ISSN: 0975-833X

Available online at http://www.journalcra.com

International Journal of Current Research

Vol. 16, Issue, 11, pp.30509-30510, November, 2024
DOI: https://doi.org/10.24941/ijcr.47987.11.2024

INTERNATIONAL JOURNAL OF CURRENT RESEARCH

RESEARCH ARTICLE

EARLY BEHAVIOURAL SYMPTOMS IN A CASE OF ADULT ONSET LEUKODYSTROPHY- A CASE REPORT

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ARTICLE INFO

Article History:
Received 24th August, 2024
Received in revised form
17th September, 2024
Accepted 29th October, 2024
Published online 30th November, 2024

Key Words:
Leukodystrophy, Behavioral Symptoms.

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ABSTRACT

Leukodystrophies are rare genetic disorders characterized by progressive damage to the white matter of the brain, manifesting with neurological and motor symptoms. In adults, the onset of leukodystrophy symptoms is typically delayed, with the disease being frequently misdiagnosed due to its broad symptomatology. This case report describes a unique presentation of early behavioral symptoms in an adult patient with leukodystrophy, emphasizing the importance of recognizing neuropsychiatric changes as potential early indicators. The patient, a previously healthy adult, presented initially with depressive and irritable behavior since 7 years, treated initially with antidepressants showing minimal improvement and later developed cognitive symptoms along with 1 manic episode 4 years back. Neuro-imaging and genetic testing ultimately confirmed the diagnosis, showing the utility of advanced diagnostic tools in cases of atypical leukodystrophy presentation. This case highlights the need for increased awareness of behavioral changes as possible early signs of leukodystrophy, which may facilitate earlier intervention and improved patient outcomes

Citation: Dr. Pradyumna, Dr. Madhusudhan, S. and Dr. Harshitha Handral. 2024. "Early behavioural symptoms in a case of adult onset leukodystrophy- A case report*". International Journal of Current Research*, 16, (11), 30509-30510.

INTRODUCTION

Leukodystrophies are genetic-derived diseases that primarily affect the white matter affecting both the construction and maintenance of myelin as well as the key neurological cells involved.[1]Though there is a continuum with certain leukodystrophies occurring from childhood to adulthood with milder phenotypes, certain leukodystrophies present exclusively in the adult population as described in few rare case reports. The spectrum of adult leukodystrophies is much less well understood due to their relative rarity, varied presentations and perceived diagnostic challenge due to the greater proportion of confounding mimics such as multiple sclerosis and small vessel disease. Early diagnosis is essential as several of them are potentially treatable when diagnosed early in the disease course. This case report, elaborates unusual behavioural presentation in an adult onset leukodystrophy, adding to the limited existing literature on this topic.

CASE REPORT

A 39-year-old female, 7th passed, married, home maker with no familial history of psychiatric or neurological illness and no known comorbidities presented with depressive symptoms, wandering behaviour, nervousness 7 years back which was insidious in nature gradually progressive over next two years. Initially, a trial of SSRIs was done to alleviate these symptoms, but with minimal improvement despite 1 year of regular medications. Since 5 years there is slowness of movement, slurring of speech, difficulty in walking, increasing progressively over years. She also developed elevated mood, irritability, experienced decreased need for sleep, aggressive behaviour, reduced appetite and also displayed wandering behaviour. In view of these manic symptoms 4 years back olanzapine was added and showed significant improvement in manic symptoms. Since 3 years there is progressive cognitive decline, worsening of gait, frequent falls and slurring of speech and on examination showed bipyramidal signs and gait ataxia with MMSE score of 16/30, MOCA score of 13/30. Basic blood investigations like CBC, RFT, LFT, S/E, Vitamin B12, TSH, CPK were within normal limits.HCV, HIV, HBsAg were non-reactive. MRI-Brain revealed diffuse symmetric Confluent subcortical and deep white matter T2 hyperintensityin B/L frontal, parietal, temporal and occipital lobes.

Genetic testing: Whole exome test showed mutationsin LAMB1, TIA1, HTT genes. Initially, patient was treated with SSRIs, later SNRI for anxiety and depressive symptoms along

with T. Mirtazapine. Despite one year of regular medication, there was only minimal improvement in symptoms. T. Olanzapine was effective in treating manic symptoms. Currently patient is being maintained on T. Olanzapine 15 mg. Supportive care (physiotherapy and orthopaedic care) are being provided for neurological symptoms. Patient's two daughters have received genetic counselling.

DISCUSSION

Leukodystrophies can present variably and at any age. Typically, the age of onset inversely correlates with the rate of progression and severity [2,3]. In general, adult-onset leukodystrophies have a wider phenotypic spectrum with more neuropsychiatric and behavioural symptoms. Predominant symptoms in adult-onset leukodystrophies include motor impairment, neuropsychiatric symptoms, ataxia, and cognitive deficits. In contrast to children, psychiatric symptoms and/or slowly worsening cognitive deterioration can often be the first manifestation and may precede neurological signs years in advance [4]. This case reports provides relatively a detailed description about the course of the neuropsychiatric manifestations in a patient of adult onset leukodystrophy. The psychiatric symptoms may be the early manifestation of neurological condition or may be exist as a co-morbidity. A thorough neurological examination is necessary in all psychiatric condition for early detection of a progressively emerging underlying neurological condition.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for use of her clinical information to be reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal identity.

Financial support and sponsorship: Nil

Conflicts of interest: Nil

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