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A CASE REPORT ON DENTATORUBRAL - PALLIDOLUYSIAN ATROPHY (DRPLA)

Neurology

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ABSTRACT

Dentatorubral - Pallidoluysian Atrophy (DRPLA), an uncommon hereditary neurodegenerative disorder is characterized by the progressive degradation of particular brain areas[1]. Numerous attempts have been made to manage DRPLA using a variety of treatment methods, which includes gene therapy and disease-modifying medicines. This is a case report of a 15 year old patient with DRPLA (ATN1 gene positive), highlighting the clinical features , diagnostic evaluation, and management strategies employed. DRPLA is a multifaceted neurodegenerative illness with numerous clinical symptoms with varying presentation[2]. Although there is now no curative treatment as of now, continuous research efforts are opening the road for potential therapeutic treatments.

KEYWORDS

ATN1 gene , DRPLA

INTRODUCTION

Dentatorubral-pallidoluysian atrophy or DRPLA, is a rare genetic illness characterized by progressive brain degeneration, resulting in symptoms such as involuntary movements, seizures , cognitive impairment and behavioral disorders. It is a neurodegenerative illness attributable to unstable CAG trinucleotide repeat in the DRPLA (ATN1 gene) on chromosome 12q12, resulting in severe brain alterations[3]. Common symptoms include progressive myoclonus, epilepsy, ataxia, cognitive decline, psychiatric problems and choreoathetosis.

Pathogenesis of DRPLA is ascribable to different molecular processes such as protein aggregation, mitochondrial dysfunction, and defective control of transcription. These changes cause neurons to capitulate further leading to typical breakdown of the dentatorubral and pallidoluysian systems. It is also important to take note that the link between continued eye blinking and anxiety, as seen in this case of DRPLA, may be different in different individuals, and not every patient with DRPLA will present with these following symptoms.DRPLA diagnosis can be difficult due to its rarity, varied presentation, and symptoms that overlap with other neurodegenerative illnesses. ATN1 gene mutation testing, neuroimaging investigations, and a full clinical evaluation together play crucial role in confirming the diagnosis and ruling out other probable diagnosis[4].

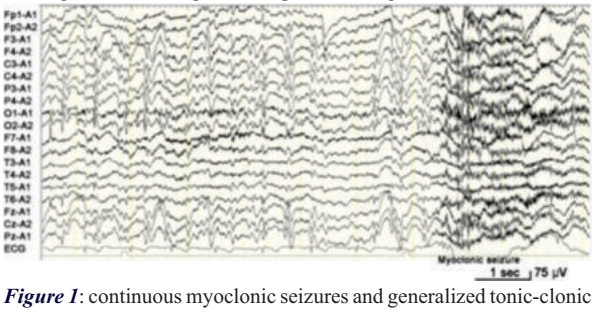

Figure 1: continuous myoclonic seizures and generalized tonic-clonic seizure following administration of PERAMPANEL (PER) **(Sources: www.googleimages.com/DRPLAeeg)**

Case Study

A 15 year old boy presented with the complaints of anxiety, sweating and continued involuntary eye blinking, which was sudden in onset without any loss of consciousness, tonic/clonic seizure episodes. Patient was aware of the entire episode after it took place and complained about it once settled. The patient experienced similar episodes (10-11 episodes) previously over the last 6 years, which was aggravated by stress and relieved on rest.

On examination, patient was conscious, oriented to time, place and person and vitals were stable. CNS examination shows normal bulk, tone and power. Patient was able to maintain eye contact, able to converse, perform day-to-day activities at decreased pace. Cardiovascular , respiratory and per abdomen examinations were normal Electroencephalography (EEG) was done, which showed mild to moderate Bilateral Cerebral Dysfunction.

Investigations

All laboratory tests were done and found to be normal. Genetic testing for ATN1 gene was positive. MRI brain was done which showed atrophic alterations in brain stem, pontine tegmentum and cerebellum. T2 weighted images showed diffuse high single intensity regions in white matter – features consistent with DRPLA findings.

Management

Management of this patient involved a multidisciplinary approach, including pharmacological therapy for seizure control, along with psychotropic measures for mood enhancement and behavioral stabilization and regular physiotherapy and occupational therapy.

Follow up

Patient was followed up for a period of 3 years with regular psychomotor evaluations, which showed significant improvement in overall symptoms and quality of life.

CONCLUSIONS

The clinical entity of dentatorubral pallidoluysian atrophy was discovered and established in Japan. As stated earlier, DRPLA is a complex neurodegenerative disorder with a wide range of clinical manifestations. Even though there isn't a cure for the disease yet, ongoing research is finding its way for the possible effective treatment approaches. The lack of a viable treatment for DRPLA is hindered by the fact that despite continuing research and clinical studies, various therapies have demonstrated minimal efficacy and detrimental effects[5].

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