2299 GENETIC DIAGNOSES OF CHILDHOOD ONSET MOVEMENT DISORDERS

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10.1136/bmjno-2022-ANZAN.85

Objectives To identify and characterize childhood onset movement disorder phenotypes with underlying genetic etiologies, and to elucidate genetic findings, as part of the larger Neuro-CONNECT study.

Methods All previous patients known to the adult Genetics or Neurology departments at Westmead Hospital were included for analysis if they had childhood onset movement disorder phenomenology and had undergone genetic investigation. Thereafter, their comprehensive movement disorder phenotype was collated through electronic medical and clinical video recordings, neuroimaging, and biochemistry results. Their phenotypes was then compared with the results from their genetic investigations if available.

Results A retrospective cohort of 21 patients with a mean age of symptom onset of 11.6 years and subsequent genetic investigations in adulthood was collated. Genetic testing results were informative for 10 patients, uninformative or inconclusive for 10 patients and pending for 1 patient. 22 unique variants from a total of 24 variants across 19 unique genes were identified from whole genome or exome sequencing, targeted multi-, or single gene panels. 12 variants of uncertain significance (VUS), 4 likely pathogenic variants and 8 pathogenic variants were identified overall. Dystonia and tremor were the most common movement phenomena across different phenotypes, followed by myoclonus and ataxia. Neurodevelopmental disorders and/or intellectual disability were the most common secondary disorder across phenotypes.

Conclusion A causative genetic diagnosis was given for 10 adult patients (47%) with childhood onset movement disorders. Dystonia was the predominant or an observed phenotypic trait in every patient barring one (95%), and in every patient with a genetic diagnosis (100%).

2302 RHEUMATOID MENINGITIS PRESENTING WITH RECURRENT STROKE-LIKE EPISODES

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10.1136/bmjno-2022-ANZAN.86

Introduction We report a rare presentation of recurrent stroke like symptoms secondary to rheumatoid meningitis in a patient with well controlled rheumatoid arthritis.

Case A 65-year-old-gentleman with a background of rheumatoid arthritis developed sudden onset of expressive aphasia and right-sided weakness. Over the next two weeks, he had recurrent such stereotyped episodes . Pyrexia and night sweats were also noted during admission. Magnetic resonance imaging (MRI) of brain showed multifocal leptomeningeal enhancement which progressed on short interval serial imaging. Cerebrospinal fluid (CSF) analysis showed elevated protein, low glucose and elevated white blood cells which were predominately mononuclear. Electroencephalogram captured one of the episodes wherein there was corresponding pleomorphic delta slowing of the left temporal leads, suggestive of a possible epileptiform abnormality. A diagnosis of likely rheumatoid meningitis was made after exclusion of infective and alternative causes on extensive diagnostic testing including blood, CSF, radiological and nuclear medicine investigations. Treatment with pulsed intravenous methylprednisolone followed by oral prednisolone taper resulted in remission of symptoms and resolution of MRI findings. He remains well a year on.

Conclusion Rheumatoid meningitis is rarely described but important differential to consider in a patient presenting with recurrent stroke-like focal neurological symptoms associated with aseptic meningitis with a known rheumatoid arthritis. It can be seen in patients whose peripheral manifestations of rheumatoid arthritis are well controlled on disease modifying therapy, as in our patient. Early treatment with high dose corticosteroids can be helpful after exclusion of infection and malignant causes.

2304 CLADIN: CLADRIBINE AND INNATE IMMUNE RESPONSES IN MULTIPLE SCLEROSIS

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10.1136/bmjno-2022-ANZAN.87

Introduction Cladribine (Mavenclad®) is an oral treatment for relapsing remitting MS (RRMS). In RRMS clinical trials, Cladribine has been shown to reduce brain atrophy, relapse rates, and new lesions on MRI. Cladribine is thought to produce its beneficial effects through its lymphocytic actions.

Objective To investigate the mechanism of action of Cladribine on peripheral innate immune cells, in particular monocytes.

Methods This is a Phase IV, open-label, multi-centre, 3-year, translational trial. Forty RRMS patients commencing Cladribine were prospectively recruited into this study. Peripheral monocytes were isolated from whole blood using negative selection, and stained with the following markers for flow cytometric analysis (CD14, CD16, HLADR, CD11b, P2X7R, DAPI). Blood samples and clinical information were collected at: T0 (prior to cladribine commencement), week 1, week 8, week 26, and week 52.

Results There was evidence of reduction in monocyte count at week 1 compared to prior to treatment (0.55 ±0.04 vs 0.08