A Case of Familial Dwarfism, with Choroideremia, Myopia, Posterior Polar Cataract, and Zonular Cataract

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The patient is a boy aged 9 years. Visual acuity right eye with S – 7.00, C – 1.50, axis 5° = 6/12. Visual acuity left eye with S – 7.00, C – 1.00, axis 0° = 6/9, with errors.
At the demarcation of the cortex and the nucleus, the lens shows a thin layer of small, round, greenish-blue dots, and thin, brownish, scaly turbidities, and also a very small posterior polar cataract.
The fundus picture of both eyes is as follows: pale papilla, very narrow vessels, only the macular region of the pigmentary layer contains a reasonable quantity of pigment, though arranged irregularly and in clusters; the chorio-capillaris is absent; the visual fields are concentrically limited, and the blind spot is connected to the peripheral limit of the visual fields. The adaptation could not be examined because of transport difficulties of the patient, who lives in a very remote district. There are no subjective symptoms of hemeral-opia. The patient is a well-proportioned dwarf of a height of approximately 80 cm. His mother, his maternal grand-parents and two of his mother’s brothers have a body length between 1 m. and 1.50 m. The only abnormality of his mother’s eyes is a small posterior polar cataract. On account of social circumstances, no information regarding the father or his family could be obtained. The association of tapeto-retinal degeneration with dwarfism in this form has never been encountered by the author in the literature.
Nettleship (1) demonstrated in a family in which cataract and pigmentary degeneration of the retina occurred simultaneously, the separate origin in the “kwartierstaat”.
Waardenburg (2) reported the simultaneous occurrence of pigmentary degeneration and choroideremia in one pedigree.
Hansell (3) described pigmentary degeneration and dwarfism combined with other hypophyseal dysfunctions.
Bibliography.
1. Nettleship