THE BRITISH JOURNAL OF OPHTHALMOLOGY

NOVEMBER, 1924

COMMUNICATIONS

CATARACT IN DYSTROPHIA MYOTONICA (MYOTONIA ATROPHICA)

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Introduction

The object of this communication is to bring to the notice of ophthalmologists in this country who may happen to be unfamiliar with it, a remarkable and by no means very rare hereditary disease in which cataract is a common and important symptom.

This disease, dystrophia myotonica as it is now called, came to light in 1909 when Batten and Gibb⁽¹⁾ showed that certain cases that had been described as examples of muscular atrophy in Thomsen's disease really belonged to a distinct and independent category. At that time atrophy of some of the muscles and myotonia were the only known symptoms; but the discovery that cataract occurred frequently directed attention to a number of other extra-muscular symptoms that had hitherto been regarded as more or less accidental and it was soon realized that this strange disorder affected not only the muscles but many other structures as well.

The credit for recognizing the significance of the extra-muscular symptoms is due to Curschmann⁽²⁾, who elevated them to a cardinal position in the symptomatology of the disease and insisted that they must receive full consideration in any attempt to explain its nature. According to the now classical description of this writer the main features of the disease as it was known in 1912 were: (1) Muscular atrophy with the peculiar distribution first described

by Batten (facies myopathica, atrophy of the sterno-mastoids, forearms, vasti and peronei); (2) myotonia, most constant and most conspicuous in the hand-grasps; (3) extra-muscular symptoms especially cataract, atrophy of the testicles with impotence, baldness, vasomotor disturbances and general atrophy and loss of weight.

At this stage in its history dystrophia myotonica was regarded as a disease characterized by this combination of symptoms, and so it remained until 1918 when Fleischer,⁽³⁾ an ophthalmologist, published the results of his investigations into the family history of patients presenting this syndrome and showed that the syndrome represented but one aspect of a heredo-familial disease whose presence in the affected stocks may be shown in various ways. For a full account of the disease the reader is referred to a recent article in another journal⁽⁴⁾. I gave there our reasons for the following general conclusions which must serve here to indicate the state of our knowledge of the disease at the present time. Dystrophia myotonica is a disease *sui generis* within the group of heredo-familial disorders. It forms a link between other well-known members of this group in that two of its cardinal symptoms—myotonia and muscular atrophy—are identical in nature with the same symptoms in Thomsen's disease and the familial muscular dystrophies respectively. At the same time it introduces a new kind of disorder into this group of diseases in the form of certain degenerative phenomena in parts outside the muscular and nervous systems. The fully developed disease is confined in the main to one generation—the dystrophic generation as I have called it—where it occurs in some members of a number of families of the same child rank in relation to a common ancestor. The members of earlier generations do not as a rule suffer from muscular symptoms, but they often develop cataract which tends to appear at an earlier age in succeeding generations, and genealogical investigations reveal other indications of progressive deterioration in some branches of the affected stock. In the dystrophic generation itself some members remain healthy, some may have premature cataract alone, and some may suffer from the muscular disease in an incomplete form, one of the cardinal symptoms, say muscular atrophy being absent; while others present the combination of muscular atrophy with myotonia and extra-muscular symptoms for which the name dystrophia myotonica (or myotonia atrophica) has hitherto been reserved. A majority of the latter group conform to the classical descriptions of Batten and Curschmann, but a consideration of the foregoing statements shows that these descriptions apply to a type only, to a syndrome which is merely one expression of a disease with diverse manifestations.

For the disease in all its aspects no name had been suggested.

I proposed, therefore, that the name dystrophia myotonica should be transferred to the hereditary disease itself from the syndrome to which it has hitherto been attached. For the syndrome which also requires a distinctive name I suggest "dystrophia myotonica, type Batten-Curschmann."

Cataract

Among the many extra-muscular symptoms of dystrophia myotonica cataract holds the place of first importance, not because of its frequency alone, but because of the decisive part it has played in the development of our knowledge of the disease as a whole.

