THE BRITISH JOURNAL OF OPHTHALMOLOGY

APRIL, 1943

COMMUNICATIONS

ON SOME ANOMALOUS FORMS OF AMAUROTIC IDIOCY AND THEIR BEARING ON THE RELATIONSHIP OF THE VARIOUS TYPES

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Tay, in 1881, described a familial disease of infants in which, within the first year of life, there appears at the maculae of both retinae a cherry-red spot surrounded by a fairly well-defined white area. The child becomes weak, unable to hold up its head or move its limbs and finally completely paralysed and progressively demented. Blindness develops, the disc becoming white and atrophic. Hyperacusis is often present. At the onset it may be overweight but it soon begins to waste and usually dies before the age of two years, only exceptionally surviving the third year (see later). Major epileptic fits rarely occur, but various sudden mass reflex phenomena are common. Sachs, in 1887, described the autopsy findings and called it amaurotic idiocy, while Higier, in 1901, suggested the name Tay-Sachs' disease. It was first thought to be confined to Jews, but although occurring predominantly in Jews occasional non-Jewish cases have been recorded (see later). The disease has been reported from all parts of the world, including Japan. Slome, in 1933, found it to be inherited as a recessive character, as 111 cases occurred in 69 families containing two or
more members and consanguinity of the parents was present in over 50 per cent. of the cases. In one family the disease occurred in one of twins and it has been found in collateral branches of a family.

At autopsy, a brain of firm rubbery consistency is found, usually smaller than normal, with widened sulci. Microscopically the nerve cells, dendrites and, less commonly, the axis cylinders show generalised swelling with diffuse gliosis, the normal pyramidal outline of the cells being changed to a round or pear shape. The Nissl's bodies are reduced or absent revealing a fibrillary network, filled with a finely granular substance displacing the nucleus to the periphery. The latter stains poorly and ultimately disappears with the rest of the cell contents. Fat stains show the granules in the cells to be formed of pre-lipoid lecithin-like material. These changes are present throughout the central nervous system including the cerebellum, but the localization of the most marked changes varies from case to case. The ganglion cells of the retina are heavily affected leading to an enlargement of the normal small red area present at the macula and allowing more of the choroidal coat to show through, while the lipoid in the ganglion cells produces the opaque whitish appearance of the surrounding zone, the colour of which may vary according to the intensity of the process. Lipoid degeneration of the ganglion cells may occur, however, without giving rise to a cherry-red spot (Schlesinger, Greenfield and Stern). The periphery of the fundus is generally normal, though pigmentary changes have been described. In the early stages the discs are normal, but later they are atrophic. The final stages, if the patient survives, have not been described but were seen in our Cases 4, 5 and 10, and consist of a gradual spreading of the white area to involve the whole retina which atrophies with pigmentary changes. The red spot disappears and is replaced by a reticulated, white and circular area and the disc becomes dead white.

Several cases of this type of the disease are now mentioned, some of them being atypical.

Case 1. Gloria G. Female, aged 10 months. Attended Out-Patients' Department, Hospital for Sick Children, London, on July 26, 1938, under the care of Dr. R. Lightwood.

Family History.—Both parents Jewish from the East End of London. Patient was the only child. No family history of blindness, idiocy or consanguinity.

History.—Normal pregnancy and delivery, breast-fed and just weaned. Taking food well but recently seemed to take no interest in things around her. Did not sit up, had a poor grip and could not hold things.
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Examination.—Weight 17 lbs. 10 ozs. (about normal); vacant expression but appeared healthy; her eyes followed lights, pupils reacted normally but were both large. In the fundi there was a cherry-red spot at both maculae with striking pallor of the surrounding area. The discs were pale, trunk muscles flabby, legs spastic, knee and ankle jerks present, plantar responses extensor. W.R. negative. Further progress not known.


Family History.—Only child of Jewish parents living in the East End of London. No consanguinity or similar disease in family.

History.—Normal pregnancy and delivery, rapid gain in weight and growth but never attempted to grasp objects. At six months of age, arms, hands, face and thighs said to be “puffy” apparently due to abnormal deposits of fat. It was then noticed that the muscles of the head and trunk seemed weak. The child could not sit up or raise its head, and progressive mental dullness and weakness appeared.

Examination.—Marked general weakness, moved little, pupils large, reacting to light. Fundus: cherry-red spot present at both maculae, optic discs normal. Legs spastic, abdominal reflexes, knee and ankle jerks absent, bilateral extensor responses. Liver and spleen not palpable.

Investigations.—Serum cholesterol 285 mgms. per cent. (normal = 100-230). Total fat in faeces 33.3 per cent., made up as follows: combined fatty acids 25.2 per cent., free fatty acids 3.5 per cent., unsoaped fatty acids 8.1 per cent., neutral fat 46 per cent. The total fat content is somewhat high but probably within normal limits, but the ratio of split to neutral fat is normal.

The child was treated with a low fat diet but died within a fortnight. Permission for post-mortem examination could not be obtained.

Case 3. Rose C. Female, aged 1 year 9 months. Admitted to the National Hospital, March 27, 1912, under the care of Dr. Gordon Holmes.

Family History.—Father and Mother, English, born in the East End of London. No consanguinity, or Jewish blood in family. No relatives known similarly affected. The patient was the elder of two, a brother aged five weeks was apparently normal.

History.—When five to six months old it was noticed that the child seemed dull, could not hold out her hand or attempt to sit up, and took no notice of her surroundings. At the age of ten months she was able to say several words, could suck an
orange if her mother held it to her mouth, but she had gradually lost the ability to do either of these. The child apparently had difficulty in seeing, but seemed able to follow lights. The legs recently always seemed cold and her hands twisted about stiffly. She had become very constipated and the abdomen was swollen. For the last three months there had been increasing difficulty in swallowing, especially of late, and the child was only able to drink out of a feeding bottle. In the last fortnight before admission she had been having curious attacks (which were preceded by a moaning noise) in which her hands clenched, her legs flexed, her eyes rolled and she seemed to be trying to raise herself up. She might have as many as thirty attacks a day. In addition there was very marked hyperacusis, the child being easily startled at the slightest sound and screaming as if "nervous and frightened."

