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BARDET BIEDL SYNDROME WITH ATYPICAL FEATURES OF *HEMIMANDIBULAR HYPOPLESIA AND MICROTIA*

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ABSTRACT

Purpose. Bardet Biedal Syndrome (BBS) is a multiorgan syndrome affecting mainly the eyes, extremities, gonads & renal system etc. **Method**. Case report. **Results** The patient had common features of BBS like retinitis pigmentosa, mental & growth retardation, hypogenitalism, syndactyly and polydactyly along with atypical features like microtia, hemimandibular hypoplesia & pigeon shaped chest which are not reported in the literatures as for our best knowledge. **Conclusions.** Hemimandibular hypoplesia, microtia (small ear) & pigeon chest should also be considered as an association of Bardet Biedal Syndrome (BBS).

Key Words. Bardet Biedal Syndrome, Hemimandibular hypoplesia, Pigeon chest, Microtia

INTRODUCTION

Bardet- Biedl syndrome is a genetic autosomal recessive disease (formerly grouped with Laurence - Moon – Biedl syndrome but considered today as separate entity). It is characterized by retinal dystrophy or pigmentary retinopathy, Dysmorphic extremities (Syndactyly, brachydactyly, or polydactyly), mental retardation, truncal Obesity, hypogonadism or hypogenitalism (limited to male patients) [1, 2] .We report a case of Bardet-Biedl syndrome with the classical features along with atypical features like small ear, hemimandibular hypoplesia and pigeon shaped chest.

Case report

An 18 year old male patient born to non-consanguineous parents reported to Regional Institute of Ophthalmology, IGIMS, Patna, for night blindness. Birth history was uneventful, full-term, normal delivered. On examination, the BCVA in both eyes was 20/ 40. On examination, patient revealed hexadactyly in both upper limbs and syndactyly in lower limbs **(fig.-1A).** He had small ears (microtia), left sided hemimandibular hypoplesia and pigeon shaped chest **(fig.-1B)**. His external genitalia were underdeveloped. He was mentally retarded. Milestones were delayed as per age. He was short statured **(fig.-1A)**. The dilated fundus examination revealed macular wrinkling; mild attenuated vessels, and marked peripheral bone-spicules pigmentation **(fig-1C).** His visual field analysis couldn't be performed due to poor mental intelligence.

The patient had common features of BBS like retinitis pigmentosa, mental & growth retardation, hypogenitalism, syndactyly and polydactyly along with atypical features like small ears, hemimandibular hypoplesia & pigeon shaped chest. These features (last 3) were not reported in literatures as for our best knowledge.

