Case Study

Jour Cas Stu Cas Rep, 2(1), 2023, 1-3

Journal of Case Studies and Case Reports

Open access peer reviewed international indexed journal Online ISSN: 2583-4428

Content Available at www.saap.org.in

Case study on wolfram syndrome-(didmoad)

Gedda Tarun[1*], Vuriviti Pavani[2], Kanuri Sai Lavanya[2], Doddi SaijyothiPoojitha[2], Pyla Upendra[2], Baddem Bhargavi[3]

[*1,2] Pharm.D, Department of Pharmacy Practice, Avanthi Institute Pharmaceutical Sciences, Vizianagaram

[3] Clinical Research Coordinator, Actimus Bio-Vizag

Received: 01 Jan 2023 Revised: 09 Jan 2023 Accepted: 18 Jan 2023

Abstract

Wolfram Syndrome is a rare autosomal recessive genetic disorder it is a condition which is characterised by childhood onset of Diabetes mellitus, Diabetes insipidus, Optic atrophy and deafness also known as DIDMOAD Syndrome. This is an inherited condition which is mainly caused due to mitochondrial dysfunction. The mutation of WFS1 gene gets implicated in endoplasmic reticulum. A wolfram gene has recently been mapped to chromosome 4p 16.1. The best available diagnosis is childhood onset diabetes mellitus and optic atrophy. This disorder mainly affects the central nervous system. The pathogenesis is unknown. This review seeks to raise awareness of this rare case. A woman of 21years was evaluated for diabetes mellitus on following up her condition it was found that she has wolfram syndrome. Researchers are investigating on the syndrome for cure of the treatment but at present the treatment can be done by reducing the symptoms by drug management therapy.

Keywords: Wolfram syndrome, DIDMOAD, Diabetes mellitus, Diabetes insipidus, Optical atrophy, Deafness, Autosomal recessive, WFS-1 gene.

***Corresponding Author**
Gedda Tarun

doi: https://doi.org/10.37022/jcscr.v2i1.406

Introduction

Dr.Don J. wolfram was the first person who studied about diabetes mellitus and optical atrophy in four siblings in 1938. He found that the above two conditions are due to defects in genes. On further investigations he found that it is a rare autosomal recessive genetic disorder which was named after him as wolfram syndrome also known as DIDMOAD syndrome. The estimated prevalence of Wolfram syndrome type 1 is **1 in 500,000** people worldwide. Approximately 200 cases have been described in the scientific literature .Usually the onset of wolfram syndrome occurs during birth of child but it is detected during their adolescence. In early years, it is thought to be caused by mitochondrial dysfunction due to its symptoms and several reports of mitochondrial mutations. But the actual cause is endoplasmic reticulum dysfunction. Based on the researchers WFS-1 mutations alter the structure of a protein and due to this there is an incorrect pairing in aminoacids which leads to mutations. The dysfunction of CISD2 gene can cause WFS-2. Researchers convey that the actual cause of syndrome and the function of the protein are unknown.

Case Presentation

A 21years old female patient was admitted in the hospital with symptoms of abdominal pain and difficulty in swallowing of food. On examination the doctor concluded that she has a pain in the epigastric region. She has a previous medical history of diabetes mellitus, weight loss and polyurea condition (1.7lit/day). She also has a history of multiple admissions for glycemic control. The female girl has gradual diminision of vision and hearing. At the time of birth she weighed 3.75 kg and 30 kg is the present weight of the patient. On examination the patient height was 126 cm; BMI: 18.9 kgs ; Haemoglobin: 7.7 gm/dl ;Blood Urea: 10 mg/dl Pus cells: 4-6 ; Epithelial cells: 2-3 .She has a short stature with mutism and developmental delay and due to this her appearance is like a younger age. She also has

central diabetes insipidus. This case is based upon a known cause of DIDMOAD syndrome on Insulin therapy.

Discussion

After studying the patient family history, it was observed that her elder brother is also suffering from the same conditions due to some genetical mutations. So, the doctor concluded that the girl is suffering from wolfram syndrome based on the family history. Recently she was admitted in the hospital with epigastric pain. Based on this it was observed that as her age is increasing the symptoms of wolfram syndrome are being more prominent. Her brother who is 25years old but has an appearance of younger age has the same progression of symptoms of wolfram syndrome on examination. He was also suffering with urinary infections during this phase. Based on result from the pedigree analysis it was concluded that this wolfram syndrome in both children is due to consanguineous marriage.

