TITLE: The History of the Diagnosis of MDJ in Australia.

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**ABSTRACT:**

I am a medical practitioner who was employed by Menzies School of Health Research to live and work on Groote Eylandt in 1990 and 1991 to investigate the Motor System disorders of East Arnhem Land as a research topic for a PhD thesis. This is the story of the diagnosis.

The people of Angurugu on Groote Eylandt were aware that one man, born in approximately 1901, developed unsteady walking in middle age. On the adjacent mainland in North East Arhnhem Land, the anthropologist professor Janice Reid had been told of several people with motor system impairment during her field trips in the 1970s.

The term “Groote Eylandt Motor Neurone Disease” began to appear in Northern Territory health department records in the early 1970s. Kiloh *et al.* (1980) published a paper describing two syndromes , one with childhood onset involving muscle wasting, and anther of adult onset with cerebellar and upper motor neurone signs, though causation and precise diagnosis were uncertain.

Manganese had been mined on Groote Eylandt since 1965 and some investigators had raised the possibility that the cause of the motor system abnormalities may be manganese toxicity (Cawte 1984, 1989). Blood tests of a wide sample of East Arnhem people showed no association between motor system disorders and blood manganese levels, and neuropsychological testing showed no correlation of blood manganese levels with hand steadiness or rection time.

During the two years of my residence on Groote Eylandt, several more people began to manifest symptoms of ataxia and motor weakness, suggesting an autosomal dominant pattern of inheritance for this condition. The presentation of two other conditions, one of ligament laxity and another of peripheral muscle wasting was sufficiently different to propose alternative diagnoses.

Neuropathological examination of one case dying with the ataxic condition was consistent with Machado Joseph Disease, (MJD) Burt *et al.* (1993). With subsequent assistance from Prof Garth Nicholson, the gene for MJD was confirmed in four North East Arnhem Land families , Burt *et al.* (1996).

Clinical management of MJD in North East Arnhem Land and ongoing research into the causation of MJD progresses locally and internationally.

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