As long ago as 1903 Uhthoff⁽⁵⁾ demonstrated four cases of "cataract in a tetany-like disease"; the same cases were described by Bartels⁽⁶⁾ in a paper on eight cases of "cataract in tetany," and his concise account of the general symptoms makes it certain that four of these cases at least were suffering not from tetany but from the as yet unrecognized disease that now concerns us. Cataract in patients known to be suffering from dystrophia myotonica was recorded for the first time by Greenfield⁽⁷⁾ in 1911; later in the same year cases were reported by Kennedy and Oberndorf⁽⁸⁾ and Ormond⁽⁹⁾ and since then others have been described by Kennedy⁽¹⁰⁾, Bramwell and Addis⁽¹¹⁾, Hoffmann⁽¹²⁾, Tetzner⁽¹³⁾, Fearnside⁽¹⁴⁾, Curschmann⁽²⁾, Mann⁽¹⁵⁾, Löhlein⁽¹⁶⁾, Hauptmann⁽¹⁷⁾, Rohrer⁽¹⁸⁾, Fleischer⁽³⁾, Rülff⁽¹⁹⁾, Adie and Greenfield⁽⁴⁾ and no doubt by others.

This curious combination of degenerative changes in structures so dissimilar as lens and muscle at once arrested attention. Greenfield in his original paper suggested that "the degenerative conditions in the lens and muscles may be due to a deficient vitality on the part of the tissues showing itself in these somewhat specialized structures." In 1912, Hoffmann reviewed the published cases and expressed the opinion that the combination was more than fortuitous; in the same year Curschmann (who saw in the association of cataract with the other extra-muscular symptoms evidence of a general intoxication) rejected all the earlier myogenic theories and formulated the hypothesis that the disease is due to a disturbance in the organs of internal secretion or of the central nervous mechanisms that control them.

Subsequent observations on the incidence of cataract enhanced its importance still further, for increasing knowledge showed that it occurred not only in patients with the fully-developed muscular disease but also in their otherwise healthy brothers and sisters and cousins and in the otherwise healthy members of earlier generations.

In Greenfield's family five children had dystrophia myotonica,

two with and three without cataract, and two had cataract alone; a paternal aunt and grandfather also had cataract. Both parents of Ormond's patient had cataract. Fearnside's patient had myotonia, muscular atrophy and cataract, and two cousins, two aunts, an uncle and a grandfather, all had uncomplicated cataract. I have found no reference to Fearnside's paper in the literature; it deserves to be remembered because it records for the first time the incidence of cataract in a cousin of a dystrophic patient. In a family described by Hoffmann dystrophia myotonica with and without cataract, and cataract alone, were found in the dystrophic generation and a grandfather had cataract. In Curschmann's family cataract occurred in three members of the dystrophic generation, in four of the preceding generation and in one member of the generation before this. The most valuable contribution to our knowledge of this subject was made in 1918 when Fleischer⁽³⁾ published a remarkable account of no less than 35 cases of cataract in dystrophia myotonica that had come under observation in the Eye Department of the University Hospital at Tübingen. His patients were distributed over twenty-seven families; in eleven of these one of the parents of dystrophic children had uncomplicated cataract and uncomplicated cataract occurred frequently in other members of the dystrophic and earlier generations. In one family, for example, the three parents of dystrophic children, an uncle, a sister of the grandfather and a daughter of his sister as well as the common ancestor, the great-grandfather of the dystrophic children, all had cataract.

Altogether I have found records of just over one hundred cases. In 66 of these, muscular symptoms and cataract were found together, cataract occurred alone in 17 members of the dystrophic generation, in 18 parents, uncles or aunts, in 5 grandparents, and in one great-grandparent. The sexes are about equally affected and transmission seems to take place through males and females indiscriminately.

