

Adult Metachromatic Leukodystrophy with Hebephrenic Schizophrenia-like Symptoms: A Case Report

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ABSTRACT

Adult metachromatic leukodystrophy (MLD) is a rare and serious genetic demyelinating disorder. Very few cases of adult MLD manifesting as hebephrenic schizophrenia-like symptoms have been described till now. This is a case report of a 24-year-old female having hebephrenic schizophrenia-like symptoms for 2 years, who was later diagnosed to be MLD on imaging. This case highlights the importance of demyelinating disorders as a cause of apparent hebephrenic schizophrenia-like disorder. Psychiatric symptoms can be the preemergent markers of the neurodegenerative disorders.

Key words: Adult, hebephrenic, metachromatic leukodystrophy, schizophrenia

INTRODUCTION

A young female graduate presented with hebephrenic schizophrenia-like symptoms as the initial manifestation of adult metachromatic leukodystrophy (MLD). This clinical presentation has been reported very rarely, and only couple of cases of adult MLD manifesting as hebephrenic schizophrenia-like symptoms has been described in various literature.^[1] Adult MLD is a serious genetic demyelination disorders affecting the white matter in the central nervous system and the peripheral nerves.^[2]

CASE REPORT

This is a case report of a 24-year-old recently married BSc graduate female, who was brought by her mother for psychiatric consultation. She was hospitalized in the female psychiatric ward of our tertiary care hospital with a history of irrelevant talks, anger outbursts, violent and aggressive behavior, aimless wandering, decreased self-care, smiling to self, muttering to self, inability to perform daily activities of living, disinhibited behavior, reduced food intake, and sleep disturbances which were started 2 years back. Her symptoms worsened further over a period of subsequent 2 years. Her past history was not revealing any significant seizure disorder, neurological disorders, psychiatric disorders, or substance use in any form. Her family history was noncontributory. Premorbidly patient was well-adjusted and well-functioning as homemaker.

On mental status examination, she was conscious but cooperative to some extent only as she had irritable affect. Her reaction time was increased and she was inattentive and had minimal speech output. Her

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thought process and behavior were disorganized. Neurological examination was normal.

As this was the first episode presentation of psychotic symptoms, all the investigations were undertaken. Electroencephalogram of this patient showed diffuse slowing. Magnetic resonance imaging of the brain showed findings as T2/FLAIR hyperintense and T1 hypointense signals in B/L periventricular white matter with sparing of cortical U-fibers, suggestive of MLD (adult form).

She had earlier consultation with private psychiatrist and had received treatment with antipsychotics for previous 2 years. She had poor response to those medications. She was also tried with electroconvulsive therapy (ECT). 3 ECTs were given which she tolerated well but without any improvement. Psychotropic medicines were stopped in view of organic cause after establishing the diagnosis. Prognosis was explained to relatives.

Leukocyte arylsulfatase A, urinary sulfatide levels (tandem mass spectrometry), or genetic studies could not be done due to unaffordability of the relatives. Referral to a higher center for detailed workup and management was done. As the patient was lacking capacity to give consent for publication of the case report, with ensuring the confidentiality of the personal information, informed consent was obtained from the patient's mother.

DISCUSSION

Adult MLD is an autosomal recessive lysosomal disease characterized by demyelination of the white matter in the central nervous system and the peripheral nerves. Chromosome 22q13 is involved with autosomal recessive mutation, which causes severe deficiency of the lysosomal enzyme arylsulfatase A (ARSA) which hydrolyzes various sulfatides, including the major sulfate-containing lipids of the nervous system.^[2] The incidence of MLD is estimated to be in frequency of 1/40,000, i.e., between 2.5 cases per 100,000 population.^[3] Disturbances in cortico-cortical and cortico-subcortical connections are thought to be the cause of the psychotic symptoms in MLD.^[4] Progressive neurocognitive deterioration has also been seen.^[5,6] Replacement therapy can be a most promising therapeutic option for MLD, considering its etiopathogenesis. There is an ongoing clinical research to study HGT-1110 intrathecal enzyme replacement therapy. Hematopoietic stem cell gene therapy is also under consideration.^[7]

CONCLUSION

This report highlights the importance of demyelinating disorders as a rare cause of apparent hebephrenic schizophrenia-like disorder with a deteriorating course.

Clinical Significance

It is important to mention that psychiatric symptoms, especially psychotic symptoms, can be the important preemergent markers of the neurodegenerative disorders including MLD.^[8] Psychotropic drugs have minimal benefit in the course and outcome of the adult MLD. Further studies for other therapeutic options are warranted.

Limitations

Earlier mentioned biochemical or genetic investigations could not be done due to the financial constraint of the patient and her relatives.

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