A REPORT ON FOUR CASES OF LAURENCE-MOON-BIEDL-BARDET SYNDROME

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The Laurence-Moon-Biedl-Bardet Syndrome has been described as a pentad comprising a pigmentary degeneration of the retinitis pigmentosa type, obesity and hypogonadism of the Babinski-Fröhlich type, polydactyly, and mental deficiency. It is estimated that only about 20% of cases exhibit the complete syndrome (Bisland). Other associated defects have been described as occurring infrequently, such as atresia ani, deafness, syndactyly and skeletal defects.

The polydactylism may affect the hands and feet and the extra digit is usually towards the fifth finger or toe. The condition is found more frequently in males and is believed to be transmitted by a recessive gene.

Most of the cases described in the literature were in Caucasian races, but cases have been reported in Indians (Kutumbiah and Abba 1942) and Negroes (Scott and Johnson 1942, Snell, A. C. 1942). In South Africa, Kessel in 1952 recorded what appears to be the first example of this syndrome, which occurred in a pure Bantu female, aged 5½ years.

The literature on this syndrome has been fairly completely reviewed by Streiff and Zentler 1938 and Burns 1950.

Four cases exhibiting this syndrome have recently been seen in the South African Bantu. Two brothers (seen by J.E.W.) and 2 sisters (seen by S.E.). The 2 pairs are not related to each other and came from different parts of the country.

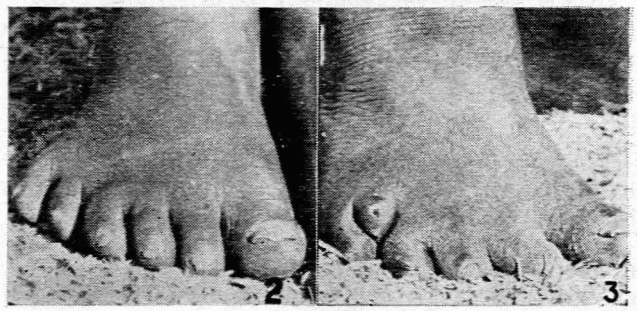
THE BROTHERS

The 2 brothers, aged 14 and 12 years, Bantu, were members of a large family, the other children being normal.

Fig. 1

Family History. Other information regarding the family was extremely difficult to obtain. However, a maternal aunt is alleged to have a sixth finger on her left hand but it appears that she does not manifest other signs or symptoms of this syndrome.

Polydactyly. Both boys were born with six digits on each limb, but the elder boy had the supernumerary



Figs. 2 and 3

digits on his hands removed surgically at an early age (Figs. 1, 2 and 3).

Mental Retardation. Both appeared to be very retarded mentally, but owing to their poor visual acuity it was rather difficult to assess the exact extent of their retardation.

Obesity started in infancy and was most marked in the face, abdomen and thighs. The genitalia appeared to be normal (Fig. 4).

Ophthalmoscopic Findings. The fundi in both cases presented the typical appearance of retinitis pigmentosa. The discs were of a waxy yellow colour, the arteries attenuated, and typical bone corpuscle pigment scattered in the mid-periphery. Refraction revealed no abnormality, but the visual acuity was less than 6/60 owing to early macular changes and the fields of vision were very constricted.

THE SISTERS

The two sisters, Bantu, were 17 and 10 years old. Their father noted that they had defective vision. His other 5 children were normal. No other significant family history was obtainable.

The 17-year-old girl had a full round face and a pituitary type of obesity. Striae were present in the skin of the abdomen and thighs and the breasts were pendulous. No evidence of hypogenitalism was noted. Mentally she was obviously retarded. Polydactyly was not present, but the fingers and toes showed brachydactyly.

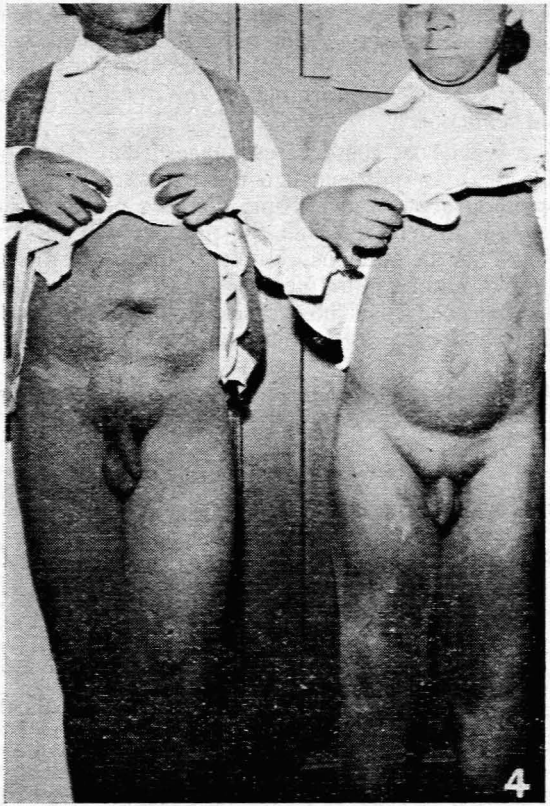


Fig. 4

The fundi presented a tapeto-retinal degeneration involving both the peripheral and macular areas. The appearance was that of an atypical retinitis pigmentosa. The media were clear. The refraction showed a myopic astigmatism. The corrected visual acuity was below 6/60 on both sides.

The 10-year-old girl had a large, round, moon face and a type of obesity similar to her sister's, but no striae were present in the skin. She had no obvious hypogenitalism. There was no polydactyly but, again, brachydactyly was a notable feature. The patient was mentally retarded.

The fundi showed an atypical retinitis pigmentosa with early macular involvement. The media were clear. The refraction showed a mixed astigmatism. The corrected visual acuity appeared to be below 6/60 on both sides.

Discussion. The two Bantu sisters represent incomplete forms of the diencephalo-retinal syndrome. They showed the typical obesity, mental retardation and tapeto-retinal degeneration. Polydactyly was absent and hypogenitalism could not be confirmed. A feature of these cases was the brachydactyly, a characteristic which does not appear to have been previously noted in association with the Laurence-Moon-Biedl-Bardet Syndrome.

REFERENCES

Burn, R. A. (1950): Brit. J. Ophthal., **34,** 65.
Kessel, I. (1952): J. Pediat. 40.1.96.
Kutumbiah and Abba (1942): Indian J. Pediat. (quoted by Burn [1]).
Scott, R. B. and Johnson, P. T. (1942): Amer. J. Dis. Child., **63,** 733.
Snell, A. C., Jun. (1942): Arch. Ophthal., **28,** 12.
Streiff, E. B. and Zeltner, C. (1938): Arch. Ophtal., **2,** 289.