CASE PRESENTATION

Bardet-Biedl Syndrome in a Bahraini Family

Dr Shaikha S Al-Arrayed*
Dr Hassan H Al-Arrayed**

Bardet-Biedl syndrome (**BBS**) is a rare autosomal recessive disorder characterised by five cardinal signs: retinal pigmentary degeneration, obesity, hypogonadism, mental retardation and polydactly.[1,2] In this review, we are reporting the first family in Bahrain with **BBS.** Two members of the family are affected, they manifest the typical signs of **BBS.**

THE CASES

A Bahraini family with eight sibs, six males and two females, has been investigated. Two of the males are affected with **BBS,** the others are normal. The parents are first cousins and there has been no previous family history of the same disorder in both sides of the family for at least three generations.

CASE 1

This 25-year-old male patient is short (163 cms in height) and very obese, weighing 160 kg at the time of the investigation (Fig 1). He had polydactyly in both hands and left foot which was excised at the age of twenty. He developed severe pain at the excision site of the toe and had another operation for the excision of an extra metatarsal bone. He had a pilonidal sinus which was excised but developed a fistula which required fistulectomy. Sigmoidoscopy showed normal mucosa with no polyps. He had poor vision especially at night with visual acuity was 6/36, in the right and left eyes(with no error of refraction).

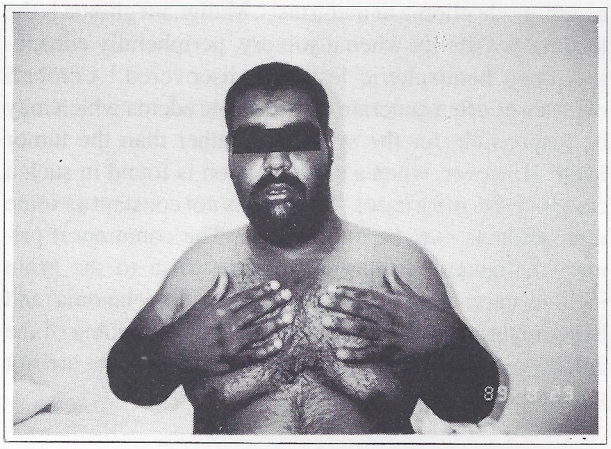

Figure 1

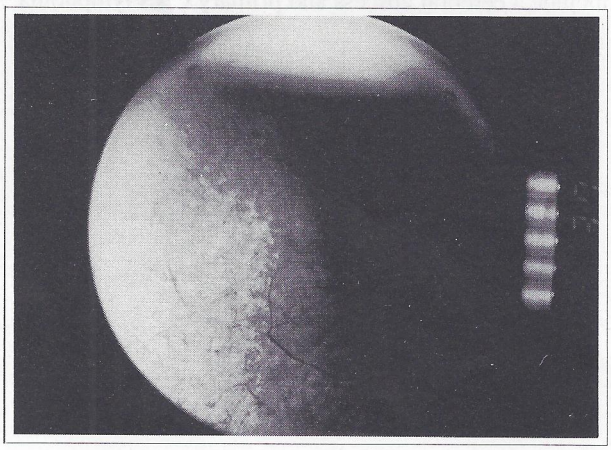

Figure 2

* Clinical Geneticist
Genetic Unit
Salmaniya Medical Centre
State of Bahrain

** Consultant and Chairman
Ophthalmology Department
Salmaniya Medical Centre
State of Bahrain

Fundus showed narrow retinal vessels, especially the arterioles with moderate tapetoretinal degeneration in mid periphery and slight pallor of optic disc. He was diagnosed as having retinitis pigmentosa (RP) (Fig 2). He also complained of difficulty in hearing. Examination of sexual characteristics revealed a very small phallus but normal hair distribution, and no urinary symptoms. His blood pressure, pulse rate, respiration and haematological and biochemical investigations were within normal limits. Chest X-ray showed cardiomegaly and increased vascular marking. Abdominal ultrasound showed a large liver with normal abdominal organs.

CASE 2

Mentally retarded, this 18-year-old male patient is short in stature (161 cms in height) and obese, weighing 148 kg at the time of the investigation (Fig 3). He had polydactyly in both hands and feet which was excised in early childhood. He had poor vision and was diagnosed as having retinitis pigmentosa. His visual acuity, as measured by Catford method, was 6/60 in both eyes (refraction - 4.00 in the right eye and - 3.00 in the left). Fundus examination showed narrowed retinal vessels, especially arterioles, very mild tapetoretinal degeneration in mid periphery (Fig 4). Examination of external genitalia revealed a small phallus; his secondary sexual characteristics revealed hypogonadism as evidenced by scanty hair growth in the face and body.

