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Case Report

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"Undiagnosed Neurodegenerative Syndrome – A Case Study"

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ABSTRACT

Introduction: Due to advancement in medical services, life span of individual has increased. Increased age also increases neurodegenerative problems generally in geriatric population. Neurodegenerative disorders affect many individuals across the world by increasing their disability.

Aim: Aim of the study was to evaluate undiagnosed neurodegenerative disorder in details to understand and gain insight into the selected case.

Case Report: Here reported case is affected by undiagnosed neurodegenerative syndrome at early age of 34 years. Patient was also invited by National Institute of Health, US under Undiagnosed Program for further investigations. Patient was managed by combined approach of allopathy, physiotherapy and naturotherapy.

Discussion: Patient was also suspected for diagnosis of Bipyramidal syndrome, Stiff man syndrome, Motor Neuron Disease, Neimann pick disease type C, Krebbee's disease, Chediak Higashi disease affecting the central nervous system by Neurophysician. The patient represents a rarest of rare case of Undiagnosed Neurodegenerative Program.

Conclusion: Patient can be benefited in symptoms by combined approach of medicine and physiotherapy in case of undiagnosed neurodegenerative.

Key Words: Bipyramidal syndrome, Neurodegenerative Disorder, Stiff man syndrome, Undiagnosed Neurodegenerative Syndrome

INTRODUCTION

Neurodegenerative diseases are regarded as a heterogeneous class of disorders those are identified by the progressive degeneration of the both structure and function of the central nervous system &/or peripheral nervous system.[1] Neurodegenerative disease is a span of different conditions which primarily affects the neurons of human brain. Neurodegenerative diseases are not curable. They are found with debilitating conditions those results in progressive degeneration and/or death of neurons. This causes lack of coordination (called ataxias) or mental functioning. The neurodegenerative diseases are Parkinson's disease (PD) and PD-related disorders, Alzheimer's disease (AD) and other dementias, Prion disease, Huntington's disease (HD), Motor neurone diseases (MND), Spinal muscular atrophy (SMA), Spinocerebellar ataxia (SCA).[2] Neurodegenerative diseases affect millions of people worldwide. Alzheimer's disease and Parkinson's disease are the most commonly seen neurodegenerative diseases. In neurodegenerative diseases, neurons in the brain or peripheral nervous system loose function over a period of time and die ultimately. Treatments may benefit to relieve some symptoms. There is currently no known cure or intervention to slow progress of the disease. The risk of being affected by a neurodegenerative disease increases dramatically with age.[3] Degenerative nerve diseases may affect body's activities such as talking, balance, movement, breathing, and heart function. Many of degenerative diseases are genetic. Sometimes the etiology is of medical reason, such as alcoholism, a tumour or a stroke. Other reasons may include chemicals, toxins as well as viruses. Sometimes the cause is unknown.[4]

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There are a number of clinical questions for which there are no easy answers, even with well-trained doctors. Pathological analysis is considered to be the gold standard in a wide spectrum of diseases, it cannot be applied to neurological processes.[5] Molecular diagnostics provide a powerful method to detect and diagnose various neurological diseases such as Alzheimer's and Parkinson's disease. The confirmation of such diagnosis allows early detection and subsequent medical counselling that help specific patients to undergo clinically important drug trials.[6] Functional imaging may be found useful for the early diagnosis of neurodegenerative disease. Traditionally, structural imaging is most commonly used tool to exclude alternate diagnoses and recent advancement in the magnetic resonance has shown greater diagnostic specificity.[7]