Examination.—The feet were red and very cold, abdomen protuberant, a few moist râles in chest, lay on one side with a vacant expression, with head a little retracted and limbs held very stiffly extended, with feet inverted. Attempts to move her caused her to cry out. She screamed if a sudden noise was made near her and occasionally seemed to recognise a little when called by name, but otherwise took no notice of her surroundings. Frequent sucking movements of the lips occurred. She seemed unable to see even a bright light. Pupils large, central, circular and inactive to light. Fundi: discs perfectly white, vessels of good calibre, macular area surrounded by a whitish area a little wider than the disc with a bright red spot in the centre. No nystagmus. Arms extended. Would not grasp anything or make voluntary movements. Spastic legs with generalised wasting. Spasticity increased on handling the arms. Reflexes: normal in arms, knee jerks present, ankle jerks not obtained owing to spasticity, abdominals just present, plantars extensor.

Discharged from hospital May 12, 1912, having gradually gone down-hill.

The L. Family

Mother and Father, alive and well, both born in Gillingham, Kent. No consanguinity, Jewish blood or blindness in family. The mother has had eight pregnancies, the first child being a girl now aged eighteen, alive and well, the second a girl, now aged seventeen (Case 4), the third a girl now aged fourteen, alive and well, the fourth a girl now aged eleven, alive and well. The fifth pregnancy resulted in a miscarriage, the sixth in a boy now aged seven (Case 5). The seventh pregnancy also resulted in a miscarriage, and the eighth in a boy who died at the age of two years and five months (Case 6).
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CASE 4. Betty L. Normal pregnancy and delivery. Developed normally until about nine months of age when she seemed backward in speaking. At the age of eighteen months it became obvious that she could not see well and her sight became rapidly worse in the course of the next three months, at which time she began to lose the power of saying the words she had so far acquired. She did not seem to learn as other children do. She seemed dull and took little interest in her surroundings but was easily startled by noises and appeared very nervous. She was seen by Dr. Paterson at Hospital for Sick Children, Great Ormond Street, in 1926, at the age of one year and nine months. Examination of the fundi showed then a typical cherry-red spot at both maculae and extensor plantar responses. From that time the patient developed normally physically, but remained grossly deficient mentally. She rapidly became completely blind and has been in a home for mentally deficient children for the last thirteen years. She is now aged seventeen years. On examination recently the girl showed a gross dementia, was able to say only a few words and there was complete blindness and pigmentary atrophy of the whole retina, the disc being dead white. Marked nystagmus was present. The limbs were somewhat spastic, tendon reflexes brisk, abdominal reflexes absent and plantar responses extensor. There was moderate degree of ataxia in movements of limbs. The girl was dirty in her habits and had to be cared for in every way.

CASE 5. David L. Pregnancy and birth normal. Developed normally until about twelve months of age then it was noticed that his sight was failing. He became dull and although previously had a normal vocabulary for a child of his age, he began to speak less and less. He became sleepy and apathetic, taking little interest in his surroundings, but easily startled by noises. He was seen at the age of fifteen months by Mr. Penman, at the Hospital for Sick Children, and in the fundi a cherry-red spot was found to be present at both maculae with some optic atrophy. Since that time his physical condition has remained good, but he has become completely blind and demented. Examination recently at the age of seven showed a similar retinal picture to that present in his sister, but the only other physical finding was that of extensor plantar responses. He is clean in his habits and quite docile.
Case 6. Terence L. Seen at the age of five months at Out-Patients' Department, Hospital for Sick Children, August 22, 1939, by Dr. Schlesinger. Normal pregnancy and delivery, breast-fed for three months. Held up his head at three months but now was taking no notice of anything, but was easily upset by noises. He cried a great deal but would occasionally smile. Examination showed a normally developed child with a head circumference of sixteen and a half inches. He took no notice of his surroundings or of people. The pupils reacted very slightly to light. The fundi were pale and atrophic. Interlacing choroidal vessels were easily seen. The discs were pale but no cherry-red spot was present. The rest of the nervous system showed no abnormal physical signs. The spleen was enlarged one inch below the costal margin and the liver one finger's-breadth.

He continued to gain weight and at the age of eight and a half months was over-weight, now weighing eighteen pounds eight ounces. Apart from that he had altered very little but had been subject to attacks of vomiting. The physical signs were unchanged except that the retinae were even more atrophied. He was apparently completely blind, said nothing and was unable to sit up. This blind, demented child continued to gain weight but remained in the same state and was admitted to a home where it died at the age of two years and five months very suddenly from "acute heart failure."

The J. Family

Parents married thirteen years. Father aged thirty-seven came from the Forest of Dean. Mother aged thirty-four from yeoman farmer stock in the same region. No Jewish blood or consanguinity. No neurological condition in either family. The parents had four children.

First child, male, aged twelve and a half years, healthy and average at school. Second child, female, died in the National Hospital, Queen Square, aged two years and eleven months from amaurotic idiocy on April 6, 1932. Post-mortem examination was
performed and this case was reported by Greenfield and Nevin as a late infantile form. The fundus oculi showed on either side slight pallor of the disc with a large dull red spot at the macula, about one-third the size of the diameter of the disc, surrounded by a greyish opaque area shading off into the surrounding retina. The third child was the patient (Case 7), and the fourth child a female, aged eleven months, healthy, talked and had most of her teeth. She was quite different from the two preceding, when they were at the age of eleven months. The pregnancy was complicated by placenta praevia.

Case 7. A.J. Male, aged 2 years 8 months. Admitted to the National Hospital, January 5, 1935, under the care of Dr. S. A. K. Wilson.

