Poster Tour:
Muscle/NMJ/Peripheral Nerve

May 6, 11:45 - 12:45

Hall 3

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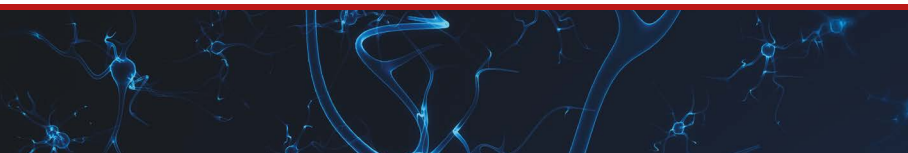

A Myositic Mirage: When Rheumatoid Vasculitis Mimics Inflammatory Myopathy

Rajesh Ambati, Shafaq Abbas, Sarah Siddiqui

East Kent Hospitals University foundation Trust

An 81-year-old man with long-standing seropositive rheumatoid arthritis (RA) presented with mobility decline, lower limb weakness and thigh pain for 3 months. Examination revealed mild distal upper limb weakness, grade 4/5, left > right, prominent proximal lower limb weakness, grade 2-3, L >R and mild left foot drop. Tendon reflexes were absent in legs and distal sensory loss noted in upper and lower limbs. Differentials included myositis (inflammatory or inclusion body) and/or mononeuritis multiplex.

Investigations showed elevated inflammatory markers (CRP 182 mg/L, ESR 104 mm/hr), normal creatine kinase, negative autoimmune serology & myositis antibodies. MRI thighs demonstrated inflammatory changes suggesting myositis. Neurophysiology showed severe, generalized, axonal sensorimotor neuropathy. Quadriceps muscle biopsy revealed lymphocytic and macrophage inflammation involving medium-sized arteries, with necrotic muscle fascicles, supporting a vasculitic process.

A diagnosis of skeletal muscle vasculitis and vasculitic mononeuritis multiplex was made. The patient was treated with intravenous methylprednisolone and rituximab, without significant improvement, with subsequent switch to cyclophosphamide, leading to clinical improvement and normalization of inflammatory markers.

This case illustrates a rare presentation of vasculitis involving muscles associated with RA. A myositis-type presentation in RA should raise suspicion of skeletal muscle vasculitis, which can be responsive to aggressive immunosuppressive therapy.

Myasthenia Gravis in Pregnancy: Outcomes from a Tertiary Centre Cohort

Bhairavi Wijayendran, Natasha Sharman, Mandish Dhanjal, Charlotte Frise, Pooja Dassan, Stuart Viegas

Imperial College NHS Healthcare Trust

Background: Myasthenia gravis (MG) affects women of reproductive age, yet contemporary real-world data describing disease behaviour and pregnancy outcomes remain limited. Pregnancy and the postpartum period are recognised risk periods for disease instability, necessitating coordinated multidisciplinary care.

Methods: We conducted a retrospective cohort study of women with MG managed through pregnancy at a tertiary neuroscience centre between 2018 and 2025. Maternal characteristics, antibody status, MG disease activity, treatment escalation, obstetric outcomes, and neonatal outcomes were analysed.

Results: Twenty-six pregnancies in 20 women were included. Fourteen women were acetylcholine receptor antibody–positive, three were MuSK-positive, two were seronegative, and one had unknown antibody status. MG exacerbation occurred in 38% of pregnancies, most frequently during mid-to-late gestation, with bulbar or respiratory involvement in a subset. Management required corticosteroid escalation and, where indicated, intravenous immunoglobulin or plasma exchange. Most deliveries occurred at term; operative delivery was common and largely driven by obstetric indications. Neonatal outcomes were favourable, with transient neonatal myasthenia occurring infrequently. Postpartum exacerbation was observed in a proportion of pregnancies.

Conclusion: MG exacerbation during pregnancy remains common, but with timely multidisciplinary management, maternal and neonatal outcomes are generally favourable.

Establishing a Tofersen service for SOD1-ALS:Audit of single centre experience at St Georges Hospital, London

Viva Levee, Derek Weidner, Lucy Yarham, Clare Galtrey

St George's Hospital London

Background: Tofersen is an intrathecal anti-sense oligonucleotide used to treat motor neuron disease due to a SOD1 gene mutation.Clinical trials show that tofersen stabilises disease progression and improves quality of life.Tofersen is currently available through the Early Access to Medicines Scheme and is being appraised by the National Institute of Health and CareExcellence.We set up a service for SOD1-ALS patients in SouthWest London and Surrey since February 2023.

