faecal incontinence (n = 4) or erectile dysfunction (n = 3; all male patients). Anorectal electrophysiology was performed in two patients, both consistent with bilateral pudendal neuropathies (Web table 1). MRI studies (fig 1) showed: scattered cerebral white matter hyperintensities in all three patients in whom this test was performed; asymmetrical left frontotemporal atrophy consistent with clinical FTD and an atrophic cord especially around the conus in II:18; and fatty change and atrophy of the gluteal muscles and low muscle volume in the quadriceps and hamstring groups in III:14. A muscle biopsy from II:14 is shown in online fig 2.

DISCUSSION
IBMPFD presents as a multisystem disorder affecting brain, skeletal and cardiac muscle, spinal cord and bone. The R155H mutation, an arginine-to-histidine substitution in the N-terminal CDC48 domain of the VCP protein, results in loss of VCP function and is found exclusively in individuals affected with IBMPFD. Clinical reports of this disease are scarce. Family members usually presented with proximal weakness, progressing to wheelchair disability and premature death. The average age of muscle weakness onset, as a familial average, was 34, contrasting with previous studies (45 and 57). The dementia frequency within this pedigree was 44% compared with 30%, 70%, and 100% in other studies. As some patients may have prematurely died of myopathy- or cardiomyopathy-related complications, it was difficult to assess FTD penetrance.

Four out of five investigated members of this pedigree had echocardiographic features of cardiomyopathy. This is the first clinical description of cardiomyopathy in a molecularly confirmed VCP mutation. Recent postmortem findings have found a dilated atrioventricular node and hypertrophic cardiomyopathy in one postmortem finding. A dilated and hypertrophic cardiomyopathy in one postmortem finding has found a dilated atrioventricular node and hypertrophic cardiomyopathy in one postmortem finding. Clinical reports of this disease are scarce. Family members usually presented with proximal weakness, progressing to wheelchair disability and premature death.

Brain MRI demonstrated a mild excess of white matter abnormalities in all three examined. Right temporal lobe atrophy and cord atrophy were seen in an older patient, correlating with her clinical FTD. This is in agreement with one other MRI brain study of IBMPFD, while another described progressive cerebral atrophy with prominent callosal and frontal white matter loss. Spinal cord atrophy is a previously undescribed feature of IBMPFD and is consistent with pathological findings of spinal cord inclusion bodies. A recent report describes MRI muscle findings in IBMPFD of “fatty degeneration” throughout predominantly proximal muscle groups, in agreement with current findings.

Members of this pedigree has been previously given other diagnoses including various muscular dystrophies and spinal muscular atrophy. MND has been diagnosed in some patients because of derangement on neurophysiology (online table 1). In conclusion, IBMPFD is a multisystem disorder, which should be considered in the differential diagnosis of autosomal dominant neuromuscular disorders, especially when there is a prominent history of dementia or “MND.” Sphincter involvement is a likely associated clinical feature of the disease.

REFERENCES

CORRECTION
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M P T Lunn, H J Willison. Diagnosis and treatment of inflammatory neuropathies (J Neurol Neurosurg Psychiatry 2009;80:249–58). There is a dosage error in this paper. In the last paragraph on page 255 the dose of prednisolone should be 1mg/kg not 1g/kg as printed.