DOI:10.24041/ejmr2021.20

BARDET BIEDL SYNDROME IN PREGNANCY: A CASE REPORT

Shweta Kumari, Shabnam Rizvi, Amrita Singh, Ayesha Ahmad, Shriya Kashiv
Department of Obstetrics and Gynecology
Era's Lucknow Medical College & Hospital, Era University, Sarfarazganj Lucknow, U.P., India-226003

Received on : 12-04-2021
Accepted on :04-06-2021

Address for correspondence
Dr. Shweta Kumari
Department of Obstetrics and Gynecology
Era's Lucknow Medical College &
Hospital, Era University, Lucknow-226003.
Email: dr053shweta@gmail.com
Contact no: +91-9430604966

ABSTRACT

Bardet Biedl Syndrome (BBS) is a rare idiopathic autosomal recessive disorder which affects multiple organs and organ systems. In India less than 15 cases have been reported so far. We present case of 41 year old third gravida who presented at 17 weeks of gestation. She had a suspicious history of delayed milestones in the previous child and ultrasound findings of echogenic kidneys, bilateral talipes, and polydactyly. Index child evaluation showed type-6 variant of BBS. Amniocentesis in the present pregnancy showed the same mutation in the fetus. The couple decided on termination of t pregnancy.

KEYWORDS: Bardet Biedl syndrome, delayed milestones, echogenic kidney, bilateral talipes, polydactyly

INTRODUCTION

Bardet Biedl Syndrome (BBS) is a rare idiopathic autosomal recessive disorder which affects multiple organs and organ systems. In India less than 15 cases have been reported. Around 21 BBS genes have been identified so far (BBS1-20, NPHP1). They are mainly involved in functioning of primary cilia. Retinal dystrophy is the most infiltrating feature of BBS. It affects around 100% of the cases. Due to this affected individual often end up becoming legally blind around 2nd to 3rd decade of life. (1)

Other important features are obesity (72-96% cases) and limb abnormalities (polydactyly, post-axial polydactyly and brachydactyly).

CASE REPORT

A 41 years old lady, presented in her third pregnancy at 17 weeks period of gestation for her first routine antenatal consultation. The pregnancy was unplanned and the couple had first degree consanguinity. Her first born, a 10-year-old boy, had clumsiness in walking and delayed milestones while her second child, a 6-year-old girl, was healthy. The first child had undergone a tendon release surgery to correct difficulty in walking, but it was not beneficial.

On examination, she was a healthy lady with a BMI of 27.7 Kg/m2. Obstetric examination showed uterus corresponding to gestational age. On ultrasound scan, we found that the fetus had echogenic kidneys (Fig. 1), bilateral talipes (Fig. 2) and polydactyly (Fig. 3). We planned a detailed evaluation of the first child by a geneticist followed by evaluation of the present pregnancy.

