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PERSPECTIVE

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Diagnosis of Primary Familial Brain Calcification Disease and its Signs and Symptoms

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Description

A disorder called primary familial brain calcification is characterised by abnormal calcium deposits (calcification) in the brain's blood vessels. These calcium deposits often develop in the basal ganglia, which are deep brain structures that aid in initiating and regulating bodily movement. They are only detectable by medical imaging. Other parts of the brain might also be impacted. Movement difficulties and psychological or behavioural issues are the predominant signs and symptoms of primary familial brain calcification. These problems typically start in mid-adulthood and get worse with time. The majority of affected people have a class of movement disorders known as Parkinsonism, which includes tremors, muscle rigidity, and extremely slow movement (bradykinesia). Additional movement issues that are prevalent in patients with primary familial brain calcification include dystonia, which is the involuntary tensing of numerous muscles, uncontrollable motions of the limbs, and an unsteady stride (gait). Twenty to thirty percent of patients with primary familial brain calcification experience psychiatric and behavioural issues. These issues can include inability to focus, memory loss, personality changes, a warped perception of reality (psychosis), and deterioration in intellectual function (dementia). Moreover, those who are affected may experience dysphagia, poor speech, headaches, vertigo episodes, seizures, or urinary issues. Primary familial brain calcification affects people differently, with some experiencing no symptoms at all while others experience severe mobility and mental health issues.

Diagnosis

The serum levels of calcium, phosphorus, magnesium, alkaline phosphatase, calcitonin, and parathyroid hormone should be tested in addition to the regular standard haematologic and biochemical examinations. To rule out the presence of bacteria, viruses, and parasites, the Cere Spinal Fluid (CSF) should be inspected. It would also be worthwhile to perform the Ellsworth Howard test, which shows a 10–20 fold increase in urine cyclic Adenosine monophosphate excretion after activation with 200 micromoles of parathyroid hormone. Toxoplasmosis serology is also recommended. The most effective way to identify and gauge the severity of cerebral calcifications is using a brain computed tomography scan. The significance of this result, if any, is unknown. Increased amounts of copper, iron, magnesium, and zinc but not calcium have been recorded in the CSF.

The following requirements must be satisfied for the diagnosis:

1. The presence of bilateral basal ganglia calcification.

2. The presence of progressively dysfunctional neuronal state.

3. The lack of a different traumatic, poisonous, infectious, metabolic, or infectious cause.

4. A background that supports autosomal dominant inheritance. Although it may not be seen on normal films of the skull, the calcification is typically detected on a CT scan.

Signs and symptoms

Deterioration of speech and motor skills, seizures, and other involuntary movements are all signs of this illness. Headaches, forgetfulness, and visual problems are other signs. Parkinson's disease has traits that are also present in pressurised fluidised bed combustion. The illness often first shows symptoms between the third and fifth decade of life, while it can also happen earlier or later in life. Clumsiness, fatigue, unsteady gait, delayed or slurred speech difficulty swallowing involuntary movements or muscle cramps are typical symptoms. Several kinds of seizures are frequent. The

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symptoms of neuropsychiatric disorders, which may be the initial or most noticeable manifestations, can range from minor memory loss and attention problems to personality and/or behaviour changes, psychosis, and dementia.