ORIGINAL RESEARCH PAPER

Psychiatry

KUFOR RAKEB SYNDROME

KEY WORDS:

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ABSTRACT

Kufor rakeb syndrome (Parkinson 9) is a rare autosomal recessive , juvenile onset disease. It may present as typical parkinsonism disease or with symptoms not associated such as paraplegia, ataxia supranuclear upgaze palsy. People with this condition also experience symptoms including anxiety, learning difficulties, visual and auditory hallucinations, and dementia and depression. World wide prevalence of KRS is unknown, with only case reports/series being published. Here we report a case of the same.

INTRODUCTION

Kufor Rakeb Syndrome (parkinsons disease 9) is a rare inherited autosomal recessive juvenile onset parkinsons disease that starts to develop before 20 years of age. It is caused by ATP13A2 gene mutations.

Symptoms can include typical Parkinson's disease like bradykinesia, rigidity, and tremor, as well as other symptoms that are not typically associated such as paraplegia, supranuclear upgaze palsy, and ataxia with cerebral atrophy and iron accumulation in basal ganglia (NBIA).

People living with Kufor Rakeb syndrome also experience non-motor symptoms including anxiety, depression, learning difficulties, visual and auditory hallucinations, and dementia.

CASE REPORT

A 20-year-old female born of consanguineous parentage presented with history of tremors in the right hand and generalized weakness present at all times since 3 months.

She also reported feeling sad , reduced interaction with family and friends, having ideas that everyone is making fun of her and hearing voices talking about her with disturbed sleep since 2 weeks.

Family gave history of deliberate self harm by consuming poisonous substance 3 days ago.

Mental status examination showed the patient to be oriented, kempt with decreased psychomotor activity. Speech was relevant with increased reaction time, mood was depressed , constricted to sadness with depressive ruminations and death wishes. She had delusions of reference but denied auditory hallucinations during interview.

On neurological examination revealed dystonia, dysdiadokinesis, past pointing, ataxia, brisk reflexes, Rhombergs and plantar reflex (extensor) were found.

The following investigations were done CBC, RFT, LFT, RBS, urine examination, thyroid function test, serum electrolytes, serology tests, vitamin B12, USG abdomen and pelvis and MRI brain and were all found to be normal.

A clinical diagnosis of severe depressive episode with psychotic symptoms with parkinson plus syndrome was made and she was started on Tab. OLIMELT 10mg, Tab FLUOXETINE 20mg and Tab THP 2mg . She was compliant on medication and reported improvement and denied any psychotic symptoms on subsequent follow up.

She was then referred to a neurologist as her neurological symptoms persisted and her CK NAC levels were abnormal (606IU/l). Further investigations such as serum ceruloplasmin, plasma ammonia, plasma lactate, serum copper, 24 hour urine copper levels, fundus examination, antinuclear antibodies and thyroid function tests were done and was found to be normal.

On discussion with the neurologist a diagnosis of Kufor Rakeb disease / parkinsons disease 9 was considered and could be confirmed with genetic sequencing and screening.

DISCUSSION

A diagnosis of Kufor Rakeb syndrome requires a thorough history, as well as a complete physical and neurological examination. KRS can be suspected in individuals that start to develop atypical parkinsonism (typical symptoms of Parkinson's disease in addition to dystonia, muscle stiffness, and rapid progression) between 10 and 20 years of age. The current patient has had relatively late onset at the age of 20 years.

MRI imaging shows brain atrophy , though not seen in this case, and possibly accumulation of iron in brain structures like basal ganglia.

Only Genetic screening allows the identification of disease-causing (pathogenic) changes (mutations) in the ATP13A2 gene and lead to a definitive diagnosis.

Case reports have been published from various parts of the world, including Pakistan and Afghanistan. Till 2015, no documented cases have been published from India. Initial description of KRS was a rapidly progressive disorder with early development of significant motor disabilities. However, newer publications have indicated variable phenotypes, including intact cognition, absence of myoclonus/tremors, and slower clinical progression similar to what has been noticed in our patient.

Though the only other case reported from India (2015) had anxiety as the exclusive nonmotor symptom, KRF patients can present with a spectrum of psychological issues like in the current patient who has sever depression with psychotic sympotms.

CONCLUSION

The few studies examining the incidence of PD depression show that depressive disorders can develop at any phase in the course of PD . Frequently, affective disorders predate the onset of motor symptoms—on average, 4–6 years before the

diagnosis of PD . Once PD is diagnosed, the annual rates of newly diagnosed depressive disorders range from 1.86 to 10 % (for major depression) and, subsequently, may have a long-term or a recurrent course .

Some studies show that the age of onset, severity, duration, stage, or subtype of PD is inconsistently related to when depressive episodes commence or their severity. In this case, her neurological symptoms preceded her depressive symptoms.

Treatment of Kufor Rakeb syndrome is similar to treatment of typical Parkinson's disease and is mainly composed of a combination of two medications called levodopa (L-DOPA) and carbidopa. Other medications, such as dopamine agonists, can also be prescribed. In this case patient had symptomatic improvement on medications.

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