Biennial National Conference of the Israeli society for Child Development and Rehabilitation

Jan 9-10, 2019

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Key Note Presentation:

Title

Epilepsy in children with neurodevelopmental disabilities: Shared causes, unique strategies

Abstract

At a busy neurodevelopmental disabilities practice, it is extremely common to see children who also have comorbid epilepsy. As both are neurological conditions with significant morbidity for the child, it is important to be able to recognize and treat both. In some children the epilepsy may be mild and easily treatable, but in some (often those with genetic or structural abnormalities as the cause for both), the epilepsy may be particularly refractory. In this lecture, we will discuss how a pediatric epilepsy specialist approaches children with comorbid neurodevelopmental disabilities – in many ways, the other side of the consult request! Recognizing that testing is expensive and sometimes requires sedation, do all children with neurodevelopment disabilities and epilepsy require MRI, EEG, genetic testing? What are the syndromes and genetic conditions that perhaps were not considered until the seizures now have occurred? Finally, what are practical tips for choosing antiseizure drugs and nonpharmacological approaches (e.g. diet and VNS) that may help seizures but also possibly issues related to the neurodevelopmental disability?

Objectives

At the end of this presentation the participants will:

- Recognize the approach of a pediatric epilepsy specialist when helping children with neurodevelopmental disabilities who have comorbid seizures
- Understand our diagnostic considerations including MRI, neuropsychological testing, EEG, and the growing use of genetic testing
- Be able to list several common epilepsy syndromes in which treating the epilepsy may improve dramatically the neurodevelopmental outcomes such as ESES, Rett syndrome, and infantile spasms

- Realize practical tips for treating children with epilepsy and neurodevelopmental disabilities including appropriate anticonvulsant drug choices and using nonpharmacological treatments (diet, VNS).

Key Note Presentation:

Title

Glut1 Deficiency Syndrome: A model neurodevelopmental disorder for specific treatment

Abstract

Glut1 deficiency syndrome is an autosomal dominant syndrome associated with early infantile seizures, moderate developmental delay, microcephaly, and a movement disorder (often ataxia). Previously solely diagnosed by lumbar puncture, the genetic cause has been identified in recent years as due to a mutation in the SLC2A1 gene, which has led to rapid (sometimes surprising) diagnoses in children with even mild (or non-existent) epilepsy and neurodevelopmental delays. As a result of research and international family support groups, the phenotypic spectrum of Glut1 deficiency syndrome has been well-described. The treatment of first choice is the ketogenic diet, which improves all of the symptoms rapidly, including cognition, but can be difficult to adhere to for the possibly years required. As a result, newer treatments such as triheptanoin, decanoate and ketones esters are under investigation as both supplements and substitutes for the ketogenic diet. Due to the wide availability of highly effective treatments, early screening has been suggested, including even newborn testing. This lecture will highlight the unique aspects of this disorder and how it represents a model for neurodevelopmental disabilities in which symptom recognition, genetic diagnosis, and aggressive treatment can have a profound impact on children's lives.

Objectives

At the end of this presentation the participants will:

- Understand the common symptoms seen in Glut1 deficiency syndrome
- Recognize the unique aspects of this neurodevelopmental disorder and how the ketogenic diet may improve not just seizures, but also cognition and ataxia
- Realize the power of strong family support groups in advocacy and even newborn screening potential in this case