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IN BRIEF

NEURODEVELOPMENTAL DISORDERS

Symptom acuity can aid selection of genotyping technique

Conventional genetic diagnostics of neurodevelopmental disorders (NDDs) yield a diagnosis in only around half of patients. Using rapid whole-genome sequencing (WGS), Soden *et al.* were able to successfully diagnose 11 out of 15 infants with acute onset of NDD symptoms soon after birth. The result suggests that rapid WGS is a cost-effective strategy to evaluate infants with a suspected NDD. Whole-exome sequencing was reported to be cost-effective in children whose symptoms manifested later.

Original article Soden, S. E. *et al.* Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. *Sci. Trans. Med.* **6**, 256ra168 (2014)

NEUROMUSCULAR DISEASE

Patients with refractory myasthenia gravis respond to immunotherapy after cyclophosphamide induction

A new study suggests that pretreatment with cyclophosphamide can enhance treatment benefit in otherwise refractory patients with myasthenia gravis. Eight patients with myasthenia gravis that was unresponsive to standard therapy received intravenous cyclophosphamide for 6 weeks. Six of the eight patients responded well to standard immunotherapies after cyclophosphamide induction, and four of them remained in remission for over 2 years.

Original article Buzzard, K. A. *et al.* Induction IV cyclophosphamide followed by maintenance oral immunosuppression in refractory myasthenia gravis. *Muscle Nerve* doi:10.1002/mus.24536

PARKINSON DISEASE

α -Synuclein antibodies might not be a reliable PD biomarker

α -Synuclein aggregates are a major pathogenetic component of Parkinson disease (PD), and naturally occurring antibodies for this protein have been investigated as a biomarker of PD. A new study used ELISA to compare levels of naturally occurring antibodies for α -synuclein in the serum and cerebrospinal fluid of patients with PD and controls. No differences were found, but the investigators reported high inter-assay variability in antibody measurements, which could have biased the results. The observation highlights the importance of established protocols and quality control to the investigation of biomarkers for PD.

Original article Heinzel, S. *et al.* Naturally occurring alpha-synuclein autoantibodies in Parkinson's disease: sources of (error) variance in biomarker assays. *PLoS ONE* **9**, e114566 (2014)

DEMENCIA

Visual hallucinations in neurodegenerative disease are associated with volume loss in the right anterior insula

Visual hallucinations are common in patients with dementia with Lewy bodies (DLB), and can also be present in Alzheimer disease (AD). Voxel-based morphometry in 78 patients with AD, half of whom experienced hallucinations, detected specific regions of grey and white matter atrophy associated with hallucinations; the right anterior insula was of core importance. Autopsy of four patients who had experienced hallucinations revealed combined AD and DLB pathology.

Original article Blanc, F. *et al.* Right anterior insula: core region of hallucinations in cognitive neurodegenerative diseases. *PLoS ONE* **9**, e114774 (2014)