Polydactyly and partial blindness

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During a survey for diabetes mellitus in rural India, a 34-year-old man with polydactyly and partial blindness drew the attention of this author. He was born of a consanguineous marriage and had healthy siblings. His parents report that the milestones of developments were apparently normal up to the age of four years when they noticed his difficulty in identifying objects and moving in familiar surroundings, initially at night but progressing. He was obese, did not attend school and was looking after the animal farm with accidental injuries. Secondary sexual hair growth was complete by 16 years. There was no history of polyuria, polydipsia or weight loss nor any symptoms to suspect renal failure. There was no family history of diabetes. On further enquiry, his niece born of consanguineous marriage had a similar history.

On examination, he weighed 68.5 kg with a height of 1.55 m and a body mass index of 28.5. Hexadactyly affecting the feet and brachydactyly of both the extremities were obvious. Injury marks were seen on both shins (figure). Simple assessment of higher functions revealed his inability to repeat four digits forwards and three digits backwards. Ophthalmologic review confirmed his inability to count fingers and limitation of vision to hand movements. Fundus showed pigmentary changes all over in both the eyes with pale discs. Examination of the genitourinary system revealed microphallus and an empty scrotal sac. Urine was checked for glucose and proteinuria and these were negative. Capillary blood glucose after a 75 g glucose load was normal (7.2 mmol/l). His condition has been explained to his parents and the potential dangers of cryptorchidism but intervention has been refused. His niece also was obese, had polydactyly of feet, brachydactyly, myopic disc with pigmentary dystrophy and normal glucose tolerance.

Questions

- 1 What is the diagnosis?
- 2 What are the cardinal features of this condition?
- 3 Name two syndromes which closely resemble this condition.

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Figure

Answers

QUESTION 1

Laurence-Moon-Biedl syndrome.

QUESTION 2

Laurence-Moon-Biedl syndrome (LMBS) is characterized by pigmentary retinopathy, polydactyly, mental retardation, obesity and hypogonadism. The presence of pigmentary retinopathy and at least three other cardinal features confirms the diagnosis.¹

QUESTION 3

Alstrom syndrome has similar features but polydactyly or mental retardation are absent. Acanthosis nigricans, nerve deafness, nephropathy and baldness are also associated with this condition. Prader-Willi syndrome is associated with dwarfism, obesity, subnormal mentality, and hypogonadism. Hypotonia characteristically manifests in infancy and hyperphagia and speech defects are common.

Discussion

These two cases with varying degrees of consanguinity support an autosomal recessive pattern of inheritance with differing degrees of penetrance in LMBS. Both are obese though not excessively and do not have glucose intolerance. The recognised features include impaired glucose tolerance, cataracts, strabismus, nystagmus, renal structural and func-

tional abnormalities with chronic glomerulonephritis, hypertension and cryptorchidism. Brachydactyly is common and hexadactyly affects feet more than the hands²; these findings were noticed in both our subjects.

Mental retardation is not uniformly accepted as a cardinal manifestation, although emotional and intellectual impairment can result.² Simple cognitive function assessment with digit span testing confirms poor abilities in our cases. However, it is not easy to assess the mental functions as they are invariably influenced by the age at which blindness started and a lack of training and learning facilities during childhood. Hypogonadism is attributed to the hypothalamo-pituitary hypofunction but there is little evidence for this. Men with LMBS have not fathered children but there are reports of women giving birth.³

There are about 600 cases of LMBS in the world literature with a higher incidence in mixed Arab populations.¹ Reports have also come from the Indian subcontinent.⁴ Renal abnormalities are increasingly recognised and have been suggested to constitute a sixth cardinal feature, manifesting in failure before the age of 30.⁵ It is interesting to note that our first subject is surviving and did not manifest renal failure. The management includes visual aids and training, diet restriction if morbidly obese, surgery for cryptorchidism and monitoring renal function.

Recent reviews^{6,7} suggest that LMBS is no longer a single entity but comprises two disorders: Bardet-Biedl syndrome in which polydactyly is common, along with other features, and Laurence-Moon syndrome in which neurologic features – spastic paraplegia and ataxia – are common while polydactyly is rare. Our subjects do not have these findings, or any other neurological involvement and hence manifest Bardet-Biedl syndrome.

Final diagnosis

Laurence-Moon-Biedl syndrome.

Keywords: Laurence-Moon-Biedl syndrome, retinal dystrophy, polydactyly

Laurence-Moon and Bardet-Biedl syndromes

- autosomal recessive inheritance with variable penetrance
- retinal dystrophy
- polydactyly
- obesity
- mental retardation
- hypogonadism
- renal disease
- spastic paraplegia in Laurence-Moon syndrome

1 Thoene JG. Laurence-Moon-Biedl Syndrome (LMBS). In: Thoene JG, ed. *Physicians guide to rare diseases*. New Jersey: Dowden Publishing Co. 1992; pp 85–7.

2 Green JS, Parfrey PS, Harnett JD, et al. The cardinal manifestations of Bardet-Biedl syndrome, a form of Laurence-Moon-Biedl syndrome. *N Engl J Med* 1989; 321: 1002–9.

3 Bell J. Laurence-Moon syndrome. In: Penrose LS, ed. *The treasury of human inheritance*. Part III, Vol. 5. London: Cambridge University Press, 1958; 51–96.

4 Malik GM, Mubarik M, Khan MD, et al. Laurence-Moon-Biedl syndrome with chorea. *J Assoc Physicians India* 1995; 43: 295–6.

5 Williams B, Jenkins D, Walls J. Chronic renal failure: an important feature of Laurence-Moon-Biedl syndrome. *Postgrad Med J* 1988; 64: 462–4.

6 Schachat AP, Maumene IH. Bardet-Biedl syndrome and related disorders. *Arch Ophthalmol* 1982; 100: 285–8.

7 Anon. Laurence-Moon and Bardet-Biedl syndromes. *Lancet* 1988; 2: 1178.