Management and Outcome

Management of wolfram syndrome is done by treating the symptoms. Insulin can be given to treat the symptoms of diabetes mellitus and hearing aid can be used in patients with hearing loss. This patient treatment regimen for treatment of her diabetes, she was given Inj. H ACTRAPID and Inj. INSULOTARD. Stomach ulcers were treated using Tab. PANTOP and Syrup SUCRATASE. Tab. DOMPERIDONE was given to the patient during her treatment to control the adverse reactions like nausea, vomiting. Inj. OPTINEURON was administered with NS saline to counter attack the nutritional deficiency. Syrup DEXORANGE is used to the patient for treatment of anemia. Fever and some infectionsoccurred during the treatment and for that Tab. DOLO, Inj.FLUCONOZOLE, Inj. PIPTAZ was administered. As wolfram syndrome mostly cause damage to the central nervous system. The patient was given Tab. VALPROATE as a precautionary drug from epileptic conditions. Tab. VERTIN was given to the patient to prevent and treat disorders of the inner ear and for abdominal infections Tab. SEPTRAN was given. Tab. BACLOFEN, Syrup MUCAINE gel is also administered. These are some of the side effects and other complaints were observed in the patient during the drug management for wolfram syndrome. And later for few days there were no fresh complaints was observed from the patient. So, the doctor suggested to the patient to manage the drugs given for wolfram syndrome on insulin therapy.

S. NO	DRUGS PRESCRIBED	DOSE	R.O.A	NO. OF DAYS GIVEN
1	Inj. ACTRAPID	8 Units (B.D)	Subcutaneous	10 days
2	Inj. INSULOTARD	6 Units (B.D)	Subcutaneous	10 days
3	T. PANTOP	40mg (B.D)	P. O	10 days
4	Syrup SUCRATASE	1 ml (TID)	P. O	10 days
5	T. DOMPERIDONE	10 mg (OD)	P. O	4 days
6	Inj. OPTINEURON	100ml (OD)	I.V	2 days
7	Syrup DEXORANGE	2ml (OD)	P. O	6 days
8	T. DOLO	650 mg (OD)	P. O	8 days
9	Inj. FLUCONOZOLE	100 mg (OD)	I.V	2 days
10	T. VALPROATE	250 mg (OD)	P. O	3 days
11	Inj. PIPTAZ	3 gm (TID)	I.V	4 days
12	T. VERTIN	8 mg (OD)	P. O	6 days
13	T. SEPTRAN	960 mg (BD)	P. O	6 days
14	Syrup MUCAINE gel	2 ml (B.D)	P. O	6 days
15	T. BACLOFEN	5 mg (O.D)	P. O	6 days

Conclusion

DIDMOAD syndrome is caused 1 in 500000 people in the world and the causing factors for this syndrome are defect and mutation in genes at present the risk of this

syndrome is increasing and one of the treatments for this syndrome is gene therapy which takes high cost and cure of this syndrome cannot be done perfectly at present. Researchers are investigating on this syndrome and at present we can reduce the symptoms of the syndrome by drug management.

References

1. T.G. Barrett, S. E. Bundey and A. F. Macleod, "Neurogeneration and diabetes: UK Nation wide study of wolfram (DIDMOAD) Syndrome," The Lancet, Vol. 346, no. 8988, pp. 1458-1463, 1995.
2. Wolfram D, wagener HP. Diabetes mellitus and simple optic atrophy among siblings: report of four cases. Proc staff meet Mayo clin 1938; 13:715-18
3. https://www.ncbi.nlm.nih.gov › articles › PMC8949990
4. **https://medlineplus.gov/genetics/condition/wolfram-syndrome/**
5. https://www.diabetes.org.uk/diabetes-the-basics/other-types-of-diabetes/wolfram-syndrome
6. B. T. Kinsley, M. Swift, R. H. Dumont, and R. G. Swift, "Morbidity and mortality in the Wolfram syndrome," Diabetes Care, vol. 18, no. 12, pp. 1566–1570, 1995.
7. R. Medlej, J. Wasson, P. Baz et al., "Diabetes mellitus and optic atrophy: A study of Wolfram syndrome in the Lebanese population," The Journal of Clinical Endocrinology & Metabolism, vol. 89, no. 4, pp. 1656–1661, 2004
8. Chaussenot, S. Bannwarth, C. Rouzier et al., "Neurologic features and genotype-phenotype correlation in Wolfram syndrome," Annals of Neurology, vol. 69, no. 3, pp. 501–508, 2011.
9. J. Stoy, E. L. Edghill, S. E. Flanagan et al., "Insulin gene mutations as a cause of permanent neonatal diabetes," Proceedings of the National Acadamy of Sciences of the United States of America, vol. 104, no. 38, pp. 15040–15044, 2007.
10. Bespalova IN, Van Camp G, Bom SJ, et al. Mutations in the Wolfram syndrome 1 gene (WFS1) are a common cause of low frequency sensorineural hearing loss. Hum Mol Genet. 2001;10(22):2501–8. doi: 10.1093/hmg/10.22.2501.
11. Barrett TG, Bundey SE, Macleod AF. Neurodegeneration and diabetes: UK nationwide study of Wolfram (DIDMOAD) syndrome. Lancet. 1995;346(8988):1458–63. Doi: 10.1016/S0140-6736(95)92473-6.