October	
Date.	25	26	27	28	29	30	1	2

Entered at Stationer's Hall. Printed and Published by Wodderspoon & Co. 7 Serle Street Lincoln's Inn Gould's Clinical Chart.

Chas. E. Cooper

Chas. E. Cooper

Holmeside
SUNDERLAND.