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Many faces of *DCTN1* (Dynactin) gene mutation in neurodegenerative diseases

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A 45 year old working lady presented to us with bradykinesia for six months, accompanied with difficulty in walking for four months. Six months ago, the patient started feeling clumsy while doing house hold work and her movements became slower as time passed by. Four months ago, she started to have difficulty in walking which gradually aggravated. Since onset, she was depressed, and experienced sleep related behavioral issues but never lost weight. Her Mother had similar symptoms but was on anti-parkinsonian drugs. P/E: increased muscle tone in all 4 limbs, right>>left with reduced right arm swing, with masked type facies. In view of positive family history, Parkinsonism symptoms, depression/apathy patient was diagnosed with definite PS (Perry Syndrome) supported by international diagnostic criteria. PSG showed airflow restriction and hypoventilation using apnea hypopnea index. Genetic test was performed which confirmed novel point *DCTN1* gene mutation. Patient was started on anti-parkinsonian agents, anti-depressants and clonazepam and her symptoms got somewhat better. It can be conclusion that we have diagnosed the first Asian case of a PS with a novel point mutation p.G67S of *DCTN1* gene in exon 2 not reported yet. Our observation suggests that patients/family members may not present with all the cardinal features of PS but still it has to be ruled out with gene testing mainly because of two reasons: (1) An early timed diagnosis can significantly modify the progression of disease and (2) improve quality of life by use of diaphragmatic pacing and can prevent life-threatening episodes of acute respiratory failure and eventually death.

Biography

Rajib Dutta is currently a pursuing Post-graduation in Neurology in China with MRCP UK London. He has completed his Diploma in Emergency Medicine and Critical Care from Royal College in UK and Diploma in Clinical Neuropsychology.

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