

## Clinical Department.

### AMAUROTIC FAMILY IDIOCY: A REPORT OF FOUR CASES.\*

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THIS rare disease of infancy was first described by Warren Tay at a meeting of the Ophthalmological Society of the United Kingdom in 1881. Since that time quite a number of cases have been seen by various observers, but so far as I have been able to ascertain, but one case has been reported as occurring in this vicinity; a case reported by Dr. Wadsworth at the meeting of the American Ophthalmological Society in 1887, as a "case of congenital zonular grayish-white opacity around the fovea"; four cases, three at the Infants' Hospital, and one at the Massachusetts Charitable Eye and Ear Infirmary, have come under my observation.

**CASE I.** Infants' Hospital, January, 1897. Male, aged fourteen months, of Jewish parents. Family history, good; two brothers and two sisters living and well, one child died of brain trouble at age of fourteen months. Previous history: Child born apparently normal and continued well for four or five months, then mother noticed that it appeared different from the other children, did not seem to gain in strength, was unable to sit up or hold the head up, the arms and legs were moved about in an aimless manner. At times the child cried stupidly, did not appear to see well. One month before entrance to the hospital the baby began to have convulsions. Physical examination: Well developed and nourished, stupid, unable to sit up, cannot hold head erect, extremities flabby and show marked loss of power. Heart and lungs negative. Ears negative. Knee jerks normal.

Examination of eyes reveal the characteristic grayish-white patch, in the macula region, in the center of which is a sharply defined red spot. Optic atrophy. This child was in the hospital at three different times and the prominent features were twitching of face, lids, corners of mouth, numerous convulsions often starting with a sigh and followed by laughter; the records state that the child "smacks its lips" a great deal.

Progressive loss of flesh and strength, death at the age of twenty-two months. No autopsy.

**CASE II.** Massachusetts Eye and Ear Infirmary. January, 1902. Service of Dr. Wadsworth. Male, aged fourteen months; first child, of Jewish parents.

This case was seen in the Out-Patient Department, hence no full report can be given. The eyes showed the characteristic change in the macula region, with optic atrophy.

This child died at the age of two years. No autopsy.

**CASE III.** Infants' Hospital, June, 1905. Female, age nineteen months, born in this country of Jewish parents. Family history, good; first child, no miscarriages, no children in either family that died in infancy of any brain trouble. Previous history: Breast fed for fourteen months; child apparently normal until three months of age, when mother consulted physician, but was told baby was all right. The child has never tried to walk or stand, is unable to sit up without falling over, cannot hold head up, never reaches for things or tries to play; very sensitive

and dislikes to be handled; rolls eyes about, but mother thinks child can see. Three weeks ago child began to have twitching of face and convulsions, since then has not taken food well. Always constipated. Physical examination: Well developed and nourished. Fair color; skin in good condition except on feet and buttocks; feet puffy and purple; circulation poor. No signs of rickets. No rigidity of head or neck. Spine flaccid; no spasm or paralysis of arms or legs. Knee jerks equal and not lively. No Kernig's sign; condition of stupor. Lungs normal except for few fine, moist râles at base. Heart, liver and spleen apparently normal. Lumbar puncture negative. Urine pale; 1,014; sl. acid; no albumin. Examination of eyes: Pupils equal and react to light, but child does not seem to follow light; eyes wander aimlessly. Myopia, myopic crescent about disk; optic atrophy; characteristic changes in macula region of each eye.

This case was under observation for about six weeks and, aside from the convulsions, the things noted were difficulty in feeding (the child had to be fed with a tube) and the hypersensitiveness to noise.

**CASE IV.** Infants' Hospital. November, 1905. Joseph C., aged eighteen months, born of Jewish parents. First child. Family history negative. Previous history: Breast fed eight and one-half months. Child got along all right to the third or fourth month, then seemed to develop slowly; at eight months of age was not considered bright mentally. Has never sat up or attempted to stand or walk. The parents were unable to tell whether child could see or not. For past three weeks there has been a marked twitching of the face.

Physical examination: Under developed and nourished; very flabby skin; fair color; slight cyanosis of lips. Unable to hold up head. Fontanelle  $\frac{3}{4}$  cm. but not tense, rather depressed. Cervical glands enlarged; no sign of rickets; incisor teeth, four upper and two lower. No evident paralysis of face or extremities; legs kept extended, resist attempt at flexing, but after rigidity is overcome, very flaccid. Unable to make out patella reflex. No ankle clonus. Heart, lungs, liver and spleen negative. Urine turbid; acid; no albumin; no sugar; 1,030. Lumbar puncture negative.

Examination of eyes reveals grayish-white patch, with brownish-red center, in the macula region of each eye. Optic atrophy. Pupils large and react. While in the hospital this child had to be fed with a tube.

The cause of this disease is unknown; the parents are generally healthy, without specific or tubercular taint, or history of intermarriage. With few exceptions, the cases reported have been of Jewish parentage. Hirsch has suggested that it may be due to a toxic condition produced by the mother's milk. Clairborne reports a case in which autopsy showed a tubercular tumor of the corpora quadrigemina about the size of a hickory nut, and Kuh reports a case in which there was hydrocephalus.

