LAURENCE - MOON - BIEDL SYNDROME *

Nihat Atav, M.D. **

The Laurence-Moon-Biedl syndrome was described in 1866 by Laurence and Moon, who observed polydactylism, obesity, and poor eyesight in a family of 8 children. In 1922 Biedl noted the familial tendency of this syndrome, reporting it in several members of the same family. Solis-Cohen and Weiss[1] proposed the term "Laurence-Moon-Biedl syndrome" in 1924 when they summarized the reported cases. Since Bardet[2] added polydactylism as one of the criteria for diagnosis, some authors prefer the term Laurence-Moon-Bardet syndrome. Reilly and Lisser, in 1932, reviewed 77 cases. In their study the syndrome was complete in 25 cases, questionably complete in 10 and partially complete in 26. 16 cases were doubtful. In 1937 Warkany, Frauenberger, and Mitchell[3] reviewed the subject. They found 102 cases in the literature up to date. Only 24 showed the combination of the five required symptoms. In the remainder of the cases in which the syndrome was complete, it was "supercomplete"; that is, there were additional heredodegenerative symptoms which could not be considered accidental. Thus 10 cases remain in which were present only the five typical symptoms of the Laurence-Moon-Biedl syndrome. Therefore, Warkany came to the conclusion that this syndrome does not represent a disease entity but is a rare combination of more or less frequent heredofamilial symptoms.

The characteristic symptoms of the Laurence-Moon-Biedl syndrome are obesity, retinal degeneration, genital hypoplasia, polydactylism, and mental retardation. Frequently associated are nystagmus, strabismus, deafness and syndactylism.

Obesity: This is a prominent feature in each case reported and is of the hypopituitary type.

Retinitis: This has been observed in 90 per cent of the cases and is usually in the form of retinitis pigmentosa. The visual disturbance is manifest by the fifth year in the form of night blindness but sometimes it may be manifest during infancy. The retinas show only scattered areas of pigmentary degeneration along the vessels.

* From the Research Institute of Child Health, Hacettepe, Ankara.
** Formerly Associate in Pediatrics.

There is seldom optic atrophy. Clay[4] stated that only 15 per cent demonstrate typical retinitis pigmentosa.

Polydactylism: This symptom is almost always present. The extra digit is usually situated near the smallest finger or toe.

Mental retardation: This is noted early; it may be mild, but the intelligence quotient is often on the moronic level. Rarely, mental retardation may begin following a normal period of mental development.

Genital hypoplasia: This symptom is also relatively common, but it is difficult to detect in young age groups.

Family tendency: There is a strong family tendency in this syndrome, which has been reported in 80 per cent of the cases.

Etiology

Cooperstock,[5] in discussing the causation, stated: "....the most acceptable concept recently set forth, is that the condition results from a hereditary developmental disturbance and that the various parts of the syndrome represent mesoblastic (polydactylism) and epiblastic (obesity, retinitis pigmentosa, mental inferiority and genital dystrophy) defects which are recessive and due to mutations of two genes of the same chromosome." Numerous other causal factors have been suggested, such as endocrine imbalance, cerebral aplasia from hydrocephalus, etc.

Treatment

Thyroid extract has been used successfully in reducing the weight of these patients. Frequently various pituitary extracts have been administered, but they are of little or no value. The combined endocrine therapy has been said to be helpful in restoring muscular tone and gives some subjective relief from symptoms referable to the eyes if begun early in the course of the disease. Therapy has no effect on the genital dystrophy. I wish to add to the literature one case.

Case Report

A. C., a 10 year old male child (Figure 1), who is originally from Pakistan, was brought to our clinic on February 27, 1959 because of mental retardation, failure to see in the dark and obesity.

History: According to the father, the child's mental and motor development had been very slow since infancy. He was still unable to do even very simple things. He was slow in understanding and

could not talk properly. He had not yet been in school. Failure of vision had been slowly progressive from the age of two years. During the last few years he had been having considerable difficulty in getting about at night, often stumbling over objects, but in the light he could even run. He had shown polydipsia and polyphagia since infancy and obesity had been progressive.

Pregnancy, birth history and neonatal period were normal. At birth it was noticed that he had six toes on each foot. At four years of age the sixth digit on the left foot was removed surgically, but the right foot was not touched. He had severe diarrhea at three months of age. There is a history of frequent upper respiratory tract infections throughout life.

