Rare disease

Oro-dental findings in Bardet–Biedl syndrome

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The Bardet–Biedl syndrome (BBS) is a human genetic disorder with an array of clinical features affecting many body systems. BBS is a pleiotropic disorder with mostly monogenic causes. It is also considered a primary ciliopathy syndrome. It is characterised by obesity, pigmentary retinopathy, polydactyly, mental deficiency and hypogonadism and recently a sixth feature, renal disease, has also been described. Since none of the diverse symptoms of BBS by itself is diagnostic of the disorder and many of the symptoms only become apparent over time, diagnosis of the BBS is often delayed until about 9 years of age when visual problems first appear.

BACKGROUND

Oro-dental findings in Bardet–Biedl syndrome (BBS) are rare clinical features. The aim of this paper is to highlight the disabilities caused due to multiple disorders and their effect on the proper functionality of the individual. Diagnosis of BBS can be established on the basis of both the clinical findings,¹ as well as molecular genetic testing. There is no known cure for BBS per se.

CASE PRESENTATION

A male patient aged 17 years of age reported to the Department of Oral Medicine and Radiology with swelling and bleeding from the gums in the left upper jaw region for 2 weeks. No other dental problem was reported. The patient was on antibiotics under private consultation. On anamnesis, no relevant medical history was reported by the parents except night blindness, postaxial polydactyly, subnormal intelligence and obesity. Upon eliciting the family history, a history of consanguineous marriage (Grade 1) between the parents was reported. No history regarding

other ocular or systemic disorders within family was given except for few cases of polydactyly in some relatives of their family. The patient had a brother who was of normal intelligence and built but had the inability to speak. The patient's parents were ethnically Indian in origin.

On examination, exophthalmos, hypertelorism, a convex facial profile with incompetent lips and a retruded mandible, long philtrum, anteverted nares, flat nasal bridge and strabismus of left eye were observed. There was facial asymmetry with enlarged left side and down turned left corner of the mouth. The facial nerve was normal upon examination. On intraoral examination (figures 1 and 2), inflamed interdental papillae in the maxillary left quadrant with a palatal swelling measuring approximately 6 cms in length by 1.5 cms in width and extending up to the soft palate was seen. The teeth in the left upper quadrant showed Grade 1 mobility and the gingival tissues of that area were fragile and bled upon probing. The left central incisor (21) was discoloured and showed pathological migration towards the right side. There was a cavity in

Figure 1 Frontal view. **Figure 2** Profile view.

Figure 3 (A, B) Intraoral views.

the palatal aspect of the tooth suggestive of an attempted access opening and root canal treatment left incomplete due to non-co-operation by the patient. Periodontal pockets were clinically present with respect to the left upper quadrant. The patient showed a high arched palate with anterior deep bite and increased overjet. The patient had Angle's Class II malocclusion with overjet and overbite. The tongue was normal on examination (figure 3A,B). On systemic examination – the patient was obese, short statured with a height of 126 cms. and weighed 46 kgs.(body mass index-28.97 kg/m²). He had short and stubby limbs. Postaxial polydactyly (hexadactyly) was observed on the right upper limb and both the lower limbs (figure 4A,B). The skins of the feet were cracked and dry (xerosis). Truncal obesity was observed with distension of the stomach and rhizomelic deposition of fat over the abdomen. The spleen and liver were not palpable. No evidence of skeletal dysplasia and abnormality of the spinal cord or shoulders were detected. The patient had an impaired vision with the inability to see during the night-time (nyctopia). There was slight incoherence of speech. The patient had learning disability due to mental deficiency and was slow to follow verbal commands. He was not able to do his routine chores without the help of his parents. The patient's parents also gave the history of his inability to balance and stand by himself at the age of 2 years.

Based on the above findings a provisional diagnosis of BBS was arrived at.

Figure 4 (A, B) Postaxial polydactyly.

INVESTIGATIONS

1. Intra oral periapical radiograph in relation to 21 (figures 5 and 6)
2. Orthopantomogram (figure 7)
3. Hand and wrist radiographs (figures 8 and 9)
4. Ophthalmological assessment
5. Neurological assessment
6. Ultrasonography.

