Progressive Chronic External Ophthalmoplegia with Disorders of Retinal Pigmentation

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's Gravenhage

A girl aged 16 years was presented. Up to her 14th year she had been normal. At that age the development of ptosis of both eyes commenced. Now, the palpebral levator muscles and the superior rectus muscles are completely paralysed. The internal and external rectus muscles allow a deviation of approximately 10° , the inferior rectus muscles of approximately 20° . The oblique muscles are also completely or nearly completely paralysed. The internal eye muscles show normal activity. The paralyses do not react to prostigmine, so that myasthenia gravis is excluded. Besides ophthalmoplegia, there are also abnormalities in the ocular fundi, viz. grey spots, which cover almost the entire fundi, but not yet the maculae luteae. Vision in both eyes is 0.8.

We found only 4 similar cases in the literature. These were described by Chamlin and B7/ef in Arch. Ophthalm. 1950. In the Netherlands, some time ago, Thiel described a few cases of paralysis of the external eye muscles occurring in Amsterdam; he believed the cause to be in the muscles. However, these cases did not show abnormalities of the fundi. In the present patient, slight abnormalities of the CSF were observed, which require a closer investigation in the further course of the disease.

Discussion.

Mrs. Schappert-Kimmijser remarked: The article by J. H. Thiel, which De Ruyter mentioned, was published in 1954 under the title of "Chronic progressive ophthalmoplegia with extra-ocular extension (ocular myopathy)". In this article, among others, 2 patients observed by me are described. The patients were 2 sisters with progressive chronic ophthalmoplegia, complicated by extension of the paralysis to the muscles of the face and the throat.

The younger sister was sent to me for keratitis and lagophthalmus. Before I examined the patient, Professor Biemond had established the diagnosis of myasthenia gravis, but it was later changed to the above-mentioned diagnosis. The affection was attributed to myopathy.

Halbertsma reported that he had seen the patient a year ago

but at that time she had only ptosis

and the fundus did not yet show the picture that it exhibits now.