CASE REPORT

Here reported case is of 43 years old male suffering from Undiagnosed Neurodegenerative Syndrome associated with movement disorder, primary bradykinesia and lead pipe rigidity. Based on this case and a review of the literature, we discuss the pathogenic mechanisms, clinical features, physiotherapy treatment and outcome. Case had no any complaint before he was 34 years old. By occupation, patient was automobile insurance adjuster after graduating as mechanical and automotive engineer. He started developing progressive weakness and stiffness in right side of the body at the age of 34 years. After three years of onset of symptoms, at the age of 37 years, patients also started developing abnormal posturing of the right upper limb with difficulty in gait and also imbalance. At the age of 38 years, slowness was observed by patient in his activity of daily life including turning, opening door, bathing and eating etc. It was found that stiffness was increasing with time with right side of body affected. At the age of 39 years, gradually patient noticed difficulty in opening fist and fingers tend to remain clawed. While walking, he felt right lower limb stiffer and found that it was difficult to flex. These symptoms lasted for variable duration from two minutes to many hours. Symptoms had tendency to worsen by cold and stress and to disappear during sleep. It was observed during this time only that symptoms also started to left side of body. In the time where patient had no posturing problem, patient was able to continue his activity of daily life including driving activity excluding few find motor skilful movement. Patient gradually developed severe limitation in mobility and activity of daily life. Secondary impairments were present in range of motion with more affection in harmstrings muscles with soft tissue contracture bilaterally. He had bilateral ankle clonus. Up to the writing of this article, i.e. at the age of 43 years, though all above symptoms worsened markedly, patient has no any complain of cognitive impairment, visual impairment, seizures, psychiatric complaints, bulbar symptoms, drug abuse or intake, radicular symptoms in extremities or flexor spasms. Functionally, patient requires total assistance to come from supine lying to sitting position and moderate assistance to come from sitting to standing position. Patient requires maximum assistance to come to sitting from standing position. Sitting and standing balance are poor and patient has tendency to fall posteriorly in sitting position and to lean laterally on right side in standing position. Patient was bradykinetic in walking few steps with maximum assistance by any one person i.e. physiotherapist, care taker or relative. Family history was not significant for any neurological disorder. Symptoms are more during stress and fatigue and less in morning. Patient uses office chair with wheels and stair lift at home which are operated by family members.

Patient was investigated for Complete Blood Count, Creatine Phospho Kinase, Renal functions, Thyroid function, Uric acid, Blood Sugar, Vitamin B12, Liver Functions, Ultrasonography of abdomen, Human Immunodeficiency Virus, Hepatitis, CSF analysis with other blood tests periodically and were found normal. MRI Brain and MRI Spine were abnormal. MRI Brain at the time of onset showed no abnormality. MRI Brain after 5 years of onset of symptoms suggested subtle altered signal along the bilateral corticospinal tracks at the level of posterior third internal capsule, superiorly extending into corona radiate and centrum semiovale without diffusion restriction (Motor Neuron Disease?). After 7 years of onset of symptoms, Positron Emission Tomography/ Computed Tomography (PETCT) showed reduced F-Dopa localization in bilateral caudate nuclei with preserved uptake in the putamen. MRI Spine revealed posterior disc bulge and osteophytes formation at C3-4 and C4-5 level. Electrophysiological studies were done repeatedly. First study, immediately at the time of onset of symptoms, was found normal. Second study, after 15 days of onset, concluded presence of fibrillations and fasciculations in distal right upper limb muscles. Third study, after 2 years 2 months of onset of symptoms, showed occasional fasciculations in right deltoid with presence of severe neurogenic pattern in recording of voluntary activity. Colour Doppler was suggestive of Trivial Tricuspid Regurgitation.

Patient was invited by National Institutes of Health Clinical Centre, Bethesda, Maryland, US under Undiagnosed Disease Program. Patient was presented to this centre with presence of significant stiffness with characteristics of lead pipe rigidity, hyper-reflexia, bradykinesia, dysarthria and past history of apparent dystonia. He has significant impairment in his activity of daily life. He also has problem in dysphagia and coughing on administration of thin liquids. Patient desires some mobility and feeding himself independently. Possibility of dystonia, secondary Upper Motor Neuron lesion or Stiff limb syndrome was expected.