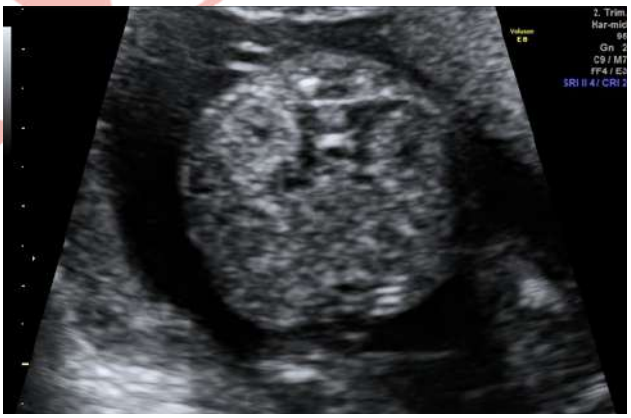

Fig. 1: Echogenic Kidney

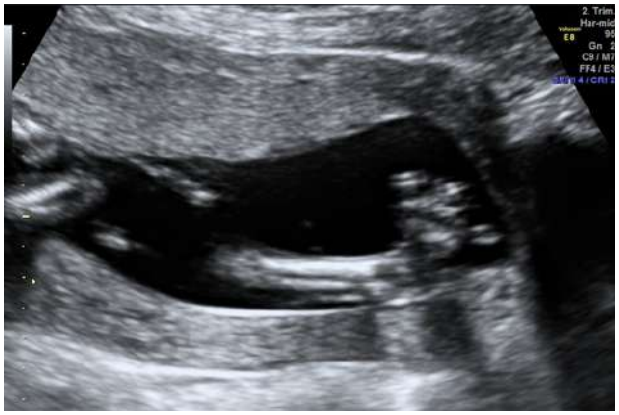

Fig. 2: Bilateral Talipes

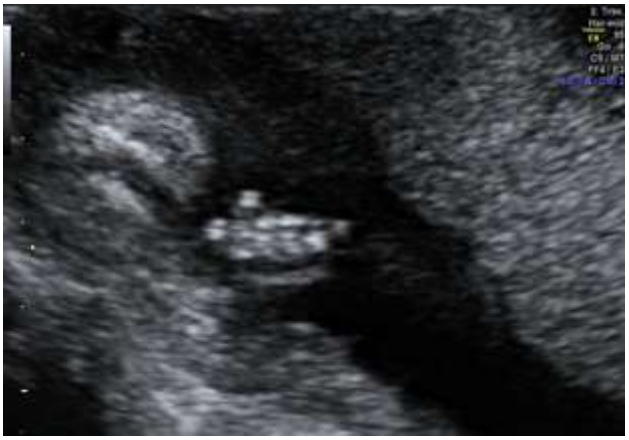

Fig. 3: Echogenic Kidney

Index child work up revealed myopia, post axial polydactyly, mild obesity and intellectual disability. A provisional diagnosis of Bardet-Biedl syndrome (BBS) was made and Clinical Exome Sequencing (CES) sent. He was found to have Type 6 variant of BBS. This was followed by amniocentesis in the present pregnancy, with samples sent for karyotyping and CES. The reports confirmed the same mutation of BBS in the fetus. The couple decided on termination of the present pregnancy.

DISCUSSION

BBS is an autosomal recessive disorder. There is scant data available from India. The presence of echogenic kidneys, polydactyly and bilateral talipes should prompt a suspicion of BBS. (2) Both bi -allelic and tri -allelic inheritance is found, although former is more common, being found in 80% subjects. (3) The overall changes of recurrence are 25%.

Similar case report by Saber et al. in (2021) (4) shows obesity, delayed milestones, acanthosis nigricans, bilateral gynecomastia as a main feature of Bardet Biedl syndrome. Another case report by Kumar S et al. shows underdeveloped genitalia ,behavioral problems & diminished vision in Bardet Biedl syndrome. In both of these studies, there is history of consanguineous marriage along with the family history of similar child. & both of these cases diagnosed late.

Exome sequencing can be done. It is an efficient as well as cost-effective technique aiding in detection of genetic mutations. While whole genome sequencing (WGS) remains expensive, CES can be utilized if a reasonable tentative diagnosis can be made on the basis of clinical features. (5-6)

The present treatment modalities of BBS focus on management of clinical features. Progression of vision loss cannot be halted. Early evaluation by a specialist team can help children by providing vision aids, mobility training and planning for future blindness. A multimodal approach can minimize obesity and complications by education, diet planning, exercise and behavioral therapies. Intellectual disability requires special education and speech therapy, started early in life. Decision for hormone replacement therapy can be considered as the child approaches puberty.

CONCLUSION

Timely diagnosis in fetus is important to plan management of the pregnancy. In our patient, index child workup had missed the diagnosis. However, a suspicious history of delayed milestones in previous child along with the ultrasound findings of current pregnancy initiated detailed evaluation of the affected child. We want to highlight the importance of a focused obstetric history taking in every antenatal woman. One should always enquire in detail about previous pregnancies and any problems observed in previous baby (delayed milestones in the present case). This will help in early detection and timely decisions regarding continuation of pregnancy.

ETHICAL CONSIDERATIONS: Patient identifiers have been anonymized.

REFERENCES

1. Forsyth R, Gunay-Aygun M. Bardet-Biedl syndrome overview. Seattle: University of Washington; 2020.
2. Baker TM, Sturm EL, Turner CE, et al. Diagnosis of Bardet-Biedl syndrome in consecutive pregnancies affected with echogenic kidneys and polydactyly in a consanguineous couple. Case reports in genetics. 2013;2013:159143.
3. Abu-Safieh L, Al-Anazi S, Al-Abdi L, et al. In search of triallelism in Bardet–Biedl syndrome. European journal of human genetics. 2012; 20(4): 420-427.
4. Saber S, Hossain MD, Alam MT, et al. Bardet Biedl Syndrome: A Rare Case Report in a Tertiary Care Teaching Hospital, Dhaka, Bangladesh. EJMED. 2021; 3(1): 11-14.
5. Ajmal M, Khan MI, Neveling K, et al. Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. Molecular vision. 2013;19: 644-653
6. Majewski J, Schwartzentruber J, Lalonde E, et al. What can exome sequencing do for you?. Journal of medical genetics. 2011; 48(9): 580-589.

How to cite this article : Kumari S., Rizvi S., Singh A., Ahmad A., Kashiv S. Bardet Biedl Syndrome In Pregnancy: A Case Report. Era J. Med. Res. 2021; 8(1): 96-97.