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# A CASE OF AMAUROTIC FAMILY IDIOCY WITH AUTOPSY

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presented by the author

# A CASE OF AMAUROTIC FAMILY IDIOCY WITH AUTOPSY.[1]

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This case was brought to my office in November, 1897. It was then an infant three months old, female, child of Russian Hebrew parents. The mother was 28 years old at the time of its birth, and had had five children and one miscarriage. Of the five children, the first was seven years old and normal, the second five and one-half years old (a blind idiot now on Randall's Island), the third a normal child of four years, the fourth a blind idiot, which died at the age of ten months, and the fifth is the case of amaurotic idiocy described in this paper. Thus there were three cases in this family. In the second child the blindness was not noted until the age of six months. In the fourth and fifth cases it was not noted until the infants were four weeks old. The blindness, however, may have existed at birth. My patient was sent to Randall's Island in January, 1898, and died there, March 16th, at the age of 7 months and 20 days.

The following history is from notes taken by myself and my internes at the Randall's Island Hospital for Idiots, Drs. Elizabeth Sturgis and F. O'Neil:

L. L., 7 months, 20 days.

Family History. — Mother, 28 years, German, healthy. Father, 29 years, Russian, said to have some lung trouble causing dyspnœa on exertion, but which is not consumption. History negative on both sides for syphilis, tuberculosis, insanity, epilepsy or nervous disease, as far as was known. No relationship exists between parents.

Personal History.—Born at term, labor normal; nursed four weeks and was then fed on milk and water, equal parts.

[1] Read before the American Neurological Association, May 27th, 1898.

Up to the time of admission, mother declared her to be as bright as her other children, laughing and playing with her hands, but crying much both night and day. Did not follow objects with her eyes, but pressed her hands into her eyes to a certain extent.

Examination showed a well-nourished baby, plump and of good muscular development.

Skin.—A nævus under chin and pigmented nævus on left thigh.

Eyes.—Media clear; pupils somewhat dilated, equally; a rotatory up and down movement, but no fine tremor; conjunctivæ, good color; owing to extreme restlessness of child, the fundus could not be seen. The eyes were examined by Dr. Percy Fridenberg.

Lungs, heart, liver and spleen normal.

Reflexes not increased, no rigidity, no paralysis.

Head.—Antero-posterior circumference, 15⅝ inches.

Chest.—Circumference, 16¼ inches.

Length (of child).—25¼ inches.

Child doing very well, but losing weight until Feb. 10th, then exposed to measles. Stools became green and watery; food was refused or vomited; temperature was raised, 101 to 102 deg. F.

Feb. 14th, coryza and distinct eruption seen on tonsils and throat.

Feb. 16th: Temperature, 104.

Feb. 17th: Sent to measles ward; no distinct rash ever seen on body. Gastrointestinal symptoms continued while in quarantine, refused food. About February 27th, rigidity of neck and knees, some twitchings of muscles. Heart and lungs negative.

March 9th, returned to Infant's Hospital. Became weaker, did not take much nourishment, and just before death she developed what was diagnosed as purpura.

Unfortunately the autopsy on this case was delayed fully forty hours, so that, although it was cold weather, and the body was well preserved, the examination of the finer nerve structures was to some extent interfered with. The autopsy was performed by Dr. D. Hunter McAlpin, Jr., to whose great courtesy I am indebted for the following notes, and also for the brain, spinal cord and parts of other organs used later for microscopical investigation.

Autopsy.—Body is that of a very emaciated child. Post-mortem discoloration present over abdomen and back. There is a large number of small hemorrhage spots of a purplish color in the skin covering the abdomen. There is a dark brown pigmented spot ¾ in. in length by ½ in. in width on anterior surface lower third thigh, evidently a birth mark.

The skull-cap is of usual thickness.

The surface of the brain is markedly œdematous and is congested. The brain weighs 22 ounces. Placed at once in 4% solution of formalin for further examination. The pia mater of the spinal cord is congested. The cord is quite firm throughout

The peritoneum is smooth and glistening. The intestines are distended with gas. The vermiform appendix is 1¼ inches in length, its lumen is patent. The mesenteric glands are not enlarged.

The pleura covering the upper and middle lobes is smooth; adherent to the costal and diaphragmatic surfaces of the pleura. The right lung is also adherent to the diaphragm.

