Management of seizures in children with therapy-resistant epilepsy

Epilepsies in children represent a heterogeneous group of disorders and syndromes with different etiology, severity, prognosis and treatment. Early diagnosis, accurate recognition of underlying aetiologies leads to more effective management and treatment and improve overall health and quality of life. Genetic testing is very important in the cases of therapy-resistant seizures. The purpose of study was to recognize the possible reason of failed AED treatment and to find the ways to overcome it.

Methods 45 patients with different forms of epilepsy aged from 3 months to 16 years not the candidate for surgical treatment have been studied. The long duration EEG, high resolution MRI, blood biochemical tests, blood level lactate and ammonia, amino acid, organic acid and disturbance of fatty oxidation by TMS, genetic investigations (mtDNA and exome sequence), measurement of autoantibodies to NR2 and GluR1 in blood serum by ELISA were performed to these children.

Results The respiratory chain disorders confirmed by mtDNA sequence were found in 11 children. Metabolic epilepsies discovered in patients have the following origins: two with glutaric aciduria type1, one – glutaric aciduria type2, one with propionic aciduria, one with methylmalonic aciduria, one with Gaucher’s disease type3, two patient with glycogenosis type 9, two patients with ceroid lipofuscinosis type 2 and 6,lysosomal storage disorders in 3 cases.

Genetic epilepsies with mutation in genes SCN8A (two patients), GRIN2A, KCNMA1, SRPX2, SCN9A, ACO2, ARHGEF9, 15q11.2q13.3,TSC- 4 patients were revealed. In other cases with normal MRI the reason of pharmacoresistant seizures was not discover yet. The elevated level of autoantibodies to glutamate NR2 and GluR1 receptors were found in children of these groups. But in patients with metabolic epilepsies the elevation level of autoantibodies to NR2 was in 4 to 7 times higher in comparing with children with genetic epilepsies. In children with metabolic disorders and energy metabolism disorders we use the specific therapy (special diet, L-carnitine, vitamins, enzyme replacement therapy etc) in cases which it possible, avoid valproic acid in treatment of children with mitochondrial disorders and glutaric acidurias, as well we use the phenytion in patient with potassium channel mutation SCN8A. These treatment management led to reduction in seizures frequency or even to seizures remission in some cases.

Conclusions The recognition and diagnostic of underlying etiologies of intractable seizures improve the treatment management in many cases. The excessive NMDA transmission might be the part of pathogenesis of seizures in children with inborn error of metabolism.

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Introduction Childhood arterial ischaemic stroke (AIS) is uncommon with a reported incidence between 1.2 and 7.91 per 100,000 per year. Previously it was thought that children with AIS had a good outcome due to brain plasticity; however, mortality has been reported in up to 28%, and morbidity in up to 70% of survivors. There are no randomised trials of mechanical thrombectomy in children. The 2017 published RCPCH stroke guidelines draw on the excellent outcomes for mechanical thrombectomy in adult trials and recommend referral for intra-arterial clot extraction in patients with NIHSS score of 6 or more and up to 12 hours post onset if there is salvageable brain tissue on imaging. There are only 29 paediatric cases published in the literature that have undergone mechanical thrombectomy, 12 of which were for posterior circulation AIS.

Lifesaving mechanical thrombectomy in paediatric stroke

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