DIFFERENTIAL DIAGNOSIS

- Laurence–Moon syndrome
- Cohen syndrome
- Hurler's syndrome
- McKusick–Kaufmann syndrome
- Biemond–Alstrom syndrome.

TREATMENT

Only symptomatic treatment offered as no specific protocols was available.

OUTCOME AND FOLLOW-UP

The prognosis of the patient was poor and he was followed upon for next 1 year with regular oral prophylaxis.

DISCUSSION

BBS is a rare, heterogeneous, autosomal recessive disorder characterised by polydactyly, obesity, mental

Figure 5 (A–D) Intraoral periapical views.

Figure 6 Bitewing view.

retardation, hypogonadism and renal failure. This is also called Laurence–Moon–Bardet–Biedl syndrome (LMBBS), but is different from Laurence–Moon–Biedl syndrome (LMBS).² Laurence and Moon described four cases of retinitis pigmentosa accompanied by obesity, hypogonadism and spastic paraplegia in 1866. Bardet and Biedl, separately described patients with obesity, retinitis pigmentosa, polydactyly, mental retardation and hypogonadism. The combination of these symptoms was known variously as LMBBS, LMBS and Laurence–Biedl syndrome. In 1970, Ammann recognised the presence of two distinct

Figure 7 Orthopantomogram view.

autosomal recessive disorders, which he termed Laurence–Moon syndrome and the BBS.³

In addition to the five main features of the syndrome (retinitis pigmentosa, polydactyly, obesity, genital hypoplasia and mental retardation) many other disturbances have been described.⁴

The retinitis pigmentosa is a degenerative process of the retina with typical distribution of pigment as polymorphic particles, concentrated mainly on the periphery of the retina. Clinically it manifests itself as night blindness, with the visual field gradually diminishing to near-blindness.⁵ Genital hypoplasia in this syndrome is manifested in the male as cryptorchism and hypoplasia of the penis, and in the female as hypoplasia of the uterus and labia. In adolescence there may be impotence and disturbance of spermatogenesis in the male, while in the female there may be disturbances of the menses.

Figure 8 Hands radiograph.

Figure 9 Feet radiograph.

The aetiology of the disease is thought to be associated with an autosomal recessive hereditary disturbance caused by a mutation of two genes within one chromosome (Durham 1960).

The gene products encoded by these BBS genes, called BBS proteins, are located in the basal body and cilia of the cell. Chromosome examination was not done in our case.

Dental anomalies regarded as secondary manifestations include hypodontia, microdontia, short roots and a deep palate.⁶

Features of BBS

Primary features of BBS	Secondary features of BBS
Retinal dystrophy	Developmental delay
Postaxial polydactyly	Behavioural problems
Obesity	Neurological problems
Hypogenitalism	Speech disorder
Renal abnormalities	Brachy-, syn-, or clinodactyly
Learning disabilities	Dental anomalies
	Nephrogenic diabetes insipidus
	Diabetes mellitus
	Hypertension
	Anosmia

Diagnosis of BBS is currently based on the presence of four primary features or three primary and two secondary features proposed by Schachat and Maumenee.⁷ The average age of diagnosis is 9 years, when visual problems first become apparent, but diagnosis after the age of 50 has also been reported. It has been suggested that BBS may be significantly underdiagnosed due to the slow development of clinical features.

The syndrome is familial and is transmitted as an autosomal recessive trait. chromosome 3 locus appears to be linked to polydactyly of all four limbs, whereas chromosome 15 is associated with early-onset morbid obesity and is mostly confined to the hands, and chromosome 16 represents the 'leanest'.

Learning points

- ▶ The primary features of BBS are retinal dystrophy, postaxial polydactyly, obesity, hypogenitalism, renal abnormalities and learning disability.
- ▶ The dental abnormalities of BBS are microdontia, taurodontism, short roots, deep palate and periodontal disease.
- ▶ A timely recognition with a multi-disciplinary approach to alleviate the existing conditions should be the foremost of choices in treatment planning.
- ▶ Also, a holistic approach towards the individuals and their treatment will help in improving the quality of life for such patients.

Competing interests None.

Patient consent Obtained.

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