The pleura over the anterior margin and in the interlobar fissure of the left lung is opaque. Apex of the lung shows a large emphysematous bleb. On section the upper lobe is of a pale pink color. On pressure a frothy straw colored fluid exudes. The pleura over the entire lower lobe is opaque and lustreless. Color of lower lobe is dark, mottled with few light areas. There are small nodules felt throughout the lower lobe. On section nodules are found firm, elevated and finely granular. On pressure a small amount of mucus exudes.

The pleura covering the upper and middle lobes is smooth; but over the lower lobe, posteriorly and inferiorly, it has lost its smooth and glistening appearance.

Upper lobe is light pink in color, and exudes a frothy fluid. At apex a mucopurulent material can be expressed from the bronchi. In the lower lobe there are few areas of consolidation similar to those in the left lung.

The pericardium is smooth and free from fluid.

The heart is of usual size. The right auricle and ventricle contain dark clots. Left auricle and ventricle are empty. Aortic valve normal. Endocardium pale and opaque. Heart muscle, firm. Color, pale pink. The other valves are normal. Right cavities dilated. Very small amount of subpericardial fat. The foramen ovale is closed by a membranous curtain, which is not adherent to the septum at its upper portion, so that a probe can be passed between the two auricles.

The kidneys are pale in color. The capsules strip off leaving smooth surfaces. The cortical portions have an opaque appearance. The markings are coarse.

The liver is normal in size. Borders shelving. Capsule smooth On section liver tissue is dry. Lobules distinct. Other zones pale gray in color, blood vessel wall slightly thickened.

Spleen small. Capsule smooth. On section dark plum color. Glomeruli prominent. Consistency firm.

Left suprarenal, pale yellow color. Firm. Small cavity in centre. Right suprarenal, same.

Pancreas, negative.

Stomach, normal size. Contains a few milk curds and dark brownish material. Mucous membrane, thin, pale. Rugæ, obliterated. Intestines, distended with gas. Contain yellowish fluid material.

Two and a half feet above the iliocæcal valve is a Meckel's diverticulum measuring ¾ inches in length. It arises near the mesentery and has a small mesenteric attachment of its own.

Mucous membrane of small intestine apparently normal. Large intestine contains yellowish material. The solitary follicles are prominent.

Bladder, empty, mucous membrane normal.

Uterus, ovaries and Fallopian tubes appear normal.

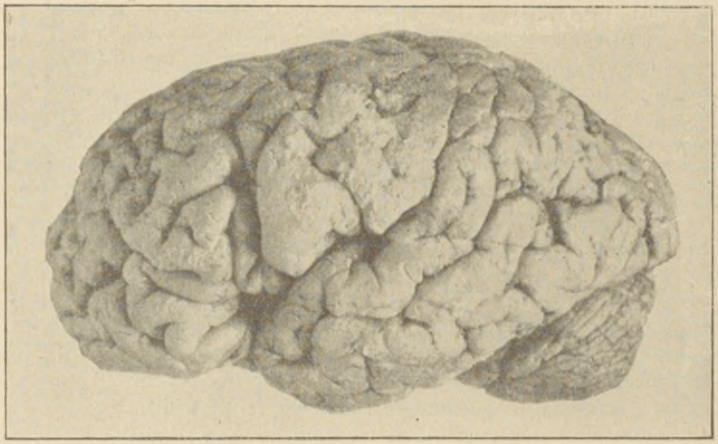

FIG. I.

Brain of amaurotic idiot. Exposure of insula; hypertrophic gyrus; confluence of central and Sylvian fissures.

Anatomical findings: Œdema and congestion of the brain, broncho-pneumonia, pleurisy, acute parenchymatous nephritis.

Macroscopic Examination of the Brain.—Grossly examined, this brain shows simply a few morphological features characteristic of defective development. On the lateral aspect (Fig. I.) we observe confluence of the central with the Sylvian fissure, exposure of the insula and one or two atrophic gyri. On the superior surface there is little to remark upon except unusual asymmetry (Fig. II.).

Microscopical Examination.—The microscopical investigation was carried out at the laboratory of the College of Physicians and Surgeons by Dr. James Ewing and the author, and the results are as follows:

The cortical areas about the calcarine fissures seem to be uniformly deficient in cells, and in some segments the number of cells appears very distinctly reduced. A striking feature of this portion of the cortex is the minute size of the cells, very few distinct somatochromes being seen except in the innermost layer. The separation of the cells into layers in this region is much less definite than is normal. The cells are moderately deficient in chromatic substance. There appears to be no difference in the structure of the right and left side in this region.