History.—Normal birth and pregnancy, full term. Weighed eight pounds at birth. Breast-fed for nine months. Cut his teeth early. Began to say simple words at nine months. At ten months he began to show signs of "nervousness," suddenly jerking on hearing a loud noise. At the age of thirteen months after being away from his mother for a month during her last pregnancy, he would have nothing to do with her on his return. He was rather slow in sitting up and late in beginning to walk, but at the age of thirteen months he could scramble about the floor. His speech progressed slowly until the age of one year and nine months, when he understood fairly well what was said to him. About one year and seven months of age his eyes began to flicker and at this time he could see his toys quite well although he never walked alone. During the past few months he had lost a lot of his alertness, but still seemed to understand his mother and father and still laughed and sang. His speech began to deteriorate and he now spoke little. He became less agile and his legs weaker. Three months before admission he began to suffer from "fits," although he had had a kind of fit about eighteen months previously, when all his body rolled into a ball for a few seconds, without loss of consciousness, whenever he was placed on his back. The present "fits" consisted of a moving up and down of either leg for a few seconds associated with a crying noise. He would grip one of his parents as if afraid, but would be normal again in a few seconds. There was no loss of consciousness, incontinence or twitching of the arms and face. They occurred two or three times daily. Soon after their commencement they became generalised in the legs and trunk and he might bend forward and his head almost touch his knees. In others his legs became stiff and straight. The fits seemed to be brought on by movement, for example, turning quickly, and were apparently a mass reflex. Shortly after the onset of the attacks his legs
gradually became weak and he was now unable to move them or sit up. In the last few days he had hardly been able to lift up his head. Recently his vision had deteriorated. He had always had to wear napkins at night owing to incontinence, but since the fits he had lost control of both bowel and bladder completely. There was no loss of weight. He also coughed a good deal when swallowing. Recently there had been no tears when he cried.

Examination.—Fairly well nourished child, lying on right side with head flexed, crying when disturbed, sometimes an almost aphonic cry without tears. He screamed if attempts were made to turn him over. He did not speak. Apparently able to hear even a whispered voice. Almost constant oscillatory nystagmus; vision obviously impaired. Fine twitching of both orbiculares palpebrarum. Pupils normal.

Fundus: August, 1934 (Mr. Williamson-Noble). Showed on the right side a disc, pale on the temporal side with a rather bright retinal reflex in the macular region which was more conspicuous.

![Cherry-red spot at the macula in a child dying at the age of 3 years and 10 months. (Case 7).](image)
than normal. On the left the same appearances were seen, but the retina around the macula was beginning to assume a greyish-white appearance (Fig. 1).


Gait.—Made no attempt to walk, when lifted into an upright position with feet on the ground, legs becoming extended and ankles plantar flexed.

Most of the primary dentition present. Liver and spleen not palpable.

Several attacks were witnessed.

1. Patient suddenly screamed, left leg moved up and down two or three times, the whole attack lasting a few minutes.

2. Patient suddenly flexed the spine and all four limbs became extended like an angry cat. Respiration became rapid for a few seconds and then the attack ceased. Corneal reflexes not lost.

3. Sudden clonic movement of all limbs lasting a few seconds. The arms were extended, head retracted and face only slightly involved. No loss of corneal reflexes. During the attack the patient appeared frightened and attempted to get hold of the bed- Hag with his right hand, uttering a few short cries.


Progress.—He slowly wasted and the head began to look relatively large in comparison with the wasted body. There seemed to be no evidence of any useful vision and he did not blink to a menace or reach out for any object. Four and a half months after admission the cherry-red spot was still visible. Two or three minor seizures still occurred daily. All the limbs were very spastic, the arms remaining in flexion at the elbows and wrists, and the legs in extension. The abdominal reflexes were absent and the plantars flexor. By thirteen months after admission, the patient was merely a mass of skin and bone, still taking liquid nourishment and vomiting occasionally. The limbs were now rigid in the decerebrate position. Rotation of the head to the left caused slight flexion of the left arm and extension of the right arm, and
vice-versa. No voluntary movements of the limbs occurred. The knee and ankle jerks were just present, the plantars extensor and the patient reacted feebly to pin-prick on both sides.

He died on February 18, 1936, at the age of three years and ten months.

Autopsy findings.—Marked shrinkage and firmness of cerebellum and considerable hydrocephalus en vacuo, the inner nuclei of the thalamus projecting into the third ventricle as large knuckles. Optic nerves thin and very firm. Posterior half of eyes removed. They showed a dark brownish-red macular spot surrounded by a greenish zone (formalin injected into vitreous immediately after death). A widely patent foramen ovale was present in the heart.

Liver normal (wt. = 285 gms.).
Spleen small (wt. = 20 gms.).
Kidneys normal (wt. = 40 gms.).
Rest of body very thin, but normal.

The microscopical findings were exactly similar to those in Greenfield and Nevin's case and will not be repeated here.

The Ha. Family

No Jewish blood or consanguinity. The parents lived in Watford, the father being of a mixture of Irish and Welsh blood. The mother is English and an epileptic (see below, Case 8).

There were three children of the marriage, first, a boy aged eighteen, alive and well, and examination showed no abnormality in the eyes or nervous system. The second, a girl, born two years later died at eleven months in January, 1927, and was apparently suffering from infantile amaurotic idiocy (case 9) and the third child born at an interval of four years, is also a girl and affected with the disease (case 10).

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Case 8. D. Ha. Female, aged 40 years. The details of this case are scanty. No satisfactory history is obtainable, but the patient was apparently quite normal until the age of thirty-one years, when she suddenly began to experience generalised epileptic attacks which have continued. From this time progressive
mental deterioration set in and the patient has become violent, necessitating restraint in the Brentwood Mental Hospital. In the last six years or so there has been a progressive failure of vision. At examination recently the only abnormalities to be found were bilateral optic atrophy of moderate degree and bilateral extensor plantar responses. Owing to inco-operation it was impossible to make as complete an examination as desired.

It was considered in view of the family history that this was a case of "infantile" amaurotic idiocy occurring in adult life.