Methods: Retrospective audit of electronic notes from February 2023.

Results:There was 100% compliance with the suggested protocol. 4 patients with SOD1-variant pathogenic mutations received tofersen. Mean age of symptom onset was 46 years and treatment initiation 48 years.Post-initiation ALSFRS progression rate ranged from 0.04 to 0.2. Adverse events included non-infective pyrexia with lower back pain and myelitis 30 months post-treatment initiation.This was treated with intravenous steroids, symptoms have essentially resolved and tofersen has been held.

Conclusion: We successfully set up an intrathecal tofersen service.We highlight the role of the multi-disciplinary team and use of evidence-based standardised operating procedures. Clinical outcomes including ALSFRS remained stable.There were significant adverse events, emphasising the importance of close monitoring.The data will contribute to the development of national MDT's discussing these complex cases.A national consensus for "stopping and starting criteria" is underway.

Progressive muscle MRI changes in periodic paralysis: a longitudinal cohort study

Murva Asad[1,2], Dipa Jayaseelan[1,2], Jasper M Morrow[1,2], John S Thornton[3,4], Sachit Shah[3,4], Tarek Y Yousry[3,4], Michael G Hanna[1,2], Vinojini Vivekanandam[1,2]

[1]National Hospital For Neurology And Neurosurgery, University College London Hospitals NHS Foundation Trust, Queen Square, [2]Queen Square Centre for Neuromuscular Diseases, UCL Queen Square Institute of Neurology, [3]Lysholm Department of Neuroradiology, National Hospital for Neurology and Neurosurgery, [4]Neuroradiological Academic Unit, UCL Queen Square Institute of Neurology

Background: Periodic paralyses (PP) are inherited muscle channelopathies characterised by episodic weakness and an increasingly recognised progressive myopathy. While cross-sectional muscle MRI abnormalities have been described, longitudinal MRI data in PP remain limited.

Objective: To characterise longitudinal lower limb muscle MRI changed in PP and correlate with clinical features and treatment.

Methods: A retrospective longitudinal study was conducted in adults with genetically confirmed PP who underwent baseline and follow-up muscle MRI as part of their clinical care. Bilateral thigh and calf 3T MRI included axial T1-weighted and STIR sequences. Semi-quantitative scores assessed fat replacement, muscle atrophy and STIR hyperintensity. Progression rates and associations with clinical measures were analysed.

Results: Fifteen participants (aged 21–69 years) were included, with a mean MRI interval of 4.5 years. All showed MRI abnormalities at baseline. Fat replacement progressed in 53% of participants. Increased STIR hyperintensity was observed in 24% of muscles, while 7% improved. Baseline muscle atrophy was minimal but progressed in over half of participants. Although functional decline was noted, MRI progression was also detected in clinically stable individuals.

Conclusions: Longitudinal muscle MRI identifies modest but measurable structural progression in PP, including subclinical changes which can be a useful outcome measure in clinical trials.

Acquired Multiple Acyl CoA Deficiency: 2 case reports

Wenona Barnieh[1], Mya Myintzu[1], Simon Heller[2], Radha Ramachandran[2], Raphael Buttigieg[2], Rebecca Cooper[1]

[1]Department of Neurology, Royal Sussex County Hospital, UHSussex NHS Foundation Trust, [2]Department of Adult Inherited Metabolic Disease, uy's and St Thomas' NHS Foundation Trust

Multiple acyl CoA dehydrogenase deficiency (MADD) is regarded as a disorder of fatty acid oxidation caused by recessive mutations affecting flavoprotein or flavoprotein dehydrogenase electron-transfer, or riboflavin metabolism. Late-onset MADD can present in adulthood with a wide range of neuromuscular symptoms, often demonstrating a good response to riboflavin supplementation. Nutritional riboflavin deficiency and sertraline have been reported to be associated with acquired forms.

We present two cases of late-onset MADD, in which no causative genetic abnormality was identified. Acylcarnitine profile and clinical features were seen to markedly improve with improved nutrition and treatment with riboflavin. Both patients had significant mental health history, were on sertraline at presentation, and shared a history of sub-optimal nutrition. Shared clinical features included a subacute onset, exertional breathlessness, head drop, and nasal speech, in addition to exercise induced myalgia. Both had moderately elevated creatine kinase levels and an abnormal muscle biopsy.

An inflammatory myositis was initially considered in both presentations.