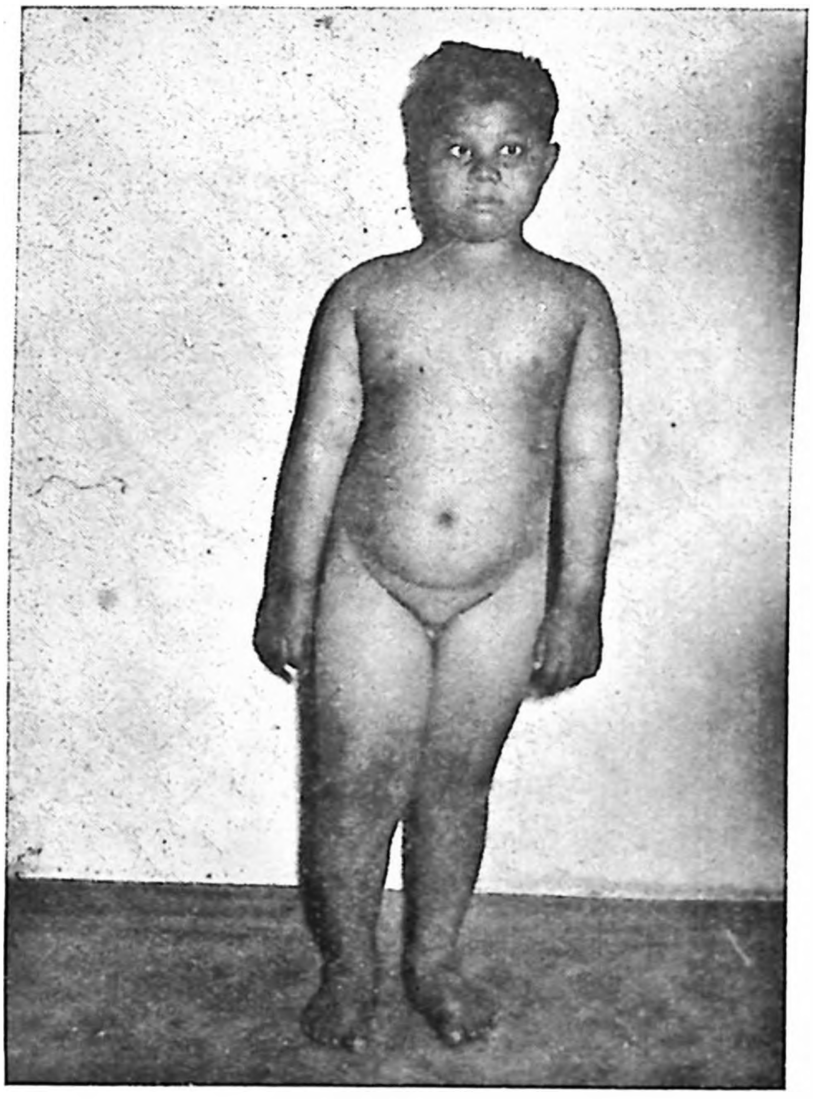

Figure 1.

Family history: He has three sisters living and healthy. One brother died at nine months of age because of diarrhea. There was one miscarriage, cause unknown. The father, 38, and mother, 29, are

living and healthy. No mental retardation, night blindness, obesity, nor any kind of foot deformity were described in the family.

Physical examination : Height 127 cm. Head circumference 49 cm. Chest circumference 80 cm. Weight 40.7 kg.

Figure 2.

The boy was obviously below par mentally. He had a type of obesity corresponding with that usually ascribed to dystrophy adiposogenitalis. Obesity was pronounced in the proximal parts of the extremities, over the lower part of the abdomen and in the region of the breasts. The neck was moderately short and thick. The skin was dark in color, smooth and warm. There was no enlargement of the thyroid. There was a fat pad over the suprapubic region and the external genitalia appeared smaller than normal for his age. The right testis could not be palpated. There was moderate relaxation of plantar arches. The big toes were larger than normal. There was a wide space between big toe and small toes. There were two toe nails on the smallest toe of the left foot (Figure 2). A small scar was noticed at the lateral aspect of the right foot, located just outside of the smallest toe. He was unable to count the examiner's fingers at a distance of three feet. Fundoscopic examination revealed pigmentary stippling at the periphery. The fundus looked pale and the retinal vessels were constricted. An eye consultation was obtained and a diagnosis of moderate myopia and retinitis pigmentosa was made.

The head was normal in shape but looked somewhat microcephalic. The remainder of the examination was not contributory.

Laboratory data: Complete urinalysis was within normal limits. Phenylpyruvic acid was absent from the urine. Complete blood count was normal. P. P. D. was negative.

Roentgenologic studies: Roentgen films of the skull did not reveal any definite abnormalities. Studies of the bones showed no evidence of delayed development.

Discussion and Conclusion

The characteristic symptoms of the Laurence-Moon-Biedl syndrome, which are obesity, retinal degeneration, genital hypoplasia, polydactylism, and mental retardation, were all present in our case. I. Q. and B. M. R. could not be determined. No family history of any similar condition could be elicited but this is not necessarily reliable because the father, informant, did not know enough about the family history. This case is considered to be a "complete Laurence-Moon-Biedl Syndrome."

REFERENCES

1. Solis-Cohen, S., and Weiss, E.: Dystrophia adiposogenitalis with atypical rétinitis pigmentosa and mental deficiency (Laurence Biedl syndrome). Am. J. M. Sc., **169**: 489-505 (April) 1925.
2. Bardet, G.: Sur un syndrome d'obesité congenitale avec polydactylie et retinite pigmentaire (contribution a l'étude des formes clinique de l'obesité hypophisaire). Thesis, Paris, no. 470, 1920.
3. Warkany, J., Frauenberger, G. S., and Mitchell, A. G.: Heredofamilial deviations: I. The Laurence-Moon-Biedl syndrome. Am. J. Dis. Child., **53**: 455 (Feb.) 1937.
4. Clay, G. E.: Laurence-Moon-Biedl syndrome. Tr. Am. Opth. Soc., **31**: 274-288, 1933.
5. Cooperstock, M.: Laurence-Moon-Biedl syndrome. Am. J. Dis. Child., **54**: 334-343 (Aug.) 1937.