



The PSP Association's International Medical Workshop 7th July 2009

ABSTRACT

Title of Talk: ATYPICAL PRESENTATIONS: HOW TO DIAGNOSE

Part 1: Speaker(s) details	
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Part 2: Abstract (Maximum 400 words) Please make your abstract easy to understand as it will appear on our website and will be read by people with PSP and their carers who are not scientists but who will want to understand your work and what it means for them.

Making a clinical diagnosis of Richardson's syndrome relies on the skilled physician. The diagnosis accurately predicts the clinical course of typical PSP for the patient and also the underlying pathology in the brain. The diagnosis in this clinical context should be readily recognisable and requires little in the way of confirmatory tests. More than a third of patients who end up having PSP pathology, do not develop such a recognisable clinical syndrome. In these cases the diagnosis is many times more difficult, and patients often experience a change in diagnosis over time or vague, general diagnostic phrases such as "atypical parkinsonism". Nevertheless when viewed over a period of time the clinical syndromes of PSP-parkinsonism (PSP-P), pure akinesia with gait freezing (PAGF) and apraxia of speech (PSP-AOS) can be identified by the astute neurologist, and used to infer a clinical prognosis and also an underlying pathology. In the early stages of the disease, and at least for the first few years, patients with PSP-P will resemble others with Parkinson's disease. Unlike in classic PSP (Richardson's syndrome) patients with PSP-P will often have tremor, improvement from medications, asymmetrical onset and normal eye movements in the early stages of disease and falls are less of a problem. Patients with PAGF also contrast sharply with those who present with Richardson's syndrome. Those with PAGF do not develop eye movement abnormalities within the first 5 years of disease, and cognition is intact. These patients do experience an insidious deterioration of their gait, with a tendency to freeze and find it difficult to take a stride. In most cases patients also developed softness of voice and small handwriting. Medications have no effect on the walking, and it slowly deteriorates over more than a decade. Others can initially developed apraxia of speech (troubles with fluency of speech) that can evolve for many years before any movement abnormalities are seen. In these difficult to diagnose scenarios adjunctive or diagnostic tests would be most helpful for the clinician. Some imaging studies may be helpful, and in particular cardiac MIBG appears to be most promising, and the future possibility of PET scanning with tau-ligands may be helpful. Clinical and electrophysiological tests have not been demonstrated to reliably predict underlying tau-pathology better than clinical diagnosis, although tests of olfactory function may have a role. Biomarker assays probably hold the greatest chance of reliably identifying underlying PSP-tau pathology, and separating PSP-P from Parkinson's disease, PAGF from vascular disease and PSP-AOS from other dementias. Spinal fluid protein analysis has been shown to achieve some of these goals.