The disease often affects several members of the same family and it usually terminates fatally at about the end of the second year. Sex has no bearing. Infants are born apparently in good health and develop normally to the third or fifth month, then the muscles become weak and flabby, the baby cannot sit up or hold up head;

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<sup>1</sup>Case presented at meeting of New England Ophth. Society, Nov. 14, 1905.

the mental development is arrested, the child becomes dull and stupid, and this condition goes on to idiocy. Twitching of the face and extremities, together with convulsions, are common symptoms. There may be difficulty of feeding. The reflexes may be increased or not, in some cases absent. The disturbance in the eye is not congenital, but comes on with the general symptoms, when it is noticed that the child does not see well, or that it rolls the eyes about aimlessly. The first thing observed in the fundus is a haziness in the macula region, this develops into the typical grayish patch with a cherry-red spot in the center. This is followed by optic atrophy. Nystagmus has been observed.

For a number of years after Tay's first case, attention was confined solely to the ophthalmoscopic appearances, and Sachs was the first to state the fact that we had to deal with an extensive disease of the central nervous system. Sachs at first thought that this condition was due to an arrest of development, but he now believes, with other investigators, that the condition is a true degenerative one.

As a result of an autopsy Peterson concludes that the brain shows a defective development; his case showed a confluence of the central with the Sylvian fissure; the pathological conditions are limited to the nerve cells of the cortex and the medulla, which were found deficient in number and development.

Sachs found the convolutions small, a confluence of central with the fissure of Sylvius; thickening of arachnoid and pia mater, hardness of cortex. Degenerative changes were present in the cortex, in the cranial nerve nuclei, and in the anterior and posterior gray matter from the cervical to the lowest lumbar and sacral segments of the cord, the greatest change taking place in the larger ganglion cells. From this Sachs concludes that the disease affects to a great extent the entire gray matter of the brain and spinal cord.

Kingdon found a marked descending degeneration of the upper part of the cord.

Treacher Collins examined two cases of Kingdon's and he reported edema of the retina, but his examination was unsatisfactory, as the retina about the macula region was thrown in folds. Mohr found edema of the outer molecular layer, and a degeneration of the outer segments of the rods and cones of the retina.

Ward Holden examined the eyes of Peterson's case, but the results were unsatisfactory, owing to post-mortem changes. Holden also examined the eyes of Hirsch's case. One eye was hardened in Müller's fluid and one in formol. The one hardened in formol showed the gray patch at the macula region as seen during life. He reports no edema of the retina, but finds a marked degeneration of the ganglion cells of the retina, the cells being enlarged and altered in shape. This change in the ganglion cells explains the fundus picture: the oval gray patch occupies exactly that part of the retina where the enlarged and altered ganglion cells are thickest, and at the

fovea centralis, where the ganglion cells are wanting, the choroid shows through, by contrast, as a cherry-red spot. The optic nerve showed degenerative changes.

Shumway and Buchanan have confirmed Holden's views; they report no edema of the retina, but find marked degenerative changes in the ganglion cells.

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## Medical Progress.

### PROGRESS IN GENITO-URINARY SURGERY.

BY F. S. WATSON, M.D., AND PAUL THORNDIKE, M.D.

#### DIAGNOSIS OF AZOÖSPERMIA.

THE author<sup>1</sup> states that spermatozoa continue to be produced by the testicles whether the exits from those organs are occluded or not. He regards this as a really important fact as determining the possibility of operative cure in cases of azoöspERMIA otherwise beyond help. To demonstrate the fact he has punctured the testis with a Pravaz needle in a series of patients who had previously suffered from acute epididymitis.

In 12 cases, punctured seven to twelve years after the acute inflammation, spermatozoa were found in all but 2. In 5 cases at a much later period after the occluding inflammation (twelve to twenty-six years) spermatozoa were found in only one instance.

#### EXPERIMENTAL POLYURIA.

In this paper, Albarran,<sup>2</sup> after showing his excellent results in 64 cases of nephrectomie for renal tuberculosis, contrasts the unreliability of many of the methods in vogue for studying the renal function with those obtained by his own plan. This he calls "experimental polyuria." It consists in studying the elimination of the two kidneys separately under normal conditions and comparing these results with those obtained from a like study under conditions of forced polyuria produced by excessive water drinking. The specimens from each kidney are obtained by ureter catheterization. Albarran believes his results indicate this as the best method for obtaining knowledge of the real working capacity of each kidney.

#### REPAIR OF OPERATIVE INJURIES TO THE URETER.

By experiments on dogs the writers<sup>3</sup> demonstrate that one ureter, injured during an operation, may be successfully implanted into the other.

<sup>1</sup> Posner: Berliner Klinische Wochenschrift, xlii, No. 35, Aug. 28, 1905.

<sup>2</sup> Albarran: Presse Médicale, Paris, Nos. 79, 80, Oct. 4-6, 1905.

<sup>3</sup> Bernasconi and Colombino: Annales des Maladies des Organes Genito-Urinaires, Paris, xxiii, no. 13.