For medical management, at the time of onset of symptoms, patient has completed Vitamin B12 injection course. Medical management included Valium initially for stiff muscle syndrome but to its insignificant effect, it was stopped due to its long term issues. Sinemet was also tried but it did not give any relief. This case was discussed in clinical meeting by Neurophysician as undiagnosed case and it was decided to continue Syndopa Plus 125 mg ½ bd, Synaptol mg bd. Patient is currently on management by use of medicines Baclofen 10 mg daily, Clonazepam 0.25 mg everyday and Sinemet every day.

Findings of physiotherapy assessment suggested rigidity of all four limbs affecting right side body more than left side. When compared unilaterally on either side, lower limbs are more rigid than upper limbs. Patient had no any subjective sensory symptom. Balance impairments were highly significant in the patient. Patient is unable to maintain sitting and standing balance independently. Patient occasionally complained of backpain and left elbow pain which appears occasionally and disappear by medicines, ice packs and rest. Goal of physiotherapy treatment is to maintain the present condition of patient with an effort to prevent complications which are possible to develop due patient's present physical condition. Physiotherapy is directed towards giving active assisted movements to all four limbs, core strengthening and maintaining hygiene by chest physiotherapy. Patient was also taking naturotherapy for relief of symptoms.

DISCUSSION

The patient represents a rarest of rare case of Undiagnosed Neurodegenerative Program. Patient was also suspected for diagnosis of Bipyramidal syndrome, Stiff man syndrome, Motor Neuron Disease, Neimann pick disease type C, Krebbee's disease, Chediak Higashi disease affecting the central nervous system by Neurophysician. The patient is selected for the study as proper medical management along with appropriate physiotherapy showed no significant improvement in patient's quality of life by increasing movement potential. There are very few researches available for role of physiotherapy in this type of diagnosis and other related associated abnormalities so more numbers of researches are needed to be done with long term duration. This can also help for creating awareness about this type of patient, combination of allopath, physiotherapy and alternative therapies may give symptomatic relief even though the condition itself is not curable. There is no current evidence-based standard of care for patients with Undiagnosed Neurodegenerative Disease and no research was found.

CONCLUSION

From present case study, though it is not proved that neurodegenerative diseases can be cured or can be slow down in progression but it is concluded that patient with neurodegenerative disease can be benefited in symptoms by combined approach of medicine and physiotherapy in case of undiagnosed neurodegenerative syndrome. Combination of Short-Wave Diathermy and Mobilization may be effective in reducing pain and increasing range of motion wherever applicable.[8]

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Declaration by Patient's relative:

Written consent was given by patient's relative.

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None

REFERENCES

1. Diseases n, Hickman R, O'Shea S, et al. Neurodegenerative diseases - Latest research and news | Nature [Internet]. Nature.com. 2022 [cited 9 January 2022]. Available from: https://www.nature.com/subjects/neurodegenerative-diseases
2. What? | JPND [Internet]. JPND | Neurodegenerative Disease Research. 2022 [cited 9 January 2022]. Available from: https://www.neurodegenerationresearch.eu/what/
3. Neurodegenerative Diseases [Internet]. National Institute of Environmental Health Sciences. 2020 [cited 2 April 2020]. Available from: https://www.niehs.nih.gov/research/supported/health/neurodegenerative/index.cfm
4. Neurodegenerative Diseases | MedlinePlus [Internet]. Medlineplus.gov. 2020 [cited 2 April 2020]. Available from: https://medlineplus.gov/degenerativenervediseases.html
5. Gómez-Río M, Moreno Caballero M, Manuel Górriz Sáez J, Mínguez-Castellanos A. Diagnosis of Neurodegenerative Diseases: The Clinical Approach. Current Alzheimer Research. 2016;13(5):469-474.
6. Agrawal M, Biswas A. Molecular diagnostics of neurodegenerative disorders. Frontiers in Molecular Biosciences. 2015;2.
7. Stoessl A. Neuroimaging in the early diagnosis of neurodegenerative disease. Translational Neurodegeneration. 2012;1(1).
8. Parmar J., Chhatlani R., Kakkad A. Study to Find the Effect of Short-Wave Diathermy, Cervical Traction and Mobilization on Pain and Range of Motion in Acute Locking of the Cervical Spine - An Interventional Study. 2022.