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IN BRIEF

MOTOR NEURON DISEASE

Genetic factors underlie variability in the amyotrophic lateral sclerosis phenotype

Previous reports have indicated that mutations in the *FUS/TLN1* gene account for 0.6–6.0% of familial amyotrophic lateral sclerosis (FALS) cases. In a recent study, Albert Ludolph and colleagues found a higher frequency of *FUS/TLN1* mutations (8.7%) in a German FALS cohort. They also showed that truncating mutations in *FUS/TLN1* produced a more severe disease phenotype than missense mutations in the same gene. These findings could have implications for patient evaluation and genetic counselling.

Original article Waibel, S. *et al.* Truncating mutations in *FUS/TLN1* give rise to a more aggressive ALS-phenotype than missense mutations: a clinico-genetic study in Germany. *Eur. J. Neurol.* doi:10.1111/ene.12031

NEURODEVELOPMENTAL DISORDERS

Gene expression profiling in blood could aid diagnosis of autism spectrum disorders

A 55-gene blood transcriptional signature, ASD55, can distinguish children with autism spectrum disorders (ASD) from controls, according to new research. In an all-male sample cohort (66 with ASD and 33 controls), ASD55 showed 72.5% accuracy. The corresponding figure for males in a validation cohort was 72.7%, but ASD55 performed less well in females (63.8% accuracy). As well as aiding diagnosis, the differentially expressed genes may provide insights into the pathophysiology of ASD.

Original article Kong, S. W. *et al.* Characteristics and predictive value of blood transcriptome signature in males with autism spectrum disorders. *PLoS ONE* 7, e49495 (2012)

ALZHEIMER DISEASE

IL-12/IL-23 pathway inhibition has therapeutic potential in Alzheimer disease

The IL-12/IL-23 signalling pathway seems to contribute to the disease process in Alzheimer disease (AD) and could represent a novel therapeutic target, a study published in *Nature Medicine* indicates. Blockade of this pathway in a mouse model of AD led to a reduction in cerebral amyloid load and improvements in cognitive function. In addition, levels of p40, a subunit that is shared by IL-12 and IL-23, were found to be elevated in the cerebrospinal fluid of patients with AD.

Original article vom Berg, J. *et al.* Inhibition of IL-12/IL-23 signaling reduces Alzheimer's disease-like pathology and cognitive decline. *Nat. Med.* 18, 1812–1819 (2012)

NEUROMUSCULAR DISEASE

Wnt7a—a treatment for Duchenne muscular dystrophy?

A team in Canada has shown that the signalling protein Wnt7a improves muscle strength in the *mdx* mouse, an animal model of Duchenne muscular dystrophy (DMD). The action of Wnt7a in this context seems to be twofold: the molecule stimulates proliferation of satellite stem cells and induces hypertrophy of myofibres. The researchers suggest that Wnt7a has the potential to be developed as an ameliorative treatment for DMD in humans.

Original article von Maltzahn, J. *et al.* Wnt7a treatment ameliorates muscular dystrophy. *Proc. Natl Acad. Sci. USA* 109, 20614–20619 (2012)