Letter to the Editor

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Laurence-moon-biedl-bardet syndrome: An overview

Dear Editor,

We read the article "*A rare case of Bardet-Biedl syndrome (BBS)" in the esteemed "Taiwan Journal of Ophthalmology*" with great interest. Shrinkhal *et al*. reported a case of a male infant who presented with retinitis pigmentosa, obesity, postaxial polydactyly, syndactyly, hypogenitalism, hepatomegaly, renal dysfunction, high-arched palate, and mental impairment. He received the diagnosis of BBS and received a prescription of refractive correction and multidisciplinary management.[1]

BBS is historically known as Laurence–Moon–Biedl–Bardet syndrome, in which each word represents the name of the first authors to publish this genetic disorder. The cause of this syndrome is probably associated with a mutation in a gene responsible for some part of the cilia anchoring structures. As a result, this disease is considered a ciliopathy.

Here, we would like to highlight some important topics that, together with the study of Shrinkhal *et al*. could lead to a better comprehension of this rare disorder. We have done the mnemonic "BARDET" to remember the main features and "BIEDL" to some secondary findings of BBS: Blindness at night with childhood-onset due to rod-cone dystrophy also known as retinitis pigmentosa; Abnormal genitourinary tract with hypogonadism in males and complex malformations in females; Renal dysfunction that is the most common cause of morbidity and death; Disable in intellectual that is variable from mild to severe; Extra digit with postaxial polydactyly; Truncal obesity; Bowel obstruction due to Hirschsprung disease; Interventricular septum and ventricular hypertrophy associated with dilated cardiomyopathy; Ear and nose abnormalities including deafness and anosmia; Diabetes insipidus and mellitus; Liver abnormalities like hepatomegaly.[1-5]

Chandrasekar *et al*. evaluated the mutation site in 64 patients diagnosed with BBS performing polymerase chain reaction with direct sequencing. They found that the BBS10 gene mutations were mainly found in the exon two of the gene. Thus, probably, there is variability in the mutation site between the population since the result suggests a hotspot for mutations in Indian individuals, and hence, other studies with a larger population are needed to supports this hypothesis.[2]

Forsythe *et al*. recent reviewed the management for BBS. Their results revealed that the majority of the studies are preclinical with only one gene therapy, and the other two studies already being in clinical trials. Therefore, even though effective therapy is provided, it is interesting to note that BBS is disease model for future genetic and nongenetic management of rare diseases.[3]

Olson *et al*. assessed the diversity and prevalence of thoracoabdominal abnormalities in 368 individuals with BBS. Their results showed that 1.6% had an abnormality localized in the thoracoabdominal region. Furthermore, they found defects of embryonic left-right axis patterning, which suggested an early embryonic involvement of the mutation of the BBS. Thus, their findings collaborate for a hypothesis of multifactorial origin for the anomalies and the necessity of investigation for other ciliopathies such as primary ciliary dyskinesia when thoracoabdominal abnormalities are found. Furthermore, the assessment of these regions with imaging for further clinical clues if the future occurrence of other pathologies localized in these sites.[4]

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Conflicts of interest

The authors declare that there are no conflicts of interests of this paper.

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