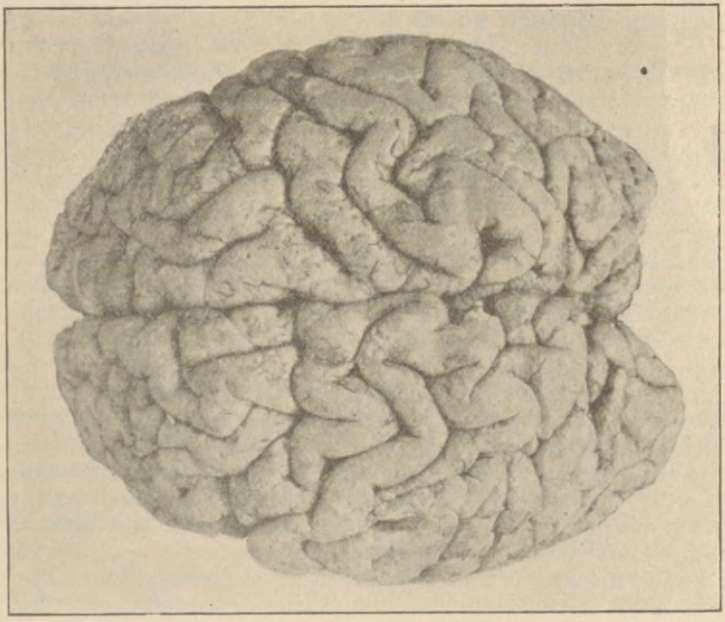

FIG. II.
Brain of amaurotic idiot.

In the motor areas the cells are markedly deficient in number, especially in the second and third layers, irregular in size, uneven in distribution, and uniformly deficient in chromatic substance. The abundance of large nuclei without demonstrable cell body in all layers of this and other regions suggests either deficient development or permanent atrophy.

In the hypertrophic lobule described in the left motor cortex the cells are markedly deficient in number.

None of the large stichochromes ordinarily seen in the motor cortex were anywhere found, but in some segments

rather large cells, forty to fifty microns in diameter were present, of nearly homogeneous appearance.

Throughout the frontal cortex, less distinctly in the temporo-sphenoidal lobe, the cells showed the same deficiency in number, and irregularity in shape and distribution, noted in the other regions.

Lumbar Cord.—Nearly all of the cells are quite normal. There are a few artificial changes, consisting in marked shrinkage of cell bodies and rupture of processes. Some cells show postmortem clouding of cell body and nucleus, and irregularity of chromatic masses. A very few cells show nearly complete absence of chromatic bodies.

Cervical Cord.—The anterior horn cells appear normal. A distinct group of medium-sized cells, situated in the posterior and external segment of the anterior horn shows typical axonal degeneration. Many posterior internal cells are moderately deficient in chromatic substance.

Nucleus XII.—The cells seem deficient in number, but show no distinct pathological alteration of the chromatic substance.

Nucleus X. (Superficial) and Nucleus Ambiguus.—The cells show moderate but uniform diminution in size of the chromatic bodies. In some cells the chromatic bodies are almost entirely absent, a few traces remaining at the periphery of the cell body only.

The limits of the third and fourth nuclei are indistinct. The cells of this region are deficient in number. Many appear shrunken. None of them contain well-formed chromatic bodies, but nucleus and cell body are homogeneous, diffusely stained, and contain few scattered granules of chromatic substance.

The olives are very well developed and cells are very abundant. The cells appear normal.

Purkinje's cells show only postmortem changes.

Corpora Quadrigemina.—The cells of this region are very deficient in number, usually of small size, ten to fifteen microns in diameter, belonging to Nissl's class of arkyochromes, although a few appear to lack distinct cell body, being classed therefore as karyochromes. A few cells also measure 30 to 35 microns in diameter. There are no distinct evidences of recent chromatolysis or of chronic atrophy. The tissue contains an unusually large number of arterioles and capillaries. In the deeper portion of the posterior corpus quadrigeminum are several groups of two to four large cells, 40 to 70 microns in diameter, circular on section, and closely resembling the cells of the posterior spinal ganglia. These cells contain an abundance of circularly arranged chromophilic bodies, and some of them show moderate central chromatolysis.

Internal Geniculate Bodies.—These nuclei contain a moder-

ate number of somatochrome cells, about 20 to 25 microns in diameter, and rather fewer karyochromes, both uniformly distributed throughout the tissue. The larger cells are arkyochrome in type and do not show any recent or old pathological changes.