**Case 9.** J. Ha. Female, born January, 1926. Normal pregnancy and delivery. She developed normally until about four months of age when she began to appear dull and apathetic and to be easily startled by noises. She did not raise her head and took no notice of anything. She rapidly became blind and wasted away and was admitted to the Archway Hospital, where examination of the eyes disclosed a cherry-red spot at both maculae. The patient died in January, 1927.

**Case 10.** D. Ha. Female, aged 9 years 9 months. Admitted to the National Hospital, August 29, 1941, under the care of Dr. Gordon Holmes.

**History.**—Normal pregnancy and delivery. Bronchitis, whooping cough, measles and broncho-pneumonia in childhood. She did not walk or talk till the age of three years and at the age of fifteen months was found to be suffering from rickets. After an attack of measles three years before admission, she was seized with severe tingling in the hands and feet, which became stiff. These attacks had occasionally occurred during the night, and they would last five minutes and gradually cease. They were apparently typical spasms of tetany. Several of these attacks lasted considerably longer. In addition since infancy she had suffered from recurrent attacks of deafness lasting for periods up to three weeks and associated with nasal discharge. She was observed during one of these and evidence of middle ear deafness was found which rapidly cleared up on treatment of the nasal condition. At the age of eight years poor vision was discovered at school during routine examination, and the condition had remained stationary since that time. Owing to the deafness and poor vision she had got on slowly at school.

**Examination.**—The patient was short for her age being four feet in height. She was somewhat precocious, but well behaved, taking an intelligent interest in events around her and cooperating excellently. She was backward in solving mental arithmetic problems, but did these easily on paper. The bridge of the nose was rather flat and wide and the forehead bulged prominently with a central boss in its upper part. The teeth were excellent.
Apart from shortness of stature the skeleton was normal. She spoke in a nasal voice suggestive of the presence of adenoids. The only other abnormality was in the retinae, the macular area being occupied by a lightish area which gave a good foveal reflex moving in the ordinary way on movement of the ophthalmoscope, and

![Fig. 2.](image)

'Typical cherry-red spot at the macula in a girl aged 10 years. (Case 10).

surrounded by a plum-coloured ring, which was again surrounded by a large white circular area extending out into the retina for about one and a half disc diameters (see Fig. 2). The discs were normal (Mr. Williamson-Noble). The visual fields showed a relative central scotoma in both eyes. Visual acuity of the right eye was 6/18 and that of the left was 6/24, unimproved by lenses.

Investigations.—C.S.F. was completely normal. W.R. in C.S.F. and blood was negative. Blood phosphorus was 2.35 mgms. per cent. and the blood calcium was 10.3 mgms. per cent.

A piece of sternal and marrow bone was trephined and this showed a general hyperplasia particularly of the erythro-blastic
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tissue. There was no maturational defect and no evidence of lipoidosis. A blood count showed 5,100,000 red cells, haemoglobin 100 per cent.; colour index 1.0. There were no abnormal cells present. The white cells were completely normal. Total number 7,700, polymorphs 63 per cent.; lymphocytes 25 per cent. No abnormal cells were present.

The blood cholesterol curve was calculated by administering 5 gms. of cholesterol in olive oil and removing blood before and after 4, 8 and 24 hours. This gave the following results: Fasting level 169 mgms. per cent.; at 4 hrs. 230 mgms. per cent.; at 8 hrs. 189 mgms. per cent. and at 24 hrs. 210 mgms. per cent.

An analysis of the lipoids in whole (oxalated) blood was performed and this gave the following:

Total fat (neutral, fatty acids and cholesterol) = 805 mgms. per cent. (Bloor's method).

Total cholesterol = 192 mgms. per cent.

The former value is in excess of the highest normal value as given by Peters and Van Slyke (Quantitative Clinical Chemistry, 1931). The value for the cholesterol is high, but not outside normal limits.

No doubly refracting cholesterol crystals were present in the urine.

X-rays of the whole skeleton and chest were normal. Splenic puncture showed no abnormal cells to be present.

These results show that some generalised abnormality in the lipoid metabolism was present in this case.

Progress.—She was followed in the Out-Patient Department and on examination in April, 1942, the visual acuity was now 6/24 in both eyes. In the fundi, the white area round the macula was considerably larger, reaching as far as the disc and being about two disc diameters wide. The cherry-red spot at the macula had disappeared and was now replaced by a dead-white reticulated area. The disc was still of good colour.

Juvenile Type

In 1903, F. E. Batten drew attention to the occurrence of "cerebral degeneration with symmetrical changes in the maculae in two members of a family." One patient developed symptoms at the age of six and his sister at the age of five. The retinæ showed generalised peppered pigmented changes and at each macula there was an irregular reddish-black spot, the region immediately surrounding the spot "being paler than the rest of the fundus, and more atrophic-looking." Cerebral degeneration was manifest by mental changes, feeble knee jerks, and extensor plantar responses. Shortly afterwards, Mayou and also Vogt,
demonstrated further multiple cases in families, and the latter tried to establish the condition as a variant of Tay-Sachs' disease and advanced histological evidence in support. Spielmeyer later described similar cases in which the fundus appearances resembled retinitis pigmentosa. Sjögren established the condition as an heredo-degeneration of a simple recessive type and showed that there is a great frequency of consanguinity among the parents, but in contrast to Tay-Sachs' disease there is no racial predilection for Jews although cases occasionally occur among them (see later). The patient develops normally until the age of five to eight years and then the first signs of the disease usually manifest themselves by failure of vision, the patient in most cases becoming blind within two years. Later, epileptic attacks occur followed by mental degeneration, disturbance of speech, ataxia, spastic weakness of the legs, and incontinence, all of which progress, the patient finally dying between fourteen and eighteen years of age, but occasionally reaching the age of twenty-five or more (see Cases 21, 23 and 24).

