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Case Report

Laurence Moon Bardet-Biedl syndrome in 12-year-old Female

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Abstract

Herein, We Report A Case of A 12-Year old Female With Clinical Manifestations of Laurence-Moon-Bardet-Biedl Syndrome (LMBBS). She Has Visual and Speech Impairment Who Is the First Child of The Family. She Had Also Variations In Clinical Laboratory Tests In Her Clinical Evaluations. Renal Failure and Obesity Were Significantly Reported to Her. She Already Lives and Has Learning Disabilities and School Skills Because Her Mental Retard.

Keywords: case report; laurence moon bardet-biedl syndrome; female child; clinical laboratory tests; genetic counseling

Introduction

Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is a rare autosomal recessive inheritance disease and genetic heterogeneity, results of consanguineous marriage. Some of sign and symptoms to this disorders which are congenital ciliopathy manifesting with primary and secondary characteristics. However functional and morphological abnormalities are observed. Laurence-Moon syndrome (LNMS) is a rare genetic disease that results in a complex association of difficulties with functions that affect several different parts of bodies such brain, eyes, ears, stomach, kidneys, and skeletal organs. However, this disease is characterized by progressive blindness, obesity, and learning disabilities, and organ abnormalities. Especially in our community genetic counseling just plays a specific role about the potential genetic diseases in consanguineous marriage. [1-15]

Case presentation

A 12-year-old female patient with visual and speech impairment who is the first child of the family and is obese with physical appearance. Nearby she has impaired lying knee joints and impaired gait. She had hyperkalemia, hypocalcaemia and mild anemia with increased platelet when she refereed. She lives not only under the surveillance of a physiotherapist and but also needs care with impaired concentration and learning. His parents are far from the conditions of consanguineous marriage. However her parents have done genetic counseling. In genetic tests, molecular PCR genetic and cellular tests have been done for each of them too. She suffers from some failures and has chronic renal failure from birth. 80% of her kidneys have damaged. So attending to renal failure progresses, hereby according to chronic polycystic renal failure and renal erythropoietin products decline so the anemia was ever reported due clinical laboratory studies. However, kidney transplantation will be required. The child is studying in exceptional school and has stuttering. And she goes to speech therapy classes. Specialists physicians prescribe to her treatment choice drugs such hydrochlorothiazide, allopurinol, calcium carbonate, clostriol, metformin and gemfibrozil. During the hospitalization of the child in the Tabriz/Iran children's hospital and examinations of urologists and blood specialist and performing renal sonography, so according to doing repeated detail clinical laboratory tests and blood gas tests and other molecular PCR and cellular genetic tests the Laurence Moon Bardet-biedl syndrome are probably considered for this case report. [16-22]

Discussion

This disease was reported the communities with poor genetic counseling facilities. Laurence-Moon-Bardet-Biedl syndrome(LMBBS) is a rare autosomal recessive inheritance disorder and genetic heterogeneity disease. Primary clinical features include dystrophies, polydactyly, central abdominal obesity, kidney abnormalities, polyuria/polydipsia, hypertension, hormonal and hypogonadism abnormalities, and mental retardation, often with poor schooling skills. Secondary clinical features include developmental delay, Anemia, speech deficit, syndactyly, dental defects, ataxia, diabetes mellitus (D2M), and congenital heart defects, learning disabilities, night blindness, retinitis pigmentosa, dwarfism, deformities of the skull, left ventricular hypertrophy, hepatic fibrosis, spasticity, hearing loss, deafness, strabismus, nystagmus, osteoarthritis, hypogonadism, polysyndactyly, and obesity. Because of the high incidence of Laurence moon biedl syndrome involved between family marriages, so genetic counseling and screening the other members of the patient families are required.[1-15]

Conclusion

Laurence-Moon syndrome (LMS) predominantly included with spasticity and distal muscle weakness which is an autosomal recessive genetic

heterogeneity disease that has a complex systemic involvement too and it is considered a major differential diagnosis. In this case report patient suffers from renal failure so kidney graft is recommended to her treatment. Therefore, genetic counseling of pre marriage and before pregnancy decision are also necessary for every family affected females. Nowadays modalities of treatment such as gene therapy and targeted therapy are recommended to these diseases.