External Geniculate Bodies.—These bodies exhibit seven distinct layers of cells. There is first a narrow, superficial layer of small karyochromes with distinct cell body. The numbers of these cells vary considerably at different points. Beneath them, increasing in breadth, as one descends are six, dividing at times into nine, layers of larger arkyochromes, 30 to 40 microns in diameter, and among which are also a few karyochromes.

No pathological changes could be detected in any of these cells, other than deficiency in number. The arterioles and capillaries of this region are very numerous.

Sympathetic System.—In some of the sympathetic ganglia, lying on the aorta, near the cœliac axis, the cells are present in normal numbers and show various grades of central chromatolysis.

Nerve Trunks and Fibre Tracts.—For the study of fibres and tracts, sections were made of the occipital cortex of either side, the geniculate bodies, corpora quadrigemina, chiasm and optic nerves, at two points in the medulla, and in the cervical and lumbar cord. These sections were stained by Van Gieson's, Marchi's, and Pal's methods, but owing to the original fixative used, formalin, satisfactory results were obtained only from the first of these methods. In sections stained by picro-acid fuchsine, no distinct abnormalities were seen in any region of the central nervous system. The various tracts in the cord and medulla seemed to be normal in development, and the axis cylinders and myelin sheaths were intact. The optic nerves were not distinctly deficient in size or number of fibres. The optic chiasm and radiations were apparently normal.

It seemed probable, on comparison with sections of normal brains, that the development of fibres in the cerebral convolutions was moderately deficient, especially in the occipital region, but this condition was far less evident than the great deficiency of cells noted in these regions.

On comparison of the various sections of the cortex and cerebral ganglia in the present case with sections from the same region in normal infants, the abnormalities described became strikingly apparent. In the normal infant's brain the cells are arranged in very distinct vertical columns, as well as in longitudinal rows which in sections stained by methylene blue are plainly visible to the naked eye. In the present case nearly all traces of the vertical columns were missing, and the

separation into longitudinal rows was very indistinct. Moreover, the cortical cells were very deficient in number as well as in size and content of chromatic substance.

These changes were noted throughout the cerebral cortex, but were specially evident in the optic centres and ganglia. In the absence of demonstrable lesions in fibres and tracts, these cellular abnormalities, together with the increased number of blood vessels previously noted, constitute the main pathological features of the case.

Viscera.—Sections of the kidney, liver, spleen, lung, suprarenal and pancreas, failed to show any noteworthy lesions other than an advanced fatty infiltration of the liver.

Eyes.—The two eyes were removed and immediately placed in formalin. One was given to Dr. Carl Koller for examination, but his report has not yet been made. The other was given to Dr. Ward A. Holden, whose report is as follows:

One eye was received in formol 5 per cent. The retina was found to be detached at the macula, a postmortem change, which prevented the recognition of any existing gross pathological conditions. Sections cut in paraffin and stained by Nissl's method showed advanced postmortem changes: the vessels contained numbers of bacilli, the rods and cones were destroyed, the ganglion cells, bipolar cells, and nuclei of the rods and cones were vacuolated. In most of the ganglion cells the vacuolation had been so excessive that the cell bodies were more or less completely broken down so that nothing can be said as to their size in life. The nuclei took on a faint diffuse stain, and the nucleoli were well marked. Nissl granules were present. An attempt to stain the optic nerve by Weigert's method was not successful after the formol hardening. Hæmatoxylin-eosin preparations of the nerve revealed no pathological changes.

From this unsatisfactory examination of the eyes it cannot be said that pathological changes existed in life. If pathological changes in the ganglion cells existed, however, they were not in an advanced stage.

Conclusions: The brain shows, both macroscopically and microscopically, a condition of defective development, and this corroborates the findings in the several autopsies made in these cases (with the single exception of that of Hirsch, reported at this meeting). The pathological conditions are limited, as far as the fine structures are concerned, to the nerve cells of the cortex and medulla, which were found markedly deficient in number and in develop-

ment in the occipital region about the calcarine fissure, in the temporo-sphenoidal lobes, in the frontal lobes, in the motor areas, in the corpora quadrigemina and geniculate bodies, and in the third and fourth cranial nuclei. Postmortem changes did not affect the importance of these findings. As regards, however, the alteration of the chromatic substance of the cell, this must be referred largely to the general condition of the patient before death, and not to the disease under discussion. No definite changes in the fibres or imperfect developments of the tracts were discovered.