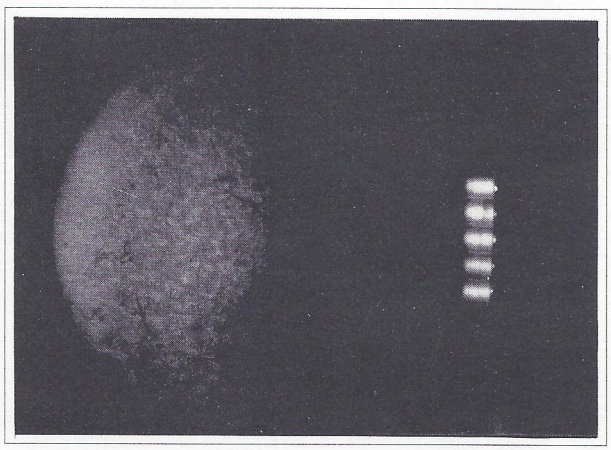

Figure 3

DISCUSSION

BBS can be detected usually in childhood or early adolescence. It is thought to result from an autosomal recessive gene. The prevalence rate of this syndrome is 1:160,000 with a primary sex ratio of one. Pigmentary retinal degeneration occurs in 90% of those patients[3-6] with severe loss of peripheral and central vision as well as pigmentary changes in the fundus. The age at which the degenerative retinal changes begins varies widely; approximately 75% of those who have pigmentary changes

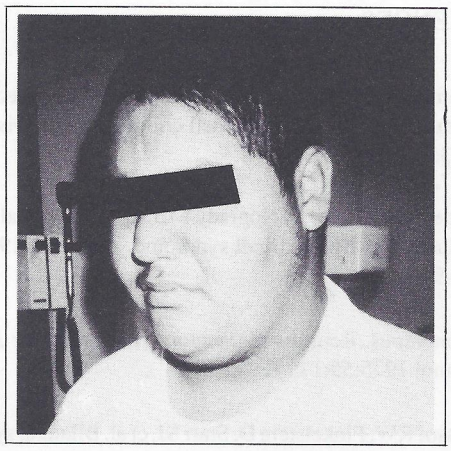

Figure 4

are legally blind by the age of 20 and 87% by age of 30 years. Night blindness may be the presenting sign. Electroretinopathy shows the pattern of tapetoretinal degeneration . Early detection can be facilitated by performing electroretinogram (ERG) in offspring and younger siblings of patients with RP. The finding of subnormal or extinguished ERG have been documented in as early as 7 months old babies.

Generally, obesity in **BBS** has its onset in infancy or early childhood and progresses with age. Some degree of mental retardation is found in 86% of patients with this condition.[1,7] Digital anomalies include syndactly with or without polydactyly and may occur on any or all limbs. Short and broad fingers and toes may occur.[1] Both of our patients have shown hypogonadism, delayed or incomplete pubertal development, as well as a wide variety of genital defects including small phallus, bifid scrotum, hypospadias, and cryptorchidism as expected of patients with **BBS.**[7,8]

Endocrine disturbances are not frequent but include diabetes insipidus, tall and short stature and rarely abnormal glucose tolerance. Renal abnormalities are frequent.[2] Cardiovascular defects are common. Other associated defects include anal atresia, progressive nerve deafness.[9]

The prognosis is good, and life span is good if no cardiac or renal abnormality is expected. If these are present, then prognosis depends upon severity of associated complication.

CONCLUSION

We recommend the establishment of a proper genetic registry for all Bahraini families with positive history of retinitis pigmentosa for early diagnosis and treatment of those family members with BBS and to provide them with the necessary genetic counselling at the appropriate time.

REFERENCES

1. Bauman ML, Hogan GR. Laurence-Moon-Biedl syndrome, report of two untreated children. Am J Dis Child 1973;123:119-26.

2. Hurley RM, Dery P, Nogrady MB, et al. The renal lesion of Laurence Moon Biedl syndrome. J Pediatr 1975;87: 206-9.

3. Anonymous. Retinitis pigmentosa [Editorial]. Br J Ophthalmol 1975;59:175-6.

4. Massof RW, Finkelstein D, Starr SJ, et al. Bilateral symmetry of vision disorders in typical retinitis pigmentosa. Br J Ophthalmol 1979;63:90-6.

5. Levy NS. Early diagnosis and evolution of dominant retinitis pigmentosa. Am J Ophthalmol 1978;86:553-6.

6. Pan HFC, Dlod CF. Differential diagnosis of eye disease. New York: George Thieme Verlag Stuttgart, 1988;355-62.

7. Cockayne EA, Krestin D, Sorsby A. Obesity, hypogenitalism, mental retardation, polydactyly: Laurence-Moon-Biedl syndrome. Q J Med 1935;4:93-120.

8. Klien D, Ammann F. The syndrome of Laurence-Moon-Bardet-Biedle and allied disease in Switzerland. J Neurol Sci 1969;9:479-513.

9. Pagon RA, Hass JE, Bunt AH, Rodaway KA. Hepatic involvement in the Bardet Biedl syndrome. Am J Med Genet 1982;13:373-81.