LAURENCE-MOON-BIEDL SYNDROME*

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THE Laurence-Moon-Biedl syndrome consists of obesity, polydactyly, hypogenitalism, mental retardation, deafness, and retinitis pigmentosa. All the typical features of the syndrome are displayed by only a few patients and it is very rare to come across all the manifestations in one individual, as in the case reported here.

Case Report

A young man aged 18 years attended the out-patients' department, complaining of defective vision in both eyes, more marked in the dusk. He had been born at full term, and was the first child of normal parents with three younger brothers who were healthy and had no ocular complaints. As far as could be ascertained there was no family history in the past three generations. The various milestones in the development of the patient had been delayed; he was of dwarfish stature (height 3ft 8in.), was rather heavily built (8.5 st.), and was gradually putting on weight. He was mentally dull and partly deaf. Fatty deposits of female type were seen in the region of the breasts, lower abdomen, buttocks, hips, and upper thighs. He had one extra finger and toe next to the normal little finger and toe on each hand and foot. He had flat feet. The testicles were descended, but the genitalia were underdeveloped, with no pubic hair.

The visual acuity in both eyes was reduced to perception of hand movements close to the face, not improved with glasses, and there was unilateral posterior polar cataract.

The discs were pale and waxy, with attenuated blood vessels, and pigment distribution in the equatorial region. The typical bony corpuscular appearance was not seen, but there were pigment deposits of various shapes and sizes near the blood vessels.

The routine blood and urine examinations were normal.

The Wassermann reaction was negative.

Discussion

Höring (1864) and Stör (1865) first described the association of polydactyly with retinitis pigmentosa, and Laurence and Moon (1866) first described the clinical entity. Bardet (1920) added the symptom of polydactyly to the condition described by Laurence and Moon and deafness was added by Burn (1950), who made a detailed study of the syndrome. Various other local and general features described are posterior polar cataract, strabismus, myopia, congenital heart disease, hypothyroidism, tetany, and dwarfism.

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The condition appears to be transmitted by a recessive gene.

Until 1958 only 280 cases had been reported in the literature (Ram and Rohatgi, 1958; Nirankari, Manchanda, and Maudgal, 1960, reported four more cases) but only forty of the total of 284 displayed all the features of the complete syndrome.

Cockayne, Krestin, and Sorsby (1935), Ellis and Law (1941), and Taylor (1947) have described four typical fundus pictures. In some the typical pigmentary disturbances are typical of retinitis pigmentosa, but in others the pigment deposition is more marked in the centre; a third group shows macular dystrophy with atypical retinitis pigmentosa, and the last group shows macular dystrophy only.

The present case shows all the typical features of the syndrome: dwarfism, hypogenitalism, fat deposition of Fröhlich's type, mental retardation, polydactyly, posterior polar cataract, and retinitis pigmentosa. Only the heredity factor seems to be missing, but Nirankari and others (1960) have shown that this is not a necessary feature.

Summary

A typical case of Laurence-Moon-Biedl syndrome with no apparent hereditary linkage is described and discussed.

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