A CASE OF AMAUROTIC FAMILY IDIOCY,

WITH A SUMMARY OF REPORTED CASES.*

MORTIMER FRANK, M.D., B. S. (Mass. Inst. Tech.).
CHICAGO.

This uncommon disease is especially interesting to ophthalmologists because of the fundus changes which are pathognomonic and because Warren Tay, an English oculist, first drew attention to the subject in 1881. From that time until 1896, when Sachs proposed the name as now used, each observer described his case under a different title.

Amaurotic family idiocy is a fatal disease, commencing in the early months of infant life with muscular weakness progressing to almost complete paralysis, associated with a distinct symmetrical fundus appearance and terminating in death about the end of the second year. No definite exciting cause has been assigned. There is no evidence that inherited syphilis, consanguinity of parents or any of the usual diatheses is a factor. There is, however, a marked racial peculiarity. In all but six of the recorded cases the disease has occurred in Jewish children. Both male and female children are affected. There is a suggestive family predisposition in that more than one child in the same family is attacked, but with no regularity in the order in which this takes place.

The child is born at full term, sound and healthy, and continues so until about the third month. It then ceases to develop along normal lines and signs of muscular enfeeblement, especially of the back of the neck, are observed, with failure of sight. Should an ophthalmoscopic examination be made at this time, definite and characteristic changes in and about the macular region will be detected. Marasmus increases slowly, and the muscular enfeeblement becomes so pronounced that the child is unable to sit up and the head falls backward if unsupported. The grasp is feeble and objects placed in the hands are soon dropped. There is a decided cessation of mental development and the child becomes apathetic. Perception of light is present for a time, while hearing remains hyperacute, so much so that any sound causes the child to start.¹ The sense of taste is preserved, and interference with deglutition occasionally happens. Finally, paresis or paralysis, either flaccid or spastic, of the greater part of the body appears. The deep reflexes may be exaggerated or diminished and occasional spasmodic contraction occurs. The temperature is normal throughout the course of the disease. Falkenheim calls attention to an explosive laughter, which could be induced in the reported case by blowing on the child's face. Total blindness finally ensues from optic atrophy. The duration of life varies from one and a half to two and a half years.

The following case was referred to me by Dr. A. C. Cotton, through whose courtesy I am enabled to publish it. Minute details are omitted, as the history will be more fully gone into when published by him:

R. H., female, aged 1 year, was examined March 27, 1905. She is the youngest of four children, the remaining being boys in good health. There is a history of one miscarriage. All the children have been breast fed. The parents are of Jewish extraction, with no history of consanguinity or syphilis. The appearance and nutrition of the child was good and the head was well formed and of normal size. She was listless, unable to sit up and apathetic and understood nothing. The reflexes were exaggerated and especially noticeable was the abnormal susceptibility of the child to noises.

* Read before the Chicago Medical Society.

1. Kingdon observed the changes in the temporo-sphenoidal lobe much less marked than in other regions of the brain, which probably explains the fact that hearing is unaffected.

The following ocular symptoms were noticed: The pupils were about 1.5 mm. in diameter, equal in size and reacted slowly to light, but light thrown into the eyes failed to give any evidence of vision. A drop of atropin solution was put into each eye to facilitate the examination. The discs were atrophic, grayish in color, and the outlines well marked. The retinal vessels were of normal size. The fundus picture, as described elsewhere, was typical of the disease.

The ocular symptoms are pathognomonic of the disease and are present early. In the beginning there is no change in the appearance of the discs and the child is able to see. Later on there is definite optic atrophy and total amaurosis. Optic neuritis has been observed to precede optic atrophy. The changes at the macula are characteristic and unique and always symmetrical. These changes remain unaltered throughout the course of the disease. About the macula is a nebulous area, grayish-white in color, slightly raised above the surface of the retina and about two to three times the size of the optic disc, with softened edges gradually blending with the normal fundus. The patch is somewhat oval in shape with the major axis horizontal. In its center, coinciding with the fovea centralis, but larger, is a dark red or liver-colored spot clear cut in outline. Surrounding the central spot can be seen distinctly the tiny retinal vessels. Strabismus and nystagmus has been observed in some cases. There is no recorded regularity as to the condition of the pupils.

In but six cases have the eyes been studied microscopically. Treacher Collins reported the first findings in Kingdon's two cases. He found the retina much thickened at the yellow spot, due to enlargement of the outer molecular layer, the tissue of which showed a spacing-out which he regarded as indicative of edema. The most marked changes occurred in the neighborhood of the fovea and became less evident toward the periphery of the affected area. The retina was detached from the choroid, and had been thrown up in folds above the macular region, probably due to faulty fixation with Müller's fluid. The optic nerve was atrophied and showed an increase of interstitial connective tissue and a large number of round cells. At the time of this examination no study had been made of the ganglion cells.

In the third case, that of Peterson's, examined by Ward Holden, the results were unsatisfactory, owing to the advanced postmortem changes.

In the fourth case, that of Hirsch, also examined by Holden, he reported the essential changes in the eye to be degeneration of the ganglion cells of the retina, and of the nerve fibers of the optic nerve and tracts.

In the fifth case, that of Mohr, examined by Preisz, he reported edema and thickening of the outer molecular layers, especially the fiber layer of Henle, which composes the outer portion of this layer in the region of the macula. This edema is ascribed to a probable angioneurotic disturbance, the result of pathologic alterations in the cervical cord. No mention is made of the condition of the ganglionic cells. The outer segments of the rods and cones were degenerated and the formation of a granular substance found between the external limiting membrane and the choroid.

