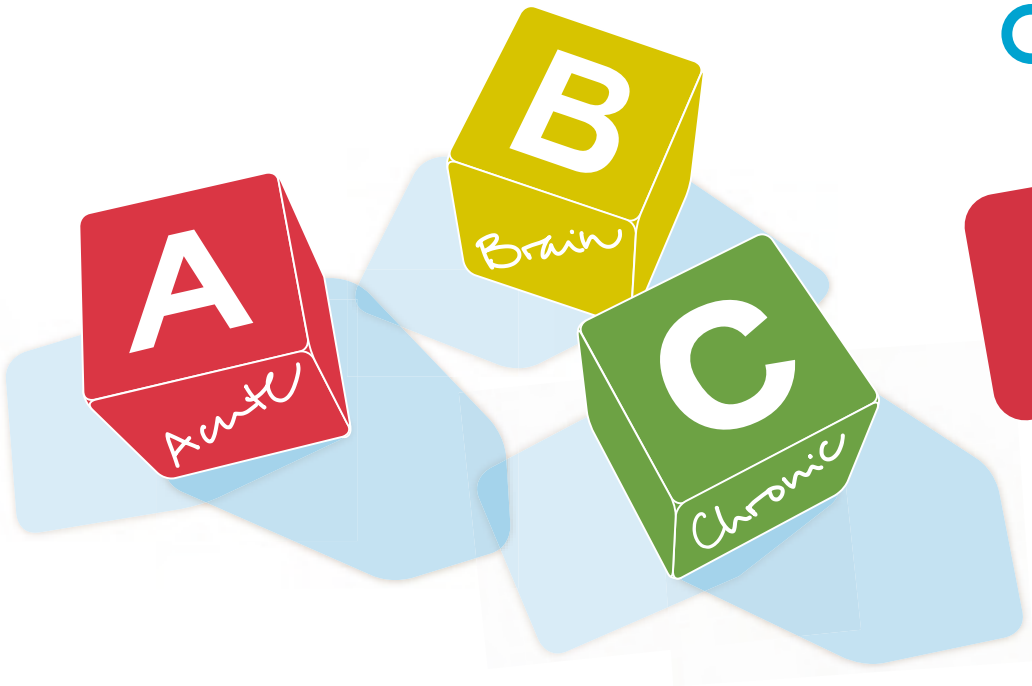
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# ABSTRACTS

Topic: Neuromuscular Disorders

**EPNS25_713 - Amyotrophic lateral sclerosis with juvenile onset. Case report**

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**Objectives:** Amyotrophic lateral sclerosis (ALS) is a motor neuron disease affecting both upper and lower motor neurons and leading to progressive paralysis. Juvenile ALS appears in a rare subgroup of patients with onset before the age of 25 years old. There are few reports of ALS in children. Here, we describe a 14-year-old boy with a rapidly progressing classical ALS phenotype caused by a mutation in FUS.

**Methods.** A previously healthy 13.5-year-old boy developed severe weakness in both upper and lower extremities and an inability to walk following a fall from a bicycle. He had also experienced intermittent weakness and imbalance in the extremities over the past three months.

**Results:** Post-fall, the weakness initially began in the left upper extremity, then in 2 weeks progressively spread to the lower extremities and the right upper extremity, leading to the loss of abilities to walk, stand, and lift his arms. Neurological examination revealed generalized muscle weakness, more pronounced in the proximal muscles. The patient required assisted walking and had axial hypotonia but no tremor, sensory deficits, or urinary/fecal incontinence. Deep tendon reflexes were normoactive, head control was normal, and no tongue fasciculations were observed. There were no swallowing or respiratory complaints. Infectious, rheumatologic, and metabolic tests were non-conclusive. EMG showed severe neurogenic involvement in the right tibialis anterior, peroneal, and gastrocnemius muscles, with moderate involvement in the right deltoid, interosseous, rectus femoris, and genioglossus muscles, suggesting an anterior horn cell disorder such as SMA or ALS. Genetic testing for SMA was negative, and CD59 deficiency-related proximal neuropathy was ruled out. Cranial and spinal MRI and MR spectroscopy were normal. There was no parental consanguinity; the patient's mother had died of an undiagnosed, rapidly progressing neurodegenerative disease with bulbar involvement at age 29. Whole-exome sequencing (WES) revealed a heterozygous *FUS* gene p525L (c.1574C>T) mutation associated with an autosomal dominant ALS6 phenotype. Since the patient had no respiratory distress or dysphagia, supportive treatment with physical therapy and vitamins was initiated.

**Conclusions:** ALS remains a rare condition in the pediatric population, but mutation in the *FUS* gene is clearly the most frequent cause of rapidly progressive pediatric ALS. Attention should be paid to searching for *FUS* mutation in pediatric ALS.