Glucose metabolism and dopamine PET correlates in a patient with myotonic dystrophy type 2 and parkinsonism

We describe a 72 year old woman who presented with a 5 year history of progressive gait uncertainty, frequent sudden falls, and difficulty rising from a chair, associated with fatigue. She noticed some degree of distal weakness in her arms when carrying weight compared with previous years. She had no complaints of pain or cramps but noticed stiffness and ‘locked’ legs when walking. She denied cranial nerve problems and had no general systemic complaints. Past medical history was significant for hypertension. Family history was unremarkable.

Examination revealed normal vital signs and intact cranial nerves. Slow and slightly slurred speech was evident. Her trunk was forcefully bent forwards in a camptocormic attitude, which could be corrected by passive extension of the trunk. Her tone was increased with rigid wrist and hips and brisk (3+) reflexes. No nystagmus was observed. Her gait was slow, and worsened during the day and over time to the extent that it limited everyday activities and often culminated in sudden falls. Pendular movements were reduced on the left. She had mild weakness in the shoulder abductors (4), finger flexors (4.5), hip flexors (4), and ankle dorsiflexors (4). Sensory examination and coordination were normal.

Routine blood investigation was unremarkable except for red blood cells (3.84 x 10^12/l), haemoglobin (11.2 mg/dl) and haematocrit (34%) in the low normal range. Haemaglobinins were 7% of total protein levels. Electrocardiography revealed first degree arteriovenous block. Brain computed tomography scan demonstrated diffuse white matter vascular hypodensity and cortical atrophy. A diagnosis of parkinsonism was made. Treatment with Sinemet CR tablets (carbidopa/levodopa 25/100 mg twice daily) was started. However, despite treatment, the patient had not improved 5 months later. Muscle pain was present. Repeat examination showed severe neck flexor weakness.

Patient details are published with consent.

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Competing interests: none declared

References
1. Diederich NJ, Goetz CG, Stebbins GT. Repeated visual hallucinations in Parkinson’s disease: are these symptoms a sign of an underlying medico-psychological disorder? Mov Disord 2005;20:130-40.

Figure 1 Statistical parametric maps of 18F-FDG PET showing significant metabolic reductions (p<0.05 corrected) bilaterally in superior temporal gyrus and parietal operculum (Montreal Neurological Institute stereotactic coordinates of local maxima for suprathreshold clusters x, y, z = 64, 28, 10, Z = -6.01, -62, -22, 2; x, y = 5.26, and thalamus (18, -22, 14, Z = 5.06, -8, -16, 12, Z = 4.23), superimposed on the T2 weighted MRI of the patient. The areas indicated in shades of yellow and red both indicate areas of hypoperfusion; yellow represents areas of maximum hypoperfusion.
Brain magnetic resonance imaging (MRI) demonstrated diffuse white matter lesions consistent with chronic hypoperfusion. The patient then underwent positron emission tomography (PET) studies with different tracers: $^{18}$F-FDG PET showed a reduction of glucose metabolism bilaterally in the superior temporal gyrus, parietal operculum, and, noteworthy, in the posterior thalamus (fig 1). The presynaptic dopamine reuptake as measured by $^{11}$C-B-CIT-FE$^2$ and the postsynaptic D2 receptor density as measured by $^{11}$C-raclopride$^3$ were both within the normal range values, thus revealing the integrity of the nigrostriatal pathway.

DISCUSSION

Our results confirm initial and more recent reports that extrapyramidal signs of different degrees may be present in DM2,$^3$ yet emphasise that the diagnosis of DM2 is still often one of exclusion and is frequently unrecognized. Despite the growing number of reports describing the concomitant findings of myotonic dystrophy and extrapyramidal signs, the frequency and significance of this association is still unclear. Still less clear is the pathophysiology of extrapyramidal involvement and its therapeutic implications.

We describe a patient having camptocormia, bradykinesia, reduction of pendular movement and rigidity, suggesting features of Parkinson’s disease (PD). This diagnosis was only subsequently excluded when there was no response to levodopa treatment, weakness and ‘locking’ worsened, and pain became a dominant symptom. Elevated CK and myotonic discharges led to the clinical diagnosis, subsequently confirmed by genetic analysis. Rigid spine syndrome was considered as a differential diagnosis because the bent spine attitude may result from weakness of the paraspinal and abdominal muscles, as in this patient. In addition, CK may be elevated and EMG may show spontaneous activity and a myopathic pattern. However, inflammatory signs on muscle biopsy are prominent in focal myositis, while other extrapyramidal signs such as those present in our patient are usually absent.

Although only carried out in a single patient, our PET studies demonstrate that parkinsonian features in DM2 are not related to neurodegenerative processes of the nigrostriatal system, such as in PD. Instead, they may be related to hypometabolism in the posterior thalamus. Studies on a larger group of patients with DM2 having extrapyramidal signs are needed to confirm these preliminary findings demonstrating the integrity of the dopaminergic system, and the therapeutic and prognostic implications.

**REFERENCES**


**Dementia with Lewy Bodies and Parkinson’s disease dementia**


Dementia with Lewy Bodies (DBL) is a rare disorder? It was, but not anymore! Following the publication of diagnostic criteria for DBL in 1996, research on this disorder witnessed a true explosion and the number of papers published each year increased by ten-folds. The wealth of knowledge that has been accumulating over the past 10 years on different aspects of DBL is now compiled in an easy-to-read volume. The book is systematically designed—an insightful first chapter on historical aspects is followed by a general overview on clinical spectrum of conditions which share Lewy bodies as their common pathological denominator and their classification. Authoritative sections on various aspects of the disease pursue including epidemiological, individual sections on cognitive, behavioural, somatic neurological and autonomic features, as well as the natural course of the disease, findings in neuro-imaging, underlying pathology and neurochemical deficits, and pharmacological and non-pharmacological treatment approaches. Some chapters provide comparison to a closely related disorder, dementia associated with Parkinson’s disease, which is at the end also covered in a chapter of its own, along with a chapter devoted to differentiation of DBL from other disorders that may cause dementia and parkinsonism. Most sections are exhaustive with extensive coverage of the literature. Especially sobering is a first-hand account of how the disease is experienced by a caregiver, which would help clinicians to understand the real difficulties and set management priorities.

It was timely that the large amount of data that has been produced in the past decade was put together in a single volume, especially so as this has been largely missing. This is an excellent piece of work that will fill that gap, with chapters written by authors who have been in the forefront of research in this field. The timing is also fitting as revised diagnostic guidelines on DBL have just been published. This book will be a useful reading as well as an excellent source on the large amount of literature for those who have an interest in dementia, movement disorders and the border zone between them.

**BOOK REVIEW**

M Emre

**NOTICE OF RETRACTION**

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