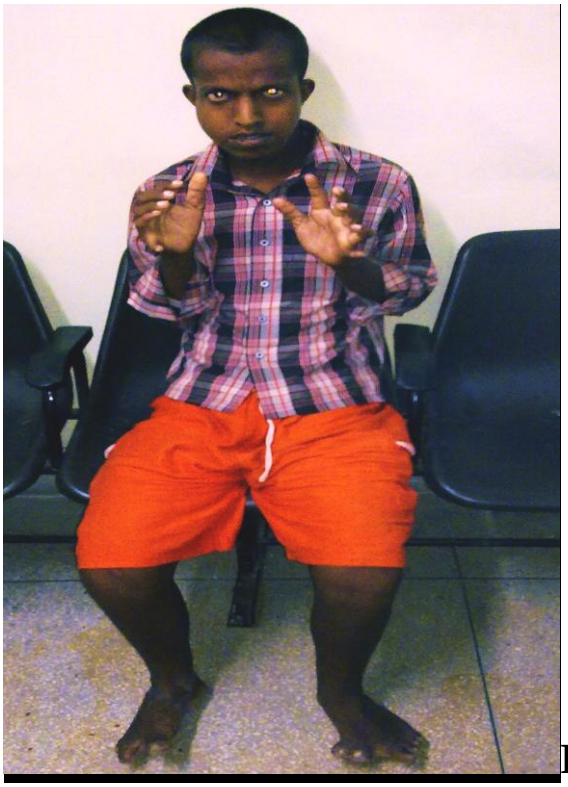

Fig.1A

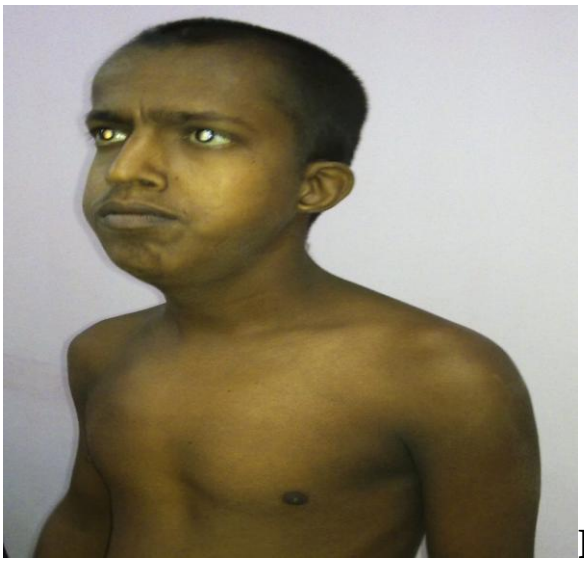

Fig.1B

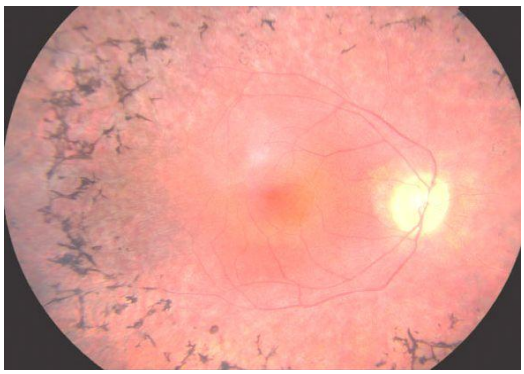

Fig.1C

DISCUSSION

Bardet - Biedl – Syndrome (BBS) is an inherited genetic condition that affects 1 in 100,000 babies born. It was described after the four doctors who described the symptoms of the syndrome. It was first described by John Z. Laurence and Robert Moon. The basic components of the syndrome were established by George Bardet in 1920 and Arthur Biedl in 1922. Laurence-moon syndrome is considered to comprise retinal dystrophy, obesity, hypogenitalism, and spastic paraparesis without polydactyly.

BBS is a combination of two syndromes, one is Bardet (described in 1920) with features of retinal dystrophy or pigmentary retinopathy, truncal obesity and dysmorphic extremities (Syndactyly, brachydactyly, or polydactyly), and other is Biedl (described in 1922) with mental retardation and hypogonadism or hypogenitalism.

The incidence has been placed at 1 in 160,000 in Switzerland. Farag and Teebi[3] have found that Bardet- Biedl is more prevalent in Arab population of Kuwait and among the Bedouin, where the estimated minimum prevalence was 1 in 13,500. Twelve genes are known to be associated with BBS: BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, and TRIM32/BBS11 and BBS12. Molecular testing is available on a clinical basis for M390R, the common mutation in BBS1 that is approximately 18%-32% of individuals with BBS.

Incomplete manifestation of the five cardinal features is the rule rather than exception in BBS. Prosperi et al.[4] estimated from previous reports that 40% to 45% of cases are incomplete. Pigmentary changes (90-100%) is the most common features followed by mental retardation (85-87%) and polydactyly (75%)[5]. The fundal involvement in BBS is an atypical pigmentary retinal dystrophy with early macular involvement. Full-field rod and cone ERG are the investigation of choice and may be abnormal as early as 14 months of age. Visual acuity, dark adaptation, and retinal vessels are normal in infancy; disk pallor and attenuated retinal vessels develop with age. Polydactyly is postaxial and may involve any or all extremities. Syndactyly or brachydactyly is present in 14.4% of patients. Hypogenitalism is present in roughly half of patients over the age of 15[6]. Vaginal atresia, urogenital sinus, uterine and ovarian hypoplasia and congenital hydrometrocolpos have been described in female BBS[7]. Infertility is particularly prominent in male BBS. Important nonocular manifestations are renal abnormalities, hirschprung's disease,

hypertension, and diabetes mellitus. Diagnosis of the BBS is based on the child's symptoms. They may be present at birth or may become noticeable as child grows. Individuals with Bardet- Biedl syndrome need visual aids and educational programs for the visually impaired. Diet, exercise, and behavioural therapies are used to manage obesity; hypercholesterolemia and diabetes mellitus are treated as in the general population. Surgery to remove accessory digits prevents functional interference and poor fitting of foot wear. Early intervention and special education address cognitive disability; speech delay or impairment is addressed with speech therapy. Hydrocolpos, vaginal atresia, or, hypospadias may be surgically corrected. Hormone replacement therapy is used to correct hypogonadism. Renal anomalies and hypertension are treated as in the general population; renal transplantation has been successful. Surveillance includes regular ophthalmologic evaluation, annual blood pressure measurement, monitoring of renal function, and regular testing for diabetes mellitus and lipid profiling.

The purpose of reporting this case is to add small ear, hemimandibular hypoplesia & pigeon shaped chest as a new atypical features of BBS

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