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Many faces of DCTN-1 (dynactin) gene mutation in neurodegenerative diseases

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Axonal transport machinery is central to neuronal health and survival, with dysfunction implicated in several neurodegenerative disorders including AD, FTLD, MND/ALS and PD and PD plus syndromes, HMN 7B and perry syndrome all associated with dynactin pathology. 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 faces. In view of positive family history, parkinsonism symptoms, depression/apathy patient was diagnosed with definite PS (Perry Syndrome) supported by international diagnostic criteria. To confirm PSG showed airflow restriction and hypoventilation using apnea hypopnea index with no respiratory acidosis in ABG. Genetic test was performed which confirmed novel point DCTN 1 gene mutation. Patient was started on anti-parkinsonian agents, anti-depressants and clonazepam and her symptoms got somewhat better. 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 in literature 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) Early timed diagnosis will lead to early symptomatic treatment which 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 a Postgraduate Neurology Trainee in China with MRCP UK London, has completed his Diploma in Emergency Medicine and Critical Care (Royal College UK), Diploma in Clinical Neuropsychology (UK), Pediatric Neurology certification BPNA (UK, ongoing).

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