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References

1. Rao AR, Nazir A, Imtiaz S, et al. (2023). Delineating the spectrum of genetic variants associated with Bardet-Biedl syndrome in consanguineous Pakistani pedigrees. Genes. 14:404.
2. Strong A, Li D, Mentch F, Bedoukian E, Hartung EA, Meyers K, et al. (2021). Ciliopathies: coloring outside of the lines. Am. J. Med. Genet. Part A. 185:687–694.
3. Subtain Hassan, Qaisar Ali Khan1, Priyadharshini Saravanan, (2023). Megaloblastic Anemia in Bardet-Biedl Syndrome: A Rare Case Report, Clinical Medicine Insights, doi.org/10.1177/11795476231193896.
4. Ena Arora, MDa, Aleksandr Fuks, MDa, et al, (2023). Prenatal diagnosis of Bardet Biedl Syndrome: A case report,Radiology Case Reports, 326-330.
5. Alhamoud M, Alnosair G, Alhashim H, (2022). Bardet-Biedl Syndrome: A Rare Case From Ophthalmology Perspective. Cureus, DOI 10.7759/cureus.29912.
6. Khan B, Shahid A, Bin Nazir M, et al. (2019). Laurence-Moon-Bardet-Biedl Syndrome: A Case Report . Cureus, 2019, DOI 10.7759/cureus.5618.
7. Brady, P. C., Farland, L. V., Racowsky, C., & Ginsburg, E S, (2020). Hyperglycosylated Human Chorionic Gonadotropin as a Predictor of Ongoing *Pregnancy,AmJ Obstet Gynecol,* 68.e1-68.e12.
8. Christensen ST, Morthorst S, Mogensen JB, Pedersen LB. (2017). Primary cilia and coordination of receptor tyrosine kinase (RTK) and transforming growth factor β (TGF-β) signaling. Cold Spring Harb. Perspect. Biol. 9:a028167.
9. Florea L, Caba L, Gorduza EV. (2021). Bardet–Biedl Syndrome—Multiple Kaleidoscope Images: Insight into Mechanisms of Genotype—Phenotype Correlations. Genes 2021;12(9):1353.
10. Álvarez-Satta M, Cástro-Sánchez S, Valverde D. (2017). Bardet-Biedl syndrome as a chaperonopathy: dissecting the major role of chaperonin-like BBS proteins (BBS6-BBS10-BBS12) Front. Mol. Biosci. 4:55. doi:10.3389/fmolb.2017.00055.
11. Kumar A, Husain A Sr, Saleem A, Khawaja UA, Virani S(2020). Laurence-Moon-Bardet-Biedl syndrome: a rare case with a literature review. Cureus. 12:10.7759/cureus.11355.
12. Forsyth RL, Gunay-Aygun M: Adam MP, Ardinger HH, Pagon RA, et al, (2020). Bardet-Biedl syndrome overview, Seattle.
13. Mahmood SH, Khan M, Qadar LT, Yousuf F, Hasan M:(2019). A unique manifestation of Bardet-Biedl syndrome with otolaryngologic symptoms and bronchopneumonia in a one-year-old girl. Cureus. 11:10.7759/cureus.5717.
14. Anosov M, Birk R: (2019). Bardet-Biedl syndrome obesity: BBS4 regulates cellular ER stress in early adipogenesis . Mol Genet Metab. 126:495-503. 10.1016/j.ymgme.2019.03.006
15. Maria M, Lamers I, Schmidts M, et al. (2016). Genetic and clinical characterization of Pakistani families with Bardet-Biedl syndrome extends the genetic and phenotypic spectrum, Sci Rep, 6:34764. 10.1038/srep34764.
16. Basiri, M. R., Ghazi-Khansari, M, Sahhaf-Ebrahimi, F. & Alilou, S, (2021).The Importance of Molecular and Clinical Laboratory Tests in Diagnosis and Treatment of Coronavirus Disease, *Journal of Pharmacy and Pharmacology*, 232-238.
17. Rezai-Basiri, M. (2023). New Findings for Diabetes II Management, Journal of Pharmacy and Pharmacology,129-131.
18. Basiri, M. R, (2022). Hypothesis on Management of Coronavirus Disease, Journal of Pharmacy and Pharmacology, 120-124.
19. Rezaei-Basiri, M., Rezazadeh, H., Asvadi-Kermani, I., et al, (2014). Antimutagenic Effects of VitaminE on Oncology and Non-Oncology Hospital Nurses byComet Assay, Drug Res, 337-42.
20. Majid, R. B., Rezazadeh, H., Asvadi-Kermani, I., et al, (2013). Effect of Vitamin E on Uroepithelial Cells and Changes of Urinary Sediments in Oncology Hospital Nursing Personnel,*Journal of Clinical and Diagnostic Research*, 2570-2572.
21. Majid Rezaei Basiri,et al,(2018).Evaluation of Abuse Drugs and Clinical Laboratory Tests Variations in Whole Blood & Urine Samples of Abusers, *Journal of Pharmacy and Pharmacology*,2018, 69-76.
22. Majid Rezaei Basiri, (2019). Evaluation of abuse drugs tests variations in fresh biological samples of abusers, *JJ Addic Ther* 6(1): 033.

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