In the retina, fine pigmentedary changes at the macula are probably the first signs. Gradually the whole retina atrophies sometimes with pigmentedary degeneration like that of retinitis pigmentosa, but differing in that the central areas are affected as well as the periphery. These changes are probably always present in advanced cases and were described by Greenfield and Holmes and Sjögren. Confirmation of these changes was found in our Case 16 (see Fig. 5 and 6). Later optic atrophy and narrowing of the vessels appears and irregular nystagmus is almost constant in the terminal stages. Microscopically the degenerative changes in the retina are less extensive in the ganglion cells than in the infantile type and the rod and cone layer, which is little affected in the latter type, is completely destroyed.

In the brain the changes found are essentially those present in the infantile type of the disease. There is, however, a greater variation in the intensity of the degenerative changes than in the infantile type, and the cerebellum, basal ganglia and medulla may be only slightly affected. The staining reactions of the lipid in the cells give different results. In the infantile type the prelipid lecithin-like material is present, whereas in the juvenile type, the lipoids are of a simpler form approaching the constitution of neutral fats. We record here a number of examples of this condition occurring in ten families and affecting fifteen persons (Cases 11-24).

Case 11. Jack Sty -- --, aged 4½ years. Admitted to the National Hospital, March 2, 1926, under the care of Dr. Collier.

Family History.—Mother and Father from the London area, alive and well. No family history of any nervous condition. No
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miscarriages. Two children, one aged 11 alive and well, the second was the patient.

History.—Normal pregnancy and confinement. "Milestones" normal. Well until the age of three years, when he began to have generalised epileptic attacks, which had been as frequent as two a day. In the last six months there had been another type of attack without loss of consciousness in which the patient would go stiff, his arms become contracted and drawn up and his mouth would drop. The attack would last for five minutes or more. For the last six months he had been unable to walk or sit properly and had been gradually going off his feed and during the same time his speech had been deteriorating and he was now only able to say a few words. Occasionally he had inexplicable screaming fits lasting for an hour or so and he also dribbled a lot.

Sudden jumps occurred irregularly often every two to three minutes. His hands had become very shaky, preventing him feeding himself. In addition there was an obvious defect of eyesight, which seemed to vary, and he was unable to see things held before him. It seemed that his head had been getting larger and his face swollen at the sides.

Examination.—The patient was small for his age and very difficult to examine, crying lustily at the slightest provocation and not answering any questions. Spontaneous speech was limited to "Mummy." If observed while sitting up in bed, his head and shoulders were in a state of continual jactitating movements, but no such movements of the limbs were seen. He was doubly incontinent. Visual acuity was difficult to ascertain, but obviously very defective though he could certainly make out objects held up before his face.

Fundi: (Mr. Williamson-Noble). Some pigmentary changes at the maculae. Discs of fair colour. No nystagmus. Tongue protruded jerkily and movement not maintained. All limbs very spastic, both arms being held by the sides with elbows slightly flexed. He grasped objects held before him and put them in his mouth. Intention tremor in both arms. Both legs in attitude of extension with bilateral pes cavus. Some contracture of the Achilles tendons on both sides. Sensation normal. All tendon reflexes very brisk. Abdominal reflexes present, bilateral extensor plantar responses. Slight bulging of the skull, at the side, not definitely hydrocephalic however. The patient had to be discharged from hospital because of the impossibility of controlling him. Further course was not followed.

Case 12. Stanley Ph., aged 8. Admitted to the National Hospital, January 13, 1923, under the care of Dr. Risien Russell.

Family History.—Father and Mother alive and well, London
born. Nine other children died from unknown causes, one brother alive and well aged ten years, two miscarriages since the birth of the patient. No nervous disease in family.

History.—The patient was a full-time child born after a difficult labour. He walked at the age of fourteen months but was late in talking and still talked badly at the age of four years. Went to school at the age of five when his defective vision was first noticed along with twitching of the face and blinking of the eyelid. Then it was noticed that if he dropped an object he did not look for it with his eyes but found it by feeling for it. He got on quite well at school and was only about half a year behind the children of his own age. He became generally nervous, moody, irritable and apathetic but at other times would be quite normal and bright. Since the age of five his vision had been getting gradually worse. He could not see in the dark at all and in daylight stumbled over objects and was apparently unable to see things around him. During the last two years he had suffered from "bilious attacks" with nausea and vomiting and some occipital headache. These usually occurred at about fortnightly intervals. He was quite normal in mental perception and took cognisance of all that was being said and going on around him.

Examination.—A well developed child, rather pale, intelligence practically normal. Co-operated well. Speech normal. Visual acuity (corrected) 6/18 right and 6/24 left. Fields showed concentric contraction on both sides. Fundi: (Mr. Paton). Right: disc waxy-looking, vessels reduced in calibre, the retina all round the disc being atrophic, allowing the choroidal vessels to be seen through it. At the macula the retina was speckled with pigment giving a pepper and salt appearance. The same appearance was present in the peripheral retina and increased near the extreme periphery.

Left: appearances similar, but vessels still more reduced in calibre, no nystagmus. No other organic signs found in the central nervous system.

Further progress not followed.

The Cr. Family

From the London area. Father alive and well aged fifty-one. Mother alive and well aged fifty. No consanguinity. Mother has a brother born instrumentally and apparently injured then. He was always defective mentally but able to earn a livelihood. The children were:

1. Henry, aged 27 years, alive and well.
2. Charles died 1925, aged 17½ years, in the National Hospital from cerebro-macular degeneration. He became affected at the
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age of nine with failing vision, mental impairment and gradually became imbecile, incontinent, blind and paralysed. He was left handed. This case was reported by Greenfield and Holmes in 1925.

3. Frederick, aged 23 years, alive and well.
4. Albert, who died in 1928 at the age of 16 years of cerebro-macular degeneration. Symptoms arose at the age of nine. He died in Darenth Mental Hospital. He was left handed. This case was also reported by Greenfield and Holmes.
5. Doris, aged 17, alive and well.
6. Gertrude (see below, Case 13), suffering from cerebro-macular degeneration.
7. Kenneth, aged 12, alive and well. Left handed.
8. Olive, aged 9 (see below, Case 14).

Case 13. Gertrude C. Female, aged 8. Admitted to the National Hospital, October 9, 1926, under the care of Dr. Gordon Holmes.