Holden was the first to show the relationship between the fundus changes, especially at the macula, to those found in the central nervous system. He likewise regards the atrophy of the optic nerve, both as an ascending and a descending degeneration, and dependent on a common cause and related to the changes in the cerebral nervous system, as both are embryologically related.

In the sixth case, that of McKee, examined by Shumway and Buchanan, the views of Holden were confirmed.

The nebulous area about the macula, it is reasonable to suppose, results from the swollen and degenerated ganglion cells present in larger numbers in the macular region rather than to an edema. Two facts speak against edema: first, the unchanging appearance of the lesion for months, and the absence of any veiling of the minutest vessels in this area (Falkenheim).

In the fovea the ganglion cells are absent, but at the margin of the fovea the ganglionic layer is six to ten cells deep, gradually growing less until at the periphery it is only a layer of single cells. Thus we have the red center void of cells, next the dense white corresponding to the deepest portion of the layer, fading away gradually as the cells become fewer. In the vertical meridian the ganglion cell layer thins out more rapidly than in the horizontal, explaining the oval area about the macula having its major axis horizontal.

Sachs,² in his last paper, lays great stress on the morbid process in this disease affecting primarily, or at least to a great extent, the larger ganglion cells of the entire gray matter of the brain and of the spinal cord. These confirmed the cellular findings previously described by Hirsch³ and recently by Spiller.⁴

The following is a summary of the statistics of all the cases reported up to July 1, 1905: Thirty-two authors report 54 cases, with histories completed, and 20 additional cases alluded to. Of these patients, 22 are males, 25 females, and in 7 no sex is reported. Twelve postmortems have been performed, and in 6 cases the eyes have been examined microscopically. Twenty-seven cases occurred in Jews, 6 in Gentiles, and in 21 cases no data is given. In 15 cases nystagmus was present. The youngest patient was four months old; the oldest three and three-fourths years.* The statistics compiled from my table differ from those published by Spiller and Falkenheim, owing to the omission from the list of the same case published by different authors.

THE ELIMINATION OF THE NOSTRUM TRAFFIC,

AN EVIDENT DUTY OF AMERICAN PHYSICIANS.*

M. I. WILBERT, PH.M.

Apothecary at the German Hospital.

PHILADELPHIA.

What is a nostrum?

Why should nostrums be eliminated?

Why should physicians be interested in their elimination?

As an answer to the last and certainly the most directly interesting of these three questions it may be pointed out that, apart from the duties and obligations imposed on physicians, by the traditional oath of Hippocrates, the practice of medicine, in every state, is hedged about and protected by laws that are designed to prevent the ignorant and the incompetent from imposing on the people of the commonwealth, and also, in a measure at least, are designed to protect regularly licensed members of the medical profession from imposition and unnecessary competition in the practice of their science or art.

The members of the medical profession, on the other hand, in thus seeking and accepting the protection secured to them under these several statutory enactments, are expected to guard and to protect the lives of the citizens of the state from infection and disease and, incidentally at least, to point out the dangers and, if possible, prevent the use, or at least the abuse, of anything that will in any way endanger the health or the well-being of the individual members of the community.

It is in this capacity, as guardians of the health and the well-being of the individual members of the community, that physicians are in duty bound to take cognizance of the present widespread inquiry into the status of nostrums and their possible deleterious effects on the health and even the lives of unwitting or indiscreet consumers.

WHAT IS A NOSTRUM?

As an answer to the question, What is a nostrum? we may safely accept the usual dictionary definition and include as nostrums, or at least as objectionable nostrums, all medicines "the ingredients of which are kept secret for the purpose of restricting the profits of sale to the inventor or proprietor." Personally, I believe that the word nostrum, in keeping with its evident derivation from the Latin word *noster*, might properly include all medicines or medicinal preparations in which any one individual or firm claims, or maintains, a proprietary right, either by secrecy, trade privileges or official letters patent, thus making the word nostrum practically synonymous with the term proprietary medicine, as used at the present time. I myself believe that this broader definition is quite in harmony with existing conditions and fully in keeping with the facts.

Restricting the word nostrum, however, as being applicable only to proprietary medicines which are in any way secret, or for which extravagant claims are made, as therapeutic agents, I am sure that you will readily agree with me that under no condition is the use of such substances in keeping with what purports to be a scientific occupation, and least of all can their use be made compatible with that most important and most responsible of all duties, the conservation and preservation of human lives.

Surely you will agree with me that if there is any one field of scientific investigation in this wide world that should be absolutely free from commercialism, or even a suspicion of secrecy, trickery and fraud, it should be the science of medicine in the very widest application of that term.

How far removed we are from even an approximate realization of this ideal becomes evident when we learn that, at the present time, nostrums appear to constitute fundamental necessities in the armamentarium of many physicians, as well as being the ever-ready cure-all recommended by blacksmiths and tailors.

The reasons for this unfortunate state of affairs are varied. In the first place we must not forget that nostrums, even the vilest of them, are not readily differentiated from what might be perfectly legitimate proprietary medicines, and we must also bring ourselves to realize that manufacturers of otherwise unobjectionable remedies have adopted, and are now using, highly objectionable methods in exploiting their preparations.

Another potent factor in developing the present widespread use of nostrums by physicians is to be found in the fact that the physician who is at all susceptible to extraneous influences can always find, ready at hand, literature suggestive of the curative qualities of nos-

2. Sachs: Jour. Ment. and Nerv. Dis., January, 1903, p. 1.

3. Hirsch: Jour. Ment. and Nerv. Dis., 1898, p. 538.

4. Spiller: Am. Jour. Med. Sci., January, 1905, p. 45.

*Read before the Philadelphia County Medical Society, Dec. 27, 1905. The report of this meeting is given in this issue, page 218.