So far I have used the term cataract without qualification, but its characters are not the same in the different generations; in patients with muscular symptoms and in their siblings, cataract, if it occurs at all, comes on at an early age (from the teens to the forties, most often between twenty and thirty-five); in the preceding generation it appears later but is still distinctly pre-senile in most instances, whilst in earlier generations it usually comes on in old age as ordinary senile cataract. That it may still be pre-senile in early generations is proved by Fearnside's observation of cataract at the age of 40 years in the grandfather of his patient. "Anticipation" was seen in eleven of Fleischer's families, and is well illustrated in one where cataract occurred in different members of the dystrophic generation at the ages of 27, 30, 31 and 40 years,

and in the preceding generation at the ages of 37, 38, 52, 56 and 65 years, whilst a member of the generation before this and his father had ordinary senile cataract. In Curschmann's family the ages were 38, 42, 44 and 47 in the dystrophic generation and 50, 50, 78 and 80 in the preceding generation.

With regard to the form of the cataract, most of the cases have been recorded by neurologists who make the bare statement that cataract was present; I therefore append a statement of the changes in the lens that have been seen by ophthalmologists:

Bartels: Case 1. "There are fine streaks and linear opacities especially in the posterior cortex producing a kind of star-shaped figure at the posterior pole; in the anterior cortex the opacities are in general more dust-like, but in places they run together to form streaks."

Case 2. "Both lenses show fine opacities left more radial and fewer than right where they are more diffuse and denser; they are situated more in the posterior than in the anterior cortex."

Case 3. "With a hand lens one sees numerous very small grey dust-like opacities; focal illumination with dilated pupils shows a grey star-shaped figure at the posterior pole with its concavity outwards."

Ormond: "An immature cortical cataract, the greater change being in the cortex behind, where the opacity has a stellate form with the concavity looking forwards; the anterior cortex is not so greatly affected."

Löhlein: "The changes consist partly in heaps of dust-like opacities in star-shaped radial lines near the anterior and posterior surfaces of the lens, partly in the form of a haze throughout the lens."

Szily (Hauptmann's case): "In both lenses a circumscribed star-shaped opacity in the posterior cortical lamellae, more dense at the posterior pole whence streaks radiate towards the equator; there are as well numerous small and very small white specks scattered throughout the lens in all its layers."

Leslie Paton saw in one of my cases: "In the right eye a complete ring of dust-like opacities at the periphery of the lens; they lie mostly anterior, with one or two streaks radiating from the periphery and lying posterior to the ring of opacities; in the left lens the same dust-like opacities and the same streaks are seen."

Fleischer: "The cataract begins in a fairly typical manner in the posterior cortex with opacity of the posterior pole and star-shaped radial processes, then with similar opacities in the anterior cortical lamellae and with simultaneous more diffuse very fine dust-like opacities at various depths in the lens. The cataract ripens fairly quickly to a total soft cataract with a small nucleus at about the same time in both eyes."

Cases of dystrophia myotonica have been reported from all parts of the world, their numbers now running into hundreds, but this disease is not mentioned in many standard text-books, and it is certainly not so well known in this country as it should be. I have seen twenty cases in London since 1919, and as the disease is always familial and as cataract occurs in some members of almost every affected stock, it is almost certain that the number of cases scattered throughout the country is a considerable one. My object will be attained if the foregoing bald narration of some of the facts leads to their more frequent detection.

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REPORT TO THE LANG CLINICAL RESEARCH COMMITTEE, ROYAL LONDON OPHTHALMIC HOSPITAL*

A BILATERAL CHRONIC AFFECTION OF THE ENDOTHELIAL FACE OF THE CORNEA OF ELDERLY PERSONS WITH AN ACCOUNT OF THE TECHNICAL AND CLINICAL PRINCIPLES OF ITS SLIT-LAMP OBSERVATION

BY

BASIL GRAVES

THE affection I am going to describe begins at or very near the central part of the deep face of the cornea, whence it spreads insidiously in a centrifugal direction. In respect of this distribution the condition is a bilaterally symmetrical one; but nearly always

* Submitted to the Lang Clinical Research Committee, August 10, 1924.