It is likely that poor nutritional status with relative riboflavin deficiency, in combination with sertraline exposure, contributed to the clinical presentation in these cases.

NF2-Schwannomatosis Associated Polyneuropathy: A Single Retrospective Case Series

Lucy Manley[1], Claire Forde[2], Tim Lavin[2]

[1]Doncaster And Bassetlaw Teaching Hospitals, [2]Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University NHS Foundation Trust

NF2-schwannomatosis is a rare autosomal dominant disorder characterised by multiple nervous system tumours. Generalised polyneuropathy is recognised in NF2 and, although previously attributed to tumour-related nerve compression, emerging evidence suggests alternative pathogenic mechanisms.

From 532 patients managed within the highly specialised NF2 service in Manchester, we identified individuals with polyneuropathy and reviewed their clinical and neurophysiological features through retrospective casenote analysis. Inclusion criteria were confirmed polyneuropathy on clinical and neurophysiological assessment without an alternative cause. Patients under 18, managed at other NF2 centres, or deceased were excluded.

Seven cases of generalised polyneuropathy were identified. Severe truncating NF2 mutations were present in 4/7 patients, while 3/7 were mosaic but exhibited severe clinical phenotypes with high tumour burden. Clinically, 6/7 demonstrated asymmetrical distal motor weakness, and 5/7 had asymmetrical distal upper-limb involvement.

Neurophysiology showed asymmetrical sensorimotor polyneuropathy of predominantly axonal type in all cases, with EMG evidence of superimposed mononeuropathies not attributable to tumour compression in 6/7.

Conclusion: Asymmetrical axonal sensorimotor neuropathy with additional mononeuropathies appears to be the predominant neuropathic phenotype in NF2-schwannomatosis. Severe truncating variants were the most frequently identified genetic subtype in affected individuals.

Monomelic weakness and numbness: A case of radiotherapy-induced Focal CIDP

Li Ying Tay, Iman Saeed, Arani Nitkunan, Ahmed Abbas

[1]St George's Hospital, [2]St George's Hospital, [3]Croydon University Hospital, [4]St George's Hospital

An 81-year-old woman received radiotherapy to her left axilla and supraclavicular fossa for left breast cancer with nodal and bony metastases. 15 months later, she progressively developed isolated left (non-dominant) flaccid arm weakness, pain and paraesthesia impacting her ability to shower, dress and cook.

MRI showed asymmetric T2 hyperintensity and smooth enhancement of the left brachial plexus and left C4-T1 nerve roots. Nerve conduction studies demonstrated significant demyelination exclusively affecting sensory and motor fibres of the left upper limb, with conduction slowing and temporal dispersion identified in the distal left forearm (median/ulnar nerves) and partial conduction block in the left arm (median nerve). Electromyography revealed active and chronic denervation in left upper limb muscles, with no myokymia. PET-CT showed no abnormal uptake in the left brachial plexus.

She fulfilled EAN/PNS 2021 clinical-electrophysiological criteria for Focal Chronic Inflammatory Demyelinating Polyneuropathy (Focal CIDP). We propose this could represent a rare association of post-radiotherapy Focal CIDP. She was treated with intravenous immunoglobulin and made good objective recovery in strength and function within 3 months. In this context, distinguishing radiation-induced Focal CIDP from more common differentials such as radiation plexopathy or classical malignant infiltrative neuropathy is crucial for accurate diagnosis and appropriate immunomodulatory treatment.

Acquired Vitamin B6 Deficiency in POEMS: A Marker of Disease Activity and Cause of Neuropathy?

Oliver Tomkins[3], Simon Pope[3], Youssef Khalil[2], Shiwen Koay[1], Jahanzaib Khawaja[3], Jonathan Sive[3], Shirley D'Sa[3], Peter Clayton[2], Philippa Mills[2], Michael Lunn[1]

[1]NHNN, [2]UCL, [3]UCLH

POEMS syndrome is a rare, life-threatening multisystem disorder driven by clonal plasma cell proliferation, characterized by disabling neuropathy, monoclonal plasma cell disorder, and paraneoplastic features. VEGF is the best known and usual diagnostic marker and correlates with disease activity, but the mechanism underlying neuropathy remains unclear. Pyridoxal 5β-phosphate (PLP), the active form of vitamin B6, is an essential cofactor; its deficiency can cause distal painful sensorimotor neuropathy.

