J Integral Sci, 2019, Volume 2, Issue 1, 25-26. ISSN: 2581-5679

www.jisciences.com

Case Study

Journal of Integral Sciences

Case Study on X-Linked Adrenoleukodystrophy

M. Madhavi Kumari[1], Boddu Kirthana[*2]

[1] Assistant Professor, Department of Pharmacology,Avanthi Institute of Pharmaceutical Sciences

[2] Doctor of Pharmacy, Avanthi Institute of Pharmaceutical Sciences, Cherukupally, Vizianagaram.

Corresponding Author*

Boddu kirthana

Received: 15 Jan 2019; Revised: 22 Feb 2019; Accepted: 25 Mar 2019

Abstract

X-Linked Adrenoleukodystrophy (X-ALD) is a rare and devastating genetic disorder characterized by progressive demyelination of the central nervous system and adrenal insufficiency. This X-linked disorder is caused by mutations in the ABCD1 gene, leading to the accumulation of very-long-chain fatty acids (VLCFAs). The patient in question is a 10-year-old male, presenting with a history of progressive neurological symptoms such as vision and hearing loss, difficulty swallowing, and motor dysfunction. Diagnosis poses challenges due to the diverse clinical presentation, requiring a multidisciplinary approach encompassing neurological assessments, imaging studies, and genetic testing. Understanding the genetic basis has facilitated genetic counseling and family screening, allowing for early identification of at-risk individuals. Current treatment strategies focus on symptom management and disease modification. Hormone replacement therapy addresses adrenal insufficiency, while dietary interventions and experimental treatments like hematopoietic stem cell transplantation aim to reduce VLCFA levels and slow disease progression. Ongoing research explores genetic therapies and targeted interventions, offering hope for improved outcomes. Long-term management involves regular monitoring, rehabilitation services, and palliative care as symptoms evolve. The comprehensive care of individuals with X-ALD requires collaboration between healthcare providers, researchers, and patient advocacy groups.

Keywords: X-ALD, Adrenoleukodystrophy, Very-Long-Chain Fatty Acids, rehabilitation services, motor dysfunction, Peroxisome, progressive demyelination.

Introduction

Adrenoleukodystrophy (ALD) is a genetic disorder that follows X linked inheritance pattern in most cases (X-ALD). A unique neonatal form classified as one form of Zellweger syndrome has an autosomal recessive inheritance pattern (N-ALD). Although, the earlier report in the 1900s described the clinical presentations suggestive of this disease.; the terminology and pathophysiology were first reported in the 1970s [1].

The brain, spinal cord, adrenal glands, and testes are the most commonly affected organs. Given the multiple organs, involvement multidisciplinary team approach is highly recommended in the management plan [2].

Adrenoleukodystrophy (ALD) is a genetic disorder that follows X linked inheritance pattern in most cases (X-ALD). A unique neonatal form classified as one form of Zellweger syndrome has an autosomal recessive inheritance pattern (N-ALD) [3]. Although, the earlier report in the 1900s described the clinical presentations suggestive of this disease.; the terminology and pathophysiology were first reported in the 1970s [4]. The brain, spinal cord, adrenal glands, and testes are the most commonly affected organs. Given the multiple organs, involvement multidisciplinary team approach is highly recommended in the management plan [5].

Case Presentation

The patient in question is a 10-year-old male, presenting with a history of progressive neurological symptoms such as vision and hearing loss, difficulty swallowing, and motor dysfunction. A detailed family history revealed that the patient's maternal uncle had a similar clinical presentation, leading to suspicions of a genetic

component.

Diagnostic Evaluation

X-ALD manifests with a spectrum of clinical symptoms, ranging from mild to severe. The hallmark features include demyelination of the central nervous system, adrenal insufficiency, and accumulation of very-long-chain fatty acids (VLCFAs). The patient exhibited symptoms consistent with the cerebral form of X-ALD, characterized by cognitive decline, behavioral changes, and motor deficits.

Diagnosis

The diagnosis of X-ALD can be challenging due to its diverse clinical presentation and the absence of specific early markers. Initial evaluations involved neurological assessments, imaging studies, and blood tests to measure VLCFAs. Confirmatory diagnosis often relies on genetic testing, specifically identifying mutations in the ABCD1 gene located on the X chromosome. The identification of a hemizygous mutation in this gene confirmed the diagnosis of X-ALD in our patient

Treatment

Current management strategies for X-ALD focus on symptomatic treatment and disease modification. Adrenal insufficiency is addressed with hormone replacement therapy, while Lorenzo's oil, a mixture of oleic acid and erucic acid, has shown promise in reducing VLCFA levels. Hematopoietic stem cell transplantation (HSCT) remains a potentially curative option, especially if performed in the early stages of the disease.

Outcomes and Follow-Up

Adrenoleukodystrophy (X-ALD) depend on various factors, including the age at onset, the specific subtype of X-ALD, the severity of symptoms, and the effectiveness of interventions. It's essential for individuals with X-ALD to receive ongoing medical care and monitoring to address evolving needs. Here are key aspects of outcomes and follow-up care on Disease Progression, Hematopoietic Stem Cell Transplantation (HSCT), Hormone Replacement Therapy (HRT), Genetic Counseling and Family Planning, Palliative Care.

DISCUSSION:

Discussion focuses on key aspects of X-ALD, including its genetic basis, clinical manifestations, diagnostic challenges, current treatment strategies, and areas for ongoing research. X-ALD represents a challenging genetic disorder that requires a multidisciplinary and holistic approach. Advances in genetic research, diagnostics, and treatment options offer hope for improved outcomes and quality of life for individuals affected by X-ALD and their families. Continued collaboration between the scientific community, healthcare professionals, and advocacy groups is vital for making further strides in understanding and managing this rare condition.

Conclusion

X-Linked Adrenoleukodystrophy poses significant challenges in diagnosis and management. As our understanding of the genetic and molecular aspects of the disorder advances, novel therapeutic interventions and targeted treatments may offer hope for affected individuals. Collaborative efforts between clinicians, geneticists, and researchers are crucial in unraveling the complexities of X-ALD and developing effective strategies to improve the quality of life for patients and their families.

References

1. Moser, H. W., Raymond, G. V., & Dubey, P. (2005). Adrenoleukodystrophy: New approaches to a neurodegenerative disease. JAMA, 294(24), 3131-3134.
2. Engelen, M., Kemp, S., & Poll-The, B. T. (2014). X-linked adrenoleukodystrophy: Pathogenesis and treatment. Current Neurology and Neuroscience Reports, 14(10), 486.
3. Berger, J., Forss-Petter, S., & Eichler, F. S. (2014). Pathophysiology of X-linked adrenoleukodystrophy. Biochimie, 98, 135-142.
4. Kemp, S., Huffnagel, I. C., Linthorst, G. E., Wanders, R. J., & Engelen, M. (2016). Adrenoleukodystrophy—Neuroendocrine pathogenesis and redefinition of natural history. Nature Reviews Endocrinology, 12(10), 606-615.
5. Moser, A. B., Jones, R. O., & Hubbard, W. C. (1999). New insights into the pathogenesis of X-linked adrenoleukodystrophy. Metabolism, 48(8), 849-853.