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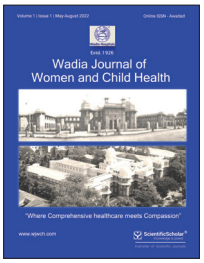

Clinical Image

A rare syndrome diagnosed in a LGA neonate!!!

Lekha Parikh[1], Tuhin Shrivastava[1]

[1]Department of Neonatology, Bai Jerbai Wadia Hospital for Children, Mumbai, Maharashtra, India.

***Corresponding author:**
Lekha Parikh,
Department of Neonatology,
Bai Jerbai Wadia Hospital
for Children, Mumbai,
Maharashtra, India.

lekha.parikh16@gmail.com

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We report a 6-day-old male late preterm large for gestational age (birth weight: 4.2 kg) neonate, who was born of a third-degree-consanguinity, to a elderly mother with history of 1 previous neonatal death. Physical examination revealed facial dysmorphism (depressed nasal-bridge, micro-retrognathia, and high-arched palate), postaxial-polydactyly in all four limbs, and hypogonadism (stretched penile length 1 cm, undescended testis) [Figure 1]. Fundoscopy was normal and auditory brainstem response showed hearing loss. Ultrasonography showed choledochal cyst and a crossed-fused ectopic kidney (5.6 × 3.4 cm) [Figure 2]. Echocardiography revealed mild concentric left ventricular hypertrophy. Clinical diagnosis of Bardet–Biedl syndrome (BBS) was suspected as this case had four major (primary) and two minor (secondary) features [Table 1]. Whole-exome genetic studies detected a likely pathogenic homozygous variant, c.886+1G>A (5'splice-site) in intron 8 of BBS9 gene.

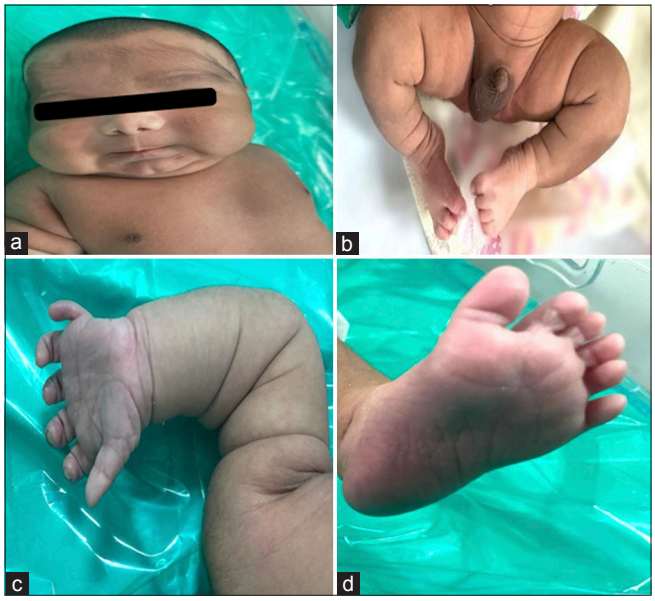

Figure 1: (a) Facial dysmorphism, (b) hypogonadism, and (c-d) polydactyly.

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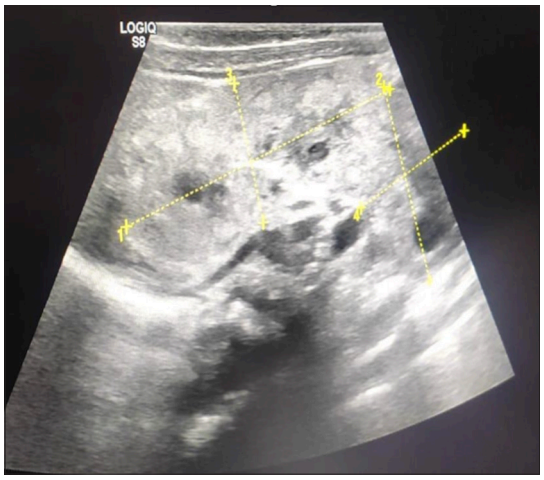

Figure 2: Crossed-fused ectopic kidney USG. Yellow dot lines are demarking and showing the crossed fused ectopic kidney. USG: Ultrasonography.

Table 1: Diagnostic criteria for BBS: At least four primary or three primary with two secondary features.

Primary features	Secondary features
• Retinal degeneration • Postaxial polydactyly • Obesity • Renal Anomalies • Hypogonadism and Genitourinary abnormalities • Learning disabilities	• Strabismus, cataracts, and astigmatism • Metabolic/Endocrine abnormalities (Metabolic syndrome, hypothyroidism) • Neurodevelopment problems (Developmental delay, behavioral problems, speech delay, ataxia, and spasticity) • Brachydactyly/syndactyly • Cardiovascular abnormalities • Liver and other gastrointestinal diseases (HD, IBD, and celiac disease)** • Anosmia/olfactory dysfunction

**HD: Hirschsprung disease, IBD: Inflammatory bowel disease, BBS: Bardet–Biedl syndrome.

BBS is a rare autosomal recessive (AR) disorder, a pleiotropic ciliopathy with dysfunction of primary cilium and loss of normal microtubular structure of non-motile cilia leading to dysfunction of various organs. There are very few neonatal cases reported for BBS. BBS cases have significant ophthalmic findings and an early onset obesity, which usually aids diagnosis at around 4–8 years of age. However, some get diagnosed even later in life with other symptoms, end-stage kidney disease requiring dialysis.

Twenty-six BBS genes have been associated with BBS till date. In Indian population, BBS3 (14%), BBS9 (10%), and BBS6 (10%) mutations are more frequent. We report a novel mutation in this case: 5' splice site variant in intron 8 of the BBS9 gene (c.886+1G>A).

Early diagnosis and multidisciplinary approach is of prime importance to ensure better outcomes of BBS cases.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.