Present Illness.—Six months before her right eye started to water and five days before admission she had a generalised epileptic attack. For the last four months “she had not been herself” and had several “little ways” as in her two affected brothers. She had been born with a tumour on one ear which had been removed at Great Ormond Street Hospital where she had also had an operation on the left nostril.

Examination.—A normal looking child, except for a congenital defect of the left nostril, which was slightly cleft in an upward direction. She seemed cheerful, bright and intelligent. Visual acuity 6/12 in both eyes.

Fundi. (Mr. Leslie Paton). Right eye: the macular area showed a very definitely marked fovea with a slightly granular area around it. Left eye: macula, not quite so clearly marked fovea but definitely abnormal (Fig. 3). No other abnormal physical signs.

C.S.F.—Normal, total Pr. 0.02 per cent. W.R. negative.

Progress.—The patient gradually became demented, had numerous epileptiform attacks and died at the age of 15 years.
CASE 14. Olive C. Female, aged 9 years. Admitted to the National Hospital, July 27, 1932, under the care of Dr. Collier.

History.—She was a full term child, delivery normal. She developed normally, walked and talked at the usual ages and got on well at school, but a year before admission she began to lose her usual brightness and this had progressed. Her school-mistress said that "she had gone back three years in the last twelve months." No change in speech had occurred nor had she complained of her vision. One month before admission she had two generalised epileptic attacks.

Examination.—A healthy looking child speaking normally but tending to giggle excessively. Intelligence well below normal. Attention fair but easily distracted. Clean in her habits. Visual acuity 6/9 in both eyes.

Fundus: (Mr. Williamson-Noble). Reflex in the macular region was rather bright. The rest of the central nervous system was normal. Liver and spleen not palpable.

While in hospital the patient had several more attacks. Six months later the fundi were again examined and at the macula there now appeared a peppering of the fundus and the fovea had become more marked. She gradually became demented and died at the age of fifteen and a half years.
The Wi. Family

All their forebears had come from London. Father alive and well, aged 54 years. Mother alive and well, aged 50 years. There were eight children.
1. Charles, aged 28, alive and well, unmarried.
2. Gladys—Case 15.
3. Lily, alive and well, aged 21, unmarried.
5. Jessie, aged 16, alive and well.
7. Dorothy—Case 18.
8. Albert, aged 31, alive and well.
Nil else in family history.

CASE 15. Gladys Wi. Normal pregnancy and delivery. Developed normally until the age of ten years when she began to have progressive failure of vision. One year later she developed generalised epileptic fits and began to deteriorate mentally and to fall about when she walked. Her speech became impossible to understand. Examination at the age of sixteen years revealed pigmentary degeneration at both maculae. She died in a mental hospital at the age of twenty-two years.

CASE 16. Edward Wi. This patient was normal until the age of nine years when he developed symptoms in every way resembling those of his sister, Gladys. He died at the age of seventeen years blind and demented.

CASE 17. John Wi. For the last five years this patient had had progressive visual and mental failure and was completely blind, suffering from numerous epileptic attacks. He was ataxic and his speech grossly abnormal.

CASE 18. Dorothy Wi., aged 8 years. Admitted to the National Hospital, March 7, 1929, under the care of Dr. Risien Russell.

History.—Quite normal at birth, labour normal, breast-fed until nearly one year old. Began to walk and talk soon after she was twelve months of age. She was quite normal till two years before admission when she became nervous and restless and her limbs
began to tremble. For the last six months she had been easily knocked over if anyone bumped into her and also tended to bump into objects. During the last two years she had become less intelligent and could not now do the simplest errands at home. During the last eighteen months her speech had become much affected and her father was now unable to understand a word she said. One year before admission she began to have an abnormal appetite and there had also been some unsteadiness of her gait which prevented her running.

Examination.—Restless, inattentive and paid little attention to anything that was said to her. She spoke in a quite unintelligible manner and often sang a few bars of a well-known tune correctly. When asked to put out her tongue she paid no attention. She did not know her first name but remembered her surname. There was no tendency to weep.

Visual acuity impossible to estimate but she did not look directly at one suggesting that the macular vision was defective. Fundi (Mr. Leslie Paton). Right optic disc pale and atrophic, arteries very much reduced in calibre. Macula showed oval area of pigmentary disturbance but without any of the central ring, characterising most cases of cerebro-macular degeneration. The periphery showed some pepper and salt appearances (Fig. 4). Left eye,
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similar, but pigment disturbance at macula less marked. Plantars extensor. The patient was unsteady and walked on a wide base and liked to have something to steady her. Rest of central nervous system and other systems normal.

The patient's condition remained unchanged while in hospital.

The We. Family

Father alive and well, aged 33 years. Mother alive and well, aged 33 years. Living in London. There were three children.
1. Winifred, developed symptoms at the age of 6.
2. Wilfred, developed symptoms at the age of 5½.
3. Brother, aged 3, alive and well.

CASE 19. Winifred We., aged 9 years. Admitted to the Maida Vale Hospital for Nervous Diseases, February 25, 1931, under the care of Dr. Wilfred Harris.

History.—The patient had had generalised epileptic attacks for the last six months. At the age of six years she developed scarlet fever, followed by otitis media and nephritis. She was in hospital for six months and on her return home she began to scream and was very nervous.

Examination.—A mentally retarded patient with no abnormal physical signs in the central nervous system except that she walked on a broad base. She was discharged to Lingfield Epileptic Colony, where six months later the fundi were examined by Mr. Rupert Scott who reported that the discs were pale, the vessels very small, with pigmentary changes of the right macula and to a lesser extent of the left. Her further course was not followed.

CASE 20. Wilfred We., aged 7 years. Admitted to the National Hospital, August 26, 1931, under the care of Dr. Purdon Martin.