Following two case reports of B6 deficiency in POEMS, we assessed PLP levels in 50 patients (18 active, 32 in remission) between September 2023 and July 2024. Active POEMS patients had significantly lower PLP (median 9.5 nmol/L) than those in remission (median 36 nmol/L; $p<0.0001$). PLP negatively correlated with VEGF ($r=-0.64$, $p<0.0001$) and paraprotein ($r=-0.38$, $p=0.0068$). In nine patients, PLP normalized after treatment-induced remission without supplementation (median 8.5 to 37 nmol/L; $p<0.002$). Two OEMS cases (POEMS features without neuropathy) maintained normal PLP. Elevated pyridoxic acid ratios in active disease suggest increased catabolism and dysfunctional B6 metabolism.

We hypothesize that severe actual and functional PLP deficiency contributes to neuropathy in POEMS. POEMS neuropathy closely resembles historical and genetic B6 deficiency. Further studies should clarify mechanisms and evaluate B6 supplementation.

From IVIg Optimisation to Home-Based Immunoglobulin Therapy

Devan Mair[1,2], Safiya Zaloum[1,3], Joela Matthews[1], Peter Arthur-Farraj[1], Apeksha Shah[1], Aleksandar Radunovic[1], Stephen Keddie[1]

[1]Barts Health NHS Trust, [2]North Bristol NHS Trust, [3]Great Western Hospitals NHS Foundation Trust

Background: Intravenous immunoglobulin (IVIg) is an effective and well-tolerated treatment for inflammatory neuropathies but is expensive, averaging £85k/year per patient, requiring significant day-unit facilities and administration. Guidance recommends regular rationalisation of dose and interval. Novel approaches such as subcutaneous immunoglobulin (SCIg) enable at-home treatment and savings from reduced hospital visits.

Methods: 61 neurology patients established on long-term IVIg (>6 months) at Barts Health Trust were identified from the National Immunoglobulin Database. Data on dose, interval and infusion days were analysed (2008–2023). Annual equivalent dose, hospital days, and costs were estimated using average NHS pricing (£66/g; £853 per infusion).

Results: Median IVIG duration was 5.5 years, with CIDP and MMN the most common diagnoses (n=31, n=22 respectively). Seventeen patients received dose reductions (median 150β120g), saving ~£275,000 annually (~£16,000 per patient). Interval lengthening occurred in 21 (median 5→8 weeks). Median hospital days fell from 35 to 17 across all patients, saving ~£666,000. A strategy of dose optimisation followed by transfer to SCIg was therefore adopted.

Conclusion: Optimising IVIg through dose reduction, interval extension, and fewer infusion days offers substantial financial and resource savings without compromising care. These efficiencies support introducing SCIg, reducing infusion costs via self-administration and improving patient convenience.

Optimising CIDP Care: A systematic review of the evidence supporting structured Immunoglobulin withdrawal trials

Muhammed Ameen Noushad[1], Aisling Carr[2,3], Chinar Osman[1]

[1]Wessex Neuroscience Centre, [2]Centre for Neuromuscular Diseases, National Hospital for Neurology and Neurosurgery, [3]Department of Neuromuscular Diseases, Institute of Neurology, UCL

Background: Neurology immunoglobulin(Ig) usage accounts for £200million of NHSE's £350million annual Ig spend The majority as maintenance management of inflammatory neuropathies, in particular CIDP. CIDP is a relapsing-remitting disease and longterm maintenance management is not always required. Data from the placebo arm of CIDP RCTs provide valuable insight into remission rates when routine treatment is withdrawn in the closely monitored clinical trial setting.

Methods: We reviewed nine CIDP treatment studies published since 2017 (including 6x RCTs: PATH, ADVANCE-CIDP 1, ADHERE, ProCID, FORCIDP, RECIPE) a rozanolixizumab and 2 rituximab studies. We extracted proportion of participants in the placebo arms who did not relapse (remission rate).

Results: A total of 878 patients with definite or probable CIDP according to 2010 EAN/PNS CIDP diagnostic criteria were included, range: 17-211. Follow up ranged from 4-48 months (median: 12 months). The Median remission rate: 37%(range:20-68.8%). ADVANCE-CIDP did not perform a pre-inclusion treatment withdrawal period and had the highest remission rate:68.8% In the open label extension, 100% of those who did relapse were rescued to previous baseline within 2 weeks on re-treatment.

Conclusion: This data advocates for structured CIDP Ig withdrawal protocols to identify patients in remission with potential for significant NHS cost savings.