We wish to acknowledge our debt to Mr. E. F. King for the sections and report on the eye, and to Dr. S. C. Dyke for similar service in respect of the thyroid tumour. To Mr. King should go no small share of the credit for the case, for he has taken great interest in it and the greater part of the work and worry have been his. We also thank Mr. Vincent Patrick for his interest and for removing the primary tumour, and Dr. J. H. Sheldon for the general examination of the patient.

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**ATYPICAL RETINITIS PIGMENTOSA ASSOCIATED
WITH OBESITY, POLYDACTYLY, HYPOGENITALISM,
AND MENTAL RETARDATION (THE LAURENCE-
MOON-BIEDL SYNDROME)**

(Clinical and Genealogical Notes on a Case)

BY

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ALTHOUGH many cases of this bizarre syndrome have been recorded since the classical paper of Laurence and Moon⁴ in 1866, reports with pedigrees are still sufficiently rare to justify description.

I first saw my patient H. D. through the kindness of Dr. Sharp and Mr. Kelly in 1933 at the Whipps Cross Hospital. He was complaining of night blindness, which, I was told, had first showed itself in 1925, when he was nine years of age. At that time he had been of fair mentality; when seen in 1933 he had degenerated so that it proved impossible to take a visual field on the perimeter, though he could still recognize letters if allowed due time. He was of a gentle, tractable, and friendly disposition, popular in spite of his deficiency with the other patients of his ward. His figure was stout, rather suggesting hypopituitarism, with slight enlargement of the breasts. An X-ray examination of the skull showed a normal sella turcica. His height was five feet, three inches. On the ulnar sides of his hands and outer sides of his feet were operation scars as if for the removal of accessory digits; but he could give no history to confirm this, and at the time it was not known that any relatives still remained alive. The genitals corresponded in development to those of a boy of twelve.

No other abnormalities were found on examination. His blood belonged to Group II.

Right vision with -2.0 D. sph : -1.0 D. cyl. axis 180 was 6/24.

Left vision with -2.0 D. sph : -1.5 D. cyl. axis 180 was 6/24.

The pupils were equal and reacted briskly to light and accommodation. Ocular movements were normal. Fundi showed slightly waxy discs and in the periphery small white dots suggesting retinitis punctata albescens.

Six months later fine pigmentary changes began to show themselves at the fundus periphery. These advanced steadily and by February, 1935, vision was so diminished that the lad knocked

"The operation scar can be seen on the outer side of the right foot (photograph by D. G. Sharp)."

into walls and into other patients. The macular regions showed pigmentation. I could not make him see 6/60, though some of this diminution might have been apparent and due to his failing mentality. Similar macular changes, however, have been noted in other cases.² Although 19 years old he had never needed to shave. He was stouter than in 1933, largely confined to a chair, with few remaining pleasures save the gustatory.

About this time a brother suddenly returned from abroad, and after some difficulty rediscovered H. D. in the hospital. The

brother was five feet seven inches in height, spare, alert, intelligent, active, and well-to-do, yet still preserving an unmistakable family resemblance of gesture and mannerism to my patient. By the brother's good offices I was able to obtain a complete family and case history.

The affected members of the family were III—9, Florence D., who died at eighteen months and had extra fingers and toes; III—13, John Stanley D., who died at three years, obese, mentally backward, with extra toes; and III—14, my patient H. D. His brother, III—8, confirmed my supposition that the operation scars on his hands and feet were from removal of accessory digits. An X-ray picture of H. D.'s left foot showed a remaining sixth metatarsal. The extra fingers and toes in every affected member of the family were "perfect and of normal size."

Exact details of the toes could not be remembered, but the extra fingers had the joints "from the base of the normal little fingers in all cases." H. D. was the only affected member of the family ever subjected to an ophthalmoscopic examination.

Details of the rest of the family are appended for completeness. I—1, was an estate agent; he died of pneumonia. His wife, I—2, died of heart disease. I—3, was a Chinese official and attached to the Chinese legation; his English wife, I—4, and he, both died at a fair age. The patient's father, II—4, was an army officer who died in middle age; his uncles II—1 and II—2, and aunt, II—3, are alive and well. II—1, is childless; II—2, has five healthy children (two girls, III—1 and III—2, and three boys, III—3, III—4, III—5); II—3, has two unaffected boys, III—6, and III—7. The patient's mother, II—5, was unrelated to his father. She died in middle age of bronchitis. Her sister, II—6, is alive, healthy and childless, 64 years of age; her brother, II—7, is healthy, childless, and 58 years of age. Of the next generation the cousins, III, I—7, are healthy. The brother Cecil D., III—8,

is 38 years of age and healthy; III—9, was affected; III—10, was an unaffected sister who died in infancy; III—11, Marie, is healthy, 23 years of age and unmarried; III—12, Doris, 22 years of age, is recently married; III—13, John Stanley D. and III—14, H. D., have both been affected as described. There are as yet no members of a generation following that of my patient.

Remarks:—My patient H. D. is evidently an example of the syndrome of obesity, hypogenitalism, mental retardation, and retinal pigmentary disturbance described by Laurence and Moon.⁴ He does not exhibit the paraplegia^{3, 5} which eventually developed in their cases, and indeed this seems to have been a feature peculiar to the original family described.² The association of polydactyly has been noted by many observers, including Biedl and Raab,¹ and recently Cockayne, Krestin, and Sorsby.² The inheritance is not sex-linked, is of the recessive Mendelian type, probably the result of mutations of two or more genes in the same chromosome, and an example of linkage of genes.² Although details are not complete, it does not seem unwarranted to assume that in the sibship described in this paper, the patient's deceased brother and sister with polydactyly also suffered from the "Laurence-Moon-Biedl" syndrome.

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THE PRECISE ORIGIN OF CORNEAL PITTING

BY

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IN the July, 1935, issue of the *Brit. Jl. of Ophthal.* MacCallan draws the unqualified conclusion, from a short paper published by me two months earlier in the same journal, that I am "not in agreement with the view that follicles are the antecedents of Herbert's peripheral pits."