History.—The patient was a full time, breast-fed baby. Normal pregnancy and confinement. Walked and talked at an early age. He attended school from the age of three and a half and made good progress. He developed well mentally until the age of five, but then development apparently ceased. Eight months before admission his mother noticed that he had to peer very closely at objects and apparently had difficulty in seeing them. He had increasing difficulty with his eyesight, especially in the last month,
and began to walk into objects, and had to feel them before he could say what they were. If he wanted to see anything he always looked away from it. In the last year he had become very sleepy, often falling asleep over his tea. Twelve months before admission he became very fat, but since that time he had been steadily losing weight.

*Examination.*—A plump, well-grown boy with obviously defective eyesight, apparently bright and intelligent. His replies to questions, however, were babyish and sometimes could not be understood, especially in spontaneous speech. Could not repeat the alphabet or count beyond two. There was a mole about one half by one quarter of an inch on the left cheek and the skin was very dry and scaly. Visual acuity: finger counting at one foot. Fundus: (Mr. Paton). Right disc pale, arteries contracted. Early pigment change in the region of the macula. Pigment in the periphery somewhat granular, but this was probably due to the light colour of the fundus, and not necessarily pathological (Fig. 5). In the left eye the changes were similar, but slightly less marked.

*FIG. 5.*

Fundus in a juvenile case at the age of 7 years. (Case 20).
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Pupils, moderate size, right slightly larger than left. React rather slowly to light. No other abnormal signs in the central nervous system. C.S.F. normal. Pr. 0·025 gms. per cent.

Progress.—The patient remained in hospital for thirteen months. Three months after admission he complained of aching in his legs when wakened up one morning and was unable to walk, except by clutching the bed end, owing to stiffness of the leg and staggering about. The legs were spastic, the knee jerks increased and the left plantar extensor. These symptoms gradually disappeared. The patient became quieter, rarely spoke and when he did so the speech was indistinct and slow. He smiled fatuously when spoken to, but his activities were limited by his visual disturbance. All the tendon reflexes became very sluggish and his vision rapidly deteriorated. The left plantar response became extensor. He was discharged on September 14, 1932, but was re-admitted on October 6, 1935, having become progressively worse since discharge. Three months before re-admission he began

FIG. 6.

Same case as in Fig. 5 four years later.
to have generalised epileptic attacks at about six-week intervals. Since their onset speech had become so affected that only his mother could understand him. He could now only just distinguish black from white and his mentality had also deteriorated. Fundi: (Mr. Williamson-Noble) now showed on the right: disc pale all over, vessels contracted down to threads, and widespread retinal degeneration with pigmentary change. Left: retinal degeneration of slighter degree with definite pigmentary change at the macula (Fig. 6). He deteriorated gradually and died at the age of eleven years.

CASE 21. J.M. Female, aged 14. Admitted to the National Hospital on August 27, 1941, under the care of Dr. Walshe.

Family History.—An only child of parents born and bred in Portsmouth. No still-births, miscarriages, or consanguinity. No nervous disease in family.

History.—Normal birth and early development. Normal until eight years of age when eyesight first began to deteriorate. At the same time her teachers began to complain of inattention at school and of her losing grip of her work. Since then her mental powers have deteriorated. Since the age of ten to eleven she had been unstable, abnormally talkative and restless. Eight months before admission she began to have generalised epileptic fits.

Examination.—A well-built girl, almost blind and showing some mental deterioration. Fundi: (Mr. Williamson-Noble) right macular area pink with yellowish-white central portion. Temporal pallor of disc. Arteries contracted. General retinal atrophy shown mainly by presence of minute dots of pigment. Left disc rather better colour, but other changes similar. C.S.F. normal. W.R. negative. Discharged unchanged.

CASE 22. Donald Sta--, aged 9 years. Admitted to the National Hospital on June 15, 1939, under the care of Dr. Gordon Holmes.

Family History.—Father alive and well. Mother suffering from melancholia and hysterical paralysis of the leg for the last five years. Patient was the only child. No family history of nervous or mental illness. Family lived in London.

History.—Full-term child, normal development, “milestones” normal. Very bright until nine months before admission, when, following a fall from a moving bus, he seemed dazed for two hours, but did not lose consciousness and he recovered completely. One month later his mother noticed that he was becoming “confused.” He went to wrong classrooms, wrote geography lessons in history books and wrote “slantwise on paper instead of in straight lines.” The words he wrote were often illegible, due to writing one letter over and over again, but he could spell aloud
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quite correctly. He began to stumble while walking, apparently due to inability to see clearly and did not seem to be able to focus his eyes well. He would place a glass of water too near the edge of the table or slide his foot along in order to find the edge of a stair. He became very preoccupied, staring into space and ignoring commands. His mother also thought that he had become childish. He liked to be waited on and fed. During the last ten weeks his condition had grown worse and his legs had become unsteady and inco-ordinated, preventing walking, and speech very slow and irregular. His hand motion had become awkward and clumsy. He no longer read or wrote and ground his teeth while asleep.

Examination.—A thin child, vacant expression, mental state definitely retarded. At times failed to understand even the simplest commands, while at others seemed to understand quite normally. His replies were childish and he was very inattentive and confused. Orientation was impaired as regards time and place. Speech slow and irregular. Dysarthric, and he tried to overcome this by speaking slowly, the words being broken and slurred. He could name objects shown to him if his attention could be retained. Visual acuity difficult to test owing to inattention, but he could name the number of fingers across the room. He did not, however, look straight at the object and could not find small objects dropped on the floor. Fundi: (Mr. Williamson-Noble) macular region more conspicuous than usual with a little abnormal pigmentation. Pupils large, react normally. No nystagmus. Motor system: titubation of head which swayed about aimlessly and then was brought stationary when his gaze was fixed. Hypotonia of all limbs with deviation of outstretched hands and considerable inco-ordination with aimless swaying about of limbs. Dysdiadochokinesis. All tendon reflexes brisk and equal. Extensor plantar responses. Sensation normal. Liver and spleen not palpable. C.S.F. Pr. 160 gms. per cent. Patient remained unchanged while in hospital.

