RESEARCH HIGHLIGHTS

IN BRIEF

MOTOR NEURON DISEASE

Mutations in ATXN2 increase the risk of amyotrophic lateral sclerosis

CAG repeat expansions in the ataxin-2 gene (ATXN2) cause spinocerebellar ataxia type 2, and could have a role in amyotrophic lateral sclerosis (ALS). Through haplotyping and DNA sequencing in an ALS pedigree, Laffita-Mesa et al. recently identified de novo CAG expansions in ATXN2 alleles in three patients with autosomal dominant ALS. Moreover, a meta-analysis of other studies showed such expansions were associated with an increased risk of ALS.

Original article Laffita-Mesa, J. M. et al. De novo mutations in ataxin-2 gene and ALS risk. PLoS ONE 8, e70560 (2013)

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MULTIPLE SCLEROSIS

RBPJ identified as an autoantigen in multiple sclerosis

The autoantigens in multiple sclerosis (MS) are not fully identified. According to a recent study, RBPJ could be an autoantigen in a subset of patients. Cerebrospinal fluid (CSF) from patients with MS and elevated IgG was screened against an array of 9,393 proteins. CSF autoantibodies against RBPJ were more prevalent in patients with MS (12.5%) than in those with other neurological diseases (1.6%). Studies to investigate the potential role of RBPJ in MS could improve our understanding of MS pathogenesis.

Original article Querol, L. et al. Protein array-based profiling of CSF identifies RBPJ as an autoantigen in multiple sclerosis. Neurology doi:10.1212/WNL.0b013e3182a43b48

DEMENTIA

Risk factors at adolescence for young-onset dementia

Young-onset dementia (YOD; diagnosis before age 65 years) is associated with a genetic predisposition, but other risk factors are unknown. In a large cohort study of Swedish males (mean age 18 years), researchers identified nongenetic risk factors at adolescence that are significantly associated with later development of YOD, including alcohol intoxication, stroke and depression. Identification of such factors could be useful for prevention strategies.

Original article Nordstrom, P. et al. Risk factors in late adolescence for young-onset dementia in men: a nationwide cohort study. JAMA Intern. Med. doi:10.1001/jamainternmed.2013.9079

MOVEMENT DISORDERS

Focused ultrasound thalamotomy shows early promise for treatment of essential tremor

Essential tremor is the most common movement disorder, and is incompletely controlled in up to 50% of patients. In an open-label, uncontrolled study high-intensity focused ultrasound, guided by MRI, was used for thermal ablation of specific thalamic regions in 15 patients with treatment-resistant essential tremor. Over 12 months of follow-up, tremor, disability and quality of life scores improved significantly from baseline. The most frequent adverse effect was paraesthesia, which persisted in four patients. These results call for larger, controlled studies of this therapeutic approach in refractory essential tremor.

Original article Elias, W. J. et al. A pilot study of focused ultrasound thalamotomy for essential tremor. N. Engl. J. Med. 369, 640-648 (2013)