A0039: Use of Early Childhood Epilepsy Severity Scale (E-Chess) in Classification and Prognostication of Children with West Syndrome: A Study from Tertiary Care Pediatric Neurology Centre

Mishra S.,[1] Mallick A.K.,[1] Mohanty G.,[1] Biswal N.[1]
[1]*Department of Neurology, S.C.B Medical College, Cuttack, Odisha, India*

Background: West syndrome is a type of pediatric epilepsy syndrome often associated with a grave prognosis. The aim of this study was to evaluate clinicoradiologically cases of West syndrome, to use E-chess scoring, and classify and use it for prognostication.

Materials and Methods: Prospective observational clinical study for 1 year in the pediatric neurology out patients department of S.C.B. Medical College, Cuttack, Odisha. Patients were included as a case of West syndrome when they met all three criteria: (1) developmental plateau or regression, (2) epileptic spasm, (3) hypsarrhythmia on electroencephalography (EEG) who came to our center for first time (with/without previous treatments) after informed consent. They were classified into three groups by E-chess scoring according to severity of disease (►**Table 1**).

Results: The total number of children included in the study was 13. Mean age of presentation was 9.4 months. Maximum patients were males. Mean duration of the disease was 3 months. Most of the patients were having hypoxic ischemic sequel in MRI. Twenty-three percent patients were categorized into type-III, 31% into type-II, 46% into type-I. Types II and III were drug-resistant epilepsy with poor response.

Conclusion: West syndrome is one of the infantile epilepsy syndromes with grave prognosis. E-chess scoring is a good and useful scoring system for classification and prognostication. This can be used in OPD basis for categorization of West syndrome. The types II and III are drug-resistant varieties with poor response to drugs. They should be planned for surgical therapy.

A0040: De Novo Mutations in TUBB2A Cause Infantile-Onset Epilepsy and Mental Retardation and Literature Review

Shuying Cai,[1] Jinliang Li,[2] Ye Wu,[2] Yuwu Jiang[2]
[1]*Department of Pediatric Neurology Rehabilitation, Maternal and Child Health Care of Xiamen, Xiamen, Fujian, China*
[2]*Department of Pediatrics, Peking University First Hospital, Beijing, China*

Objective: To investigate the clinical features of infantile-onset epilepsy and mental retardation caused by de novo mutations in TUBB2A and review the literature.

Methods: The clinical manifestations of two children with TUBB2A mutations in Pediatrics of Peking University, First Hospital were analyzed. Literature from Wanfang, Weipu, CNKI, and PubMed databases (self-built to August 2018) were searched and analyzed with the words "TUBB2A" and "epilepsy or convulsion" "epilepsy or convulsion." Summary features included clinical presentation, EEG, imaging, treatment response, and genetic mutations.

Table 1 Classification of patients by E-chess scoring according to severity of disease

Types	E-chess score
I	6–9
II	10–12
III	13–15

Results: Two cases of de novo mutations in TUBB2A were females, with infantile-onset epilepsy and global developmental delay. Case 1 is a new hybrid heterozygous mutation: c.728C > T(excon4), p.P243L(NM_001069.2), MRI images showed white matter stunting, and the left frontal angle is slightly wider; case 2 is a newborn heterozygous mutation: c 0.743 (excon4) C > T, p.A248V (NM_001069.2), brain MRI showed bilateral anterior humeral gyrus with large cerebral gyrus, considering the possibility of giant gyrus deformity, bilateral frontal lobe abnormal signal, corpus callosum dysplasia. Literature retrieval was related with four references (in English), six cases were reported de novo mutations in TUBB2A, including this study has eight cases, three cases were p.A248V mutation. The subjects were infantile-onset epilepsy (spasms), with different levels of global developmental delay. MRI images were gyri developmental deformities, bad myelination and corpus callosum dysplasia. The seizure in p.P243L mutated was easy to control, and cerebral structural change was less severe.

Conclusion: TUBB2A mutations can cause infantile-onset epilepsy and global developmental delay, and different degree of brain malformations. The p.A248v mutation may be "hot spots" mutations. The p.P243L mutation caused a slight change in brain structure, and seizures were easily controlled.

A0041: Status Epilepticus in Pediatric Patients Severity Score (STEPSS): A Clinical Score to Predict the Outcome of Status Epilepticus in Children

Sidharth Yadav,[1] Suvasini Sharma,[1] Bijoy Patra,[1] Rajeev Malhotra,[1] Virendra Kumar[1]
[1]*Lady Hardinge Medical College, Delhi, India*

Purpose In adults, Status Epilepticus Severity Score (STESS) is a good predictor of clinical outcome and treatment response. We devised a pediatric modification of this score: Status Epilepticus in Pediatric patients Severity Score (STEPSS) and evaluated it in children with status epilepticus.

Methods: In this prospective study, children aged 1 month to 18 years presenting with seizure duration of at least 5 minutes or actively convulsing to the emergency were enrolled. The parameters noted for scoring STEPSS included: level of consciousness, age, type of seizure, and previous history of seizures. Outcomes included death, Pediatric Overall Performance Category (*p*OPC) at discharge, and treatment response. The primary outcome variable was the predictive accuracy of STEPSS score for determining unfavorable outcome (death or POPC ≥ 3; indicative of moderate disability or more).

Results: One hundred forty children (mean age = 5.8 years, 94 boys) were enrolled. Overall 15 children had an