Case 23. George St---, aged 23. Admitted to the National Hospital, January 7, 1929, under the care of Dr. Collier.

Family History.—Father and Mother alive and well. The patient was the eldest of five siblings and he was born in Sheffield. The four younger children were a girl aged 19, a boy aged 15, and
two girls aged 5 and 2 years, all being alive and well. They had migrated to the Botley district of Hants. when the patient was 12 years of age. No further family history was obtainable.

History.—At the age of nine years the patient had an attack of "influenza" and soon after this he began to notice a weakness of the legs and his second toes began to turn under the soles. Twelve months before admission the weakness became more marked and he had to give up work. He was unable to stand still and had to use a stick to walk and would sometimes fall. At this time the legs began to shake, especially with the exertion of preparing for bed, and they would gradually quieten down when he got into bed. At the same time as he first noticed the weakness of the legs his arms began to shake making it difficult to lift a cup of tea, and about a year before admission it became difficult for him to work as a farm labourer for this reason. The shaking was worse on exertion. At the time of the onset also he began to have speech difficulty, stuttering to begin with and "then getting it out" suddenly. No sphincter disturbance or fits.

Examination.—A tall big-boned man. Severe degree of dementia. Co-operation only fair. Visual acuity 6/9 right and left. Fundi: (Mr. Paton) at the macula was a semicircle of small black dots, six in number, and in various parts of the periphery were similar retinal disturbances with possibly underlying choroidal disturbances, but they were mainly retinal, the discs being normal (Fig. 7). Pupils, medium size, reacted normally. No nystagmus. Left handed. Spastic gait, stooping posture. Slight general weakness of both arms with some increase in tone. Well marked tremor of both arms. Involuntary movement with fine tremor of outstretched fingers. Slight wasting of the small muscles of the hand with a little intention tremor and involuntary movement. Legs showed a spastic paraplegia of severe degree, which greatly interfered with co-ordinated movements. Considerable weakness of extensor groups. Coarse rhythmic tremors were present at wrist increased by voluntary movements. Fibrillar twitchings of all muscles of the lower limbs and glutei were present with some wasting of the muscles below the knee. There was a bilateral equino varus deformity, the great toes being dorsiflexed at the proximal joint and plantar flexed at the distal joint, the second toes being plantar flexed. Sensation normal. All the tendon jerks were extremely brisk. The abdominal reflexes were present and the plantar responses extensor. Gait extremely spastic and walking impossible without aid. Spleen and liver not palpable. To rough psychometric test, the mental age was seven years. C.S.F. normal. Total Pr. 0.03 per cent. Patient improved slightly while in hospital.
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FIG. 7.

Fundus in Case 23 at the age of 23 years, a juvenile case surviving longer than usual.

CASE 24. John Pi---, aged 25 years, born in Eastbourne. Admitted to the National Hospital, December 16, 1931, under the care of Dr. Walshe.

Family History.—Father and Mother alive and well. Patient was the eldest son, three other children being alive and well. He had been married for three years but his wife had had no pregnancies for unknown reasons.

History.—When at school his sight was found to be defective at the age of seven years by the school doctor, but he had no treatment. He himself noticed no disability except that he was
unable to play cricket, as he could not see the ball. At the age of seventeen he saw an ophthalmologist, who told him that pigmentary disturbance of the retina was present and that his sight was only half the normal. Three years before admission his vision further deteriorated and he was now unable to see things on the table, when working as a waiter. Reading had become very difficult, his eyes rapidly tiring. He was only able to read by very strong artificial light and then only by standing under it. In the last two years he had put on nearly two stone in weight.

**Examination.**—A well-developed man, attentive and co-operative, well orientated, memory excellent, intelligent. Visual acuity 6/36 right and 2/60 left. Large central scotoma in both eyes. Fundi: (Mr. Paton). Right: in the periphery there was a typical "bone-corpuscle" type of degeneration revealing the choroidal vessels. The macula showed small brownish dots the size of a pin's head scattered about. At the macula was a circular whitish area about three to four millimetres in diameter like a hole at the macula (Fig. 8.). Left: exactly similar, except that the whitish area was smaller and ran into an area downwards and inwards in which

**FIG. 8.**

Another long-surviving juvenile case at the age of 25 years. (Case 24).
there was no pigment. Both discs were white and the vessels much reduced in calibre. Fields, considerable loss in upper quadrant and whole central area. Pupils, moderate size, react normally. All deep reflexes very brisk. Flexor plantar responses. Liver and spleen not palpable. C.S.F. normal. Pr. 0.055 per cent.

Progress.—Certain subtle mental changes were present, for example, a certain facile contentment combined with a rather disproportionate emotional reaction. He was discharged unchanged.

(To be continued)

COLOUR IN PROTECTIVE NIGHT LIGHT

BY

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In the selection of a colour for a protective night light the eye factors as well as power to penetrate the external atmosphere should be taken into consideration—namely, the comparative sensitivity of the eye to coloured lights at very low intensities, visual acuity at low intensity, and the adaptation factors. Principally because of the eye factors deep red has recently been recommended as superior to the formerly widely-accepted dark blue light. The question is intricate and interesting and in its broader aspects is still an open one. It is extremely interesting in its relation to the visual functions.

Deep red light would hardly be acceptable as a night light for the illumination of many interiors, for example the lighting of hospitals. As a night light for hospitals we have used with considerable satisfaction light approximating the colour of a low-brightness kerosene flame. This is not too unpleasant and is distinctly somnolent. The possibility of using heavily frosted, colourless bulbs should not of course be ignored. In fact we believe that a comparison of test results will show in their favour. However, without the use of colour it is difficult to get light of sufficiently low intensity with the lamps that are available.

As to the suitability of blue as a colour for night-time protection, it should be kept in mind that dark blue is the least distinguishable from dark grey, and black of any of the shades of colour, and that the illumination of an interior with blue light by a suitable regulation of intensity and hue can be given a dim moonlight effect which differs little in colour from the exterior illumination on a moonlight night. Further, even if the brightness of the colours is