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COMMUNICATIONS

THE LAURENCE-MOON-BIEDL SYNDROME— RECORD OF A CASE

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THE following isolated case is submitted because of the rarity of the syndrome, and because it happens to show all the defects observed in previous cases along with several minor interesting features. I first saw the patient in June, 1936, and when I read an article* on "Obesity, Hypogenitalism, Mental retardation, Polydactyly, and Retinal pigmentation (The Laurence-Moon-Biedl Syndrome)" I realised that I had stumbled on a case showing the complete syndrome.

Family History.—The patient's mother, the second eldest of a family of twelve, is alive and healthy, and has no abnormalities. Of this family three males and one female are living. James, now aged 56 years (X on chart) has very defective vision. He has been seen by an ophthalmic surgeon, who diagnosed retinal degeneration and optic atrophy. He is able to herd cattle on a farm, but he is unable to read. He is not very bright mentally, but is not obese. There is no polydactyly. The condition of the

* A description, history and bibliography of the syndrome may be found in the admirable article by Cockayne, Krestin and Sorsby in the *Quarterly Journal of Medicine*, April, 1935.

genital organs is unknown; he is unmarried. The seventh member of the family is alive, a healthy woman with no known abnormality. The second youngest male lives in South Africa. He is unmarried, and has no known abnormality. Fred., aged 43 years (Y on chart) has retinal degeneration and nearly complete optic atrophy. He can walk about the farm alone, but is unable to see people. He is quite sound mentally, is not obese

FIG. 1.

Chart of Pedigree. (X) (Y) affected individuals. (P) patient (A.F.)

and does not show polydactyly. The condition of the genital organs is unknown; he is unmarried.

The patient is the third of a family of five, four being males and one female. There was no parental consanguinity. The second eldest son was killed in the Great War. The remaining two brothers and the sister are alive and normal; the sister has two healthy sons.

History of patient.—A. F., aged 35 years. At the age of 12 years, his vision became dim and he was unable to read small print. After leaving school his vision improved, and he was able to read all sizes of print without difficulty. Five years ago, he began to have attacks of severe headache, the headache often being relieved by epistaxis. About this time, his eyesight again began to fail. He was fitted with glasses by an optician, but as the glasses did not help him, he discontinued their use. His vision is worse in bright sunshine and during the daytime, and improves towards evening. He has no difficulty in walking about after dark. He has worked continuously as a farm-servant, and

manages to perform his duties fairly well, but has been unable to read the newspapers for about a year.

Previous illnesses.—He had measles and chicken-pox when a child, and he has had pleurisy twice. In July, 1935, he was admitted to the Aberdeen Royal Infirmary suffering from acute appendicitis. His appendix was removed and he made an uneventful recovery.

Present condition.—Apart from occasional headaches, his general health is good.

FIG. 2.

Profile view of A.F.

Physical examination.—Height 5 ft. 2 ins. Weight without clothes, 12 st. 5 lbs. Walks with a slight stoop. Limbs and body well covered, abdomen protuberant, protuberance accentuated by lordosis of lumbar vertebrae (Fig. 2). Limbs possess good power, tendon reflexes present and equal on the two sides. Co-ordination good. Sensation to pain, touch and temperature normal. Jaws edentulous. Para-central scar, right side of lower abdomen. Liver and spleen not palpable. *Genital organs.*—Penis small, $1\frac{1}{2}$ ins. in length. Hypospadias, urethral opening being about $\frac{1}{2}$ in. along under surface near lower border of glans.

FIG. 3.

Photograph of feet of A.F.

The right testicle is normal in size, the left rudimentary. *Feet.*—Polydactyly and syndactyly are present in both feet. The supernumerary toes are post-axial, being situated between the fourth and fifth toes. The skiagram shows in the right foot a bifurcated proximal phalanx, the double toes each having two additional phalanges. The additional toe on the left foot is not so well developed, consisting of an off-shoot from the distal end of the proximal phalanx. The syndactyly consists of webbing between the second and third toes of both feet, only the terminal phalanges being separate (Figs. 3 and 4). *Hands.*—The fingers are short

and stumpy, but the skiagram shows no bony abnormality. *Cardiovascular system*.—Pulse, 72 per min. Blood pressure, 140/95. Heart normal. Chest normal. Haemoglobin, 93 per cent. *Cerebro-spinal fluid*.—Cells not increased. Globulin, weakly positive. Colloidal gold negative. Wassermann reaction negative. Kahn, negative. *Urine*.—Clear. Specific gravity 1025. No abnormal constituent. *Radiological examination of head*.—Sella turcica normal. No abnormality of bones of skull.

FIG. 4.

Skiagram of feet of A.F.

Ophthalmological examination.—The pupils are medium in size, react sluggishly to light, and slightly better on convergence. They dilate fully with homatropine. The ocular tension is normal. All movements are full, there is no strabismus, or nystagmus. V.O.D. 6/60, refraction emmetropic. V.O.S. 6/60, refraction emmetropic. Reads Jaeger 6 type slowly. The fields (Fig. 5) are generally contracted, and show a defect in the left upper temporal field. An indefinite central scotoma for red and green can be demonstrated in both fields. He is not colour blind, but often makes mistakes regarding colours. Both optic discs are pale, with sharp edges. The arteries are narrower than normal. No pigmentary change could be seen at the periphery, but there is slight mottling at both macular areas, with a grey spot near the left macula (Fig. 6).

FIG. 5.

Visual fields of A.F.

FIG. 6.

Drawing of left fundus of A.F., showing optic atrophy and macular degeneration.

Mental condition.—He has a vacant solemn expression, never smiles and always speaks in a rather low husky voice. When asked a question he always takes some time to answer. His memory appears to be good, but it was very difficult to get a history of his past life; no information was volunteered, and in most cases leading questions had to be asked. It was found impossible to obtain accurate particulars of his relations from him. His mentality is definitely below normal.

Comment

THE visual symptoms appeared late, being first evident at the age of 12 years; it is interesting to note that remission occurred, vision being apparently normal from 14 to 30. Unlike the usual state of affairs in retinitis pigmentosa, the patient sees best in a dull light, and has no difficulty in finding his way about in the dark. At the periphery the retina appears to be normal, only the macular area being affected, this is not surprising when it is remembered that the macular area is photopic in function, and has a relatively high light threshold compared with the remainder of the retina which is scotopic in function; in the eye adapted for darkness it is about one thousand times more sensitive to light than the fovea.

Summary

THE patient shows the complete Laurence-Moon-Biedl syndrome.

(1) Moderate obesity; (2) Hypogenitalism and hypospadias; (3) Mental retardation; (4) Polydactyly; (5) Syndactyly; (6) Optic atrophy and macular degeneration; (7) Lordosis of lumbar vertebrae.

I am indebted to Dr. R. J. E. Paterson for supplying me with the particulars of the pedigree, and to Professor L. S. P. Davidson for the physical examination of the patient.