

Amyloid neuropathy: Five year retrospective case review at a tertiary centre

Priya D Shanmugarajah¹, Sajitha C Weerasinghe¹, Agam Jung¹

¹. Leeds General Infirmary, Great George Street, Leeds, LS1 3EX, United Kingdom

Background

- Nerve biopsy has shown to improve the diagnostic yield in patients presenting with peripheral neuropathy¹
- We describe two distinct cases of amyloid neuropathy identified from a five year retrospective case and biopsy review at a tertiary neurosciences centre

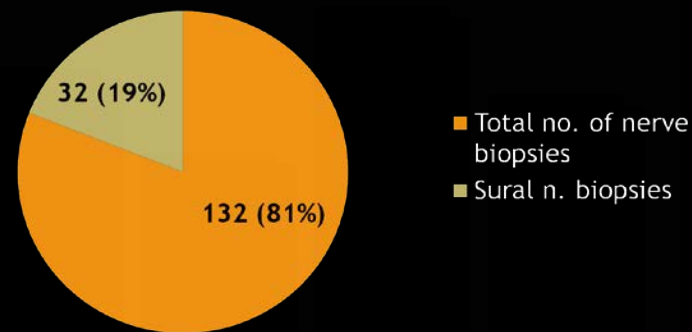
Objectives

- To analyse biopsy proven cases of amyloid neuropathy diagnosed between 2011-2016
- To assess the role of sural nerve biopsy as a diagnostic tool for peripheral neuropathy by comparing clinical and biopsy proven diagnosis

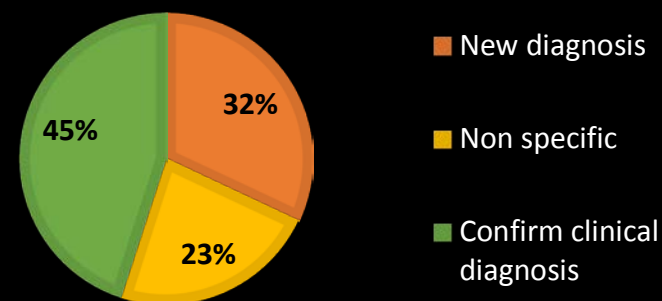
Method

- We identified all nerve biopsies performed at Leeds Teaching Hospitals NHS Trust between 2011 -2016 using the histopathology data base
- We examined the subgroup of sural nerve biopsies to compare clinical with biopsy proven diagnosis

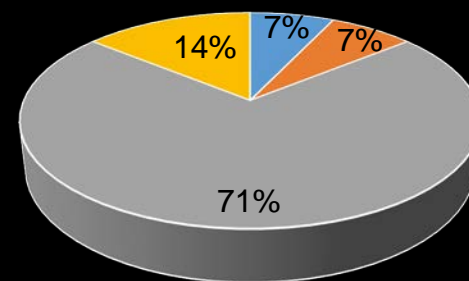
Results



Clinical vs biopsy diagnosis



Biopsy diagnosis



■ Toxic ■ Inflammatory ■ Vasculitis ■ Amyloid

Two distinct cases of amyloid neuropathy were confirmed on biopsy

	Case 1 (65 y, male)	Case 2 (67 y, male)
Background	Lambda monoclonal gammopathy of undetermined significance (MGUS)	Bilateral carpal tunnel syndrome Cardiac failure
Symptoms	Sensory impairment up to knee, foot drop, constipation, urinary retention, postural hypotension	Cold in warm weather, foot drop, dysphagia, constipation, postural hypotension, No family history
Neurophysiology showed axonal sensory motor polyneuropathy		
Sural nerve biopsy showed apple green birefringence on Congo red stain confirming clinical diagnosis of amyloid neuropathy		
Final diagnosis	Systemic light-chain (AL) amyloidosis confirmed on immunofixation electrophoresis	Genetic testing confirmed transthyretin-T60A-variant of familial amyloid polyneuropathy
Symptom onset to diagnosis	5 years	8 years

Both patients were symptomatically managed and died within 10 years of symptom onset

Conclusion

- Sural nerve biopsy is a useful diagnostic tool to confirm or alter the clinical diagnosis in peripheral neuropathy and thus has management implications
- We suggest a low threshold for nerve biopsy to guide diagnosis and treatment

Reference

- Gabriel C M et.al. Prospective study of the usefulness of sural nerve biopsy, J NeurolNeurosurg Psychiatry 2000 ; 69:442-446