with late-onset pompe disease, with and without enzyme replacement therapy

J. Muñoz-Blanco[a], I. Catalina-Álvarez[a], M.R. Jiménez-Bautista[a], I. Bretón-Lesmes[b], A. Traba-López[c]. [a]*ALS-Neuromuscular Unit Neurology, Hospital Gregorio Marañón. IiSGM, Madrid, Spain;* [b]*Nutrition Unit Endocrinology, Hospital Gregorio Marañón. IiSGM, Madrid, Spain;* [c]*Clinical Neurophysiology, Hospital Gregorio Marañón. IiSGM, Madrid, Spain*

Background: Pompe disease is a rare, autosomal recessive disorder caused by deficiency of acid alpha-glucosidase. The effects of ERT on strength, motor function, pulmonary and cardiac status, and quality-of-life measures in late-onset Pompe patients, at varying degrees of disease severity, need to be more fully documented.
Objective: Assess differences in the clinical course, with and without ERT for 6 months follow-up, of identical twins with late-onset Pompe disease.
Patients and Methods: Identical twins, 50 years old males, with heterozygous mutation (IVS1 -13 T > G; p.D645Y) in GAA gen. Onset of symptoms at 33 years with exercise intolerance, myalgias, orthopnea, pelvic girdle and axial weakness, without significant clinical differences between them. One needs NINV two months before ERT (Case A). After informed consent, we initiate a 6 months follow-up, bi-weekly, one case with ERT (algluosidase_alfa), 20 mg/kg/bi-weekly, the other with medical monitoring (Case B) by FVC (sitting_lying), 6MWT, composite MRC, hand grip, both sides, and CPK levels.
Results: No adverse effects in both.
Baseline, 6th month determination: FVCsitting (A: 84%, 85%/B: 88%, 87%); FVClying (A: 49%,65%/B: 49%, 48%); 6MWT-m (A: 556,705/B: 576,598); Grip (kg) right (A: 32, 55/B: 35, 37); Grip (kg) left (A: 29, 49/30, 28); composite-MRC (A:100, 110/B: 100, 100); CPK (U/L) (A: 530, 416/B: 550, 568). The treated case showed a significant improvement in FVClying (+16%) and the difference between FVCsitting-lying (<15%), hand grip with both hands (+71% right, +69% left) and 6MWT (+26,7%). The untreated patient remained stable.
Conclusion: ERT patient showed significant functional improvement over the case untreated, clinically and genetically identical.

doi:10.1016/j.jns.2015.08.1211

1159
WFN15-0760
Neuromuscular Disorders
Early detection of mononeuritis multiplex & diagnosis of systemic diseases thru electrophysiological work out with polyneuropathy as preceeding symptom

B. Mutluay[a], A. Koksal[a], N. Karagoz[a], F. Aysal[b], S. Baybas[a], A. Soysal[a]. [a]*Neurology, Bakirkoy Research and Training Hospital for Psychiatry Neurology and Neurosurgery, Istanbul, Turkey;* [b]*Neurology, Medipol University, Istanbul, Turkey*

Background: Mononeuritis multiplex (MNM) is a nervous system disorder that involves damage to at least two separate peripheral nerves. It is a syndrome not a disease, caused by certain systemic diseases like diabetes, vasculitis, rheumatic, infectious or paraneoplastic diseases.
Objective: To evaluate the role of electrophysiological work out on patients presenting with polyneuropathy as preceding symptom that leads to early diagnosis of MNM and an underlying systemic disease.
Patients: We retrospectively analyzed 12 MNM patients (4 females and 8 males from 19 to 62 years of age) presenting with patchily distributed weakness in all and pain at onset in nine, at our neuromuscular diseases clinic between 1993-2013. We have obtained Institutional Review Board (IRB) approval, as necessary.
Methods: Neurophysiological evaluation, routine blood chemistry, vasculitis markers, serum and protein electrophoresis, HIV, Hepatitis markers were examined in all patients. Nerve and muscle biopsies were performed in 5 patients.
Results: Neurophysiological evaluation revealed an asymmetrically distributed motor and sensory nerve involvement accompanied by neurogenic findings in all. Nerve and muscle biopsies were performed in five. The differential diagnostic work up of this patient group resulted in diagnosis of 2 Churg- Strauss syndrome, 2 rheumatoid arthritis, 2 non-necrotizing vasculitis, 2 PAN and 1 multiple myeloma, 1 CNS vasculitis related to p-ANCA, 1 Hepatitis C and 1 HIV.
Conclusion: Detailed investigation of patients with polyneuropathy as preceding symptom thru electrophysiological work out can be a valuable tool that leads to early detection and treatment of MNM and the underlying systemic disease.

doi:10.1016/j.jns.2015.08.1212

1160
WFN15-1196
Neuromuscular Disorders
First evaluation of efficacy and safety of intravenous immunoglobulin (ivig) treatment of dysimmune small-fiber predominant polyneuropathy (sfpn)

R. Treister[a], X. Liu[a], M. Lang[a], K. O'Neil[a], A.L. Oaklander[b]. [a]*Neurology, Massachusetts General Hospital Harvard Medical School, Boston, USA;* [b]*Neurology Neuropathology, Massachusetts General Hospital Harvard Medical School, Boston, USA*

Background: IVIG effectively treats autoimmune large-fiber neuropathies but it is untested for SFPN, which usually causes chronic widespread pain and/or dysautonomic symptoms. However, some SFPN patients appear to have autoimmune causality and receive IVIG empirically.
Objective: To retrospectively evaluate efficacy and safety of IVIG for treating SFPN.
Patients and Methods: Ethical permission for medical-record review was obtained. Inclusion criteria comprised physician's impression of SFPN plus confirmation by nerve biopsy, PGP9.5-immunolabeled distal-leg skin biopsy, or autonomic function testing (AFT). Autoimmune attribution required other autoimmune diagnoses or abnormal serologies, plus comprehensive exclusion of other causes. Inclusion required pre-treatment pain ≥ 3/10 and ≥ one IVIG dose of 2 grams/kg/4 weeks. Outcomes included changes in pain scores (2-tailed, paired t-test analysis) and AFT interpretation (chi-square), plus patients' Global Impression of Change (PGIC, 1-7 scale).
Results: 31 patients met inclusion criteria, all Caucasian; 81% female, and on average 43.6 ± 17.5 years old (14-71y). Treatment duration averaged 84.2 ± 81.1 weeks (5-285w). Their baseline pain of 6.2 ± 1.7 became 5.1 ± 2.3 during treatment ($p = 0.059$). Their 26 AFT results were 73% abnormal at baseline vs. 52% during