Background Hereditary transthyretin amyloidosis (ATTR) is associated with progressive peripheral neuropathy, cardiac, gastrointestinal and autonomic failure due to dominantly inherited transthyretin mutations causing amyloid deposition. The neuropathy phenotype is less well described than cardiac manifestations.

Methods Detailed clinical neurological and electrophysiological data were collected on all patients. Follow-up data was available on a subset.

Results Thirty-four patients (mean age=62 years; 70.6% male) were assessed at least once; 19 (55.8%) had serial examinations, mean follow-up: 1.9 years. The genetic breakdown was 38.2% T60A, 14.7% V30M, 8.8% I107F, 5.9% V122I, 32.4% individual mutations; 76.5% UK and/or Irish ancestry. 44.1% were treated (Diflunisal or liver transplant). 26.5% presented with neuropathy; 70.6% had neuropathy on follow-up. A length-dependent, axonal, sensorimotor neuropathy was typical; 6.7% had patchy onset, 10% had demyelinating features. Mean MRC score at presentation was 62.5 with a mean reduction of 2.7 points/year and a trend to slower deterioration in the treated group (2.4 versus 3.0 points/year; p=0.77).

Conclusion The detection of measurable annual changes in MRC score is encouraging but monitoring of sensory deficits should also be quantifiable. This small but representative study mirrors difficulties observed in recent treatment trials regarding sensitivity of current outcome measures.
NEUROPATHY PHENOTYPE IN HEREDITARY TRANSTHYRETIN AMYLOIDOSIS

Aisling Carr, Ana Pelayo, AD Wechalekar, CJ Whelan, JD Gilmore, PN Hawkins and MM Reilly

J Neurol Neurosurg Psychiatry 2014 85: e4
doi: 10.1136/jnnp-2014-309236.171

Updated information and services can be found at:
http://jnnp.bmj.com/content/85/10/e4.81

Email alerting service

These include:
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/