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Developmental Epileptic Encephalopathies in A National Cohort Study (Turkish EPISTEP -1): A Step-based Approachment for Etiologic Diagnosis

Objectives. (1) to define the etiologic trend of Developmental Epileptic Encephalopathies (DEE) in the genomic era with a step-based approach in a large cohort between 2005-2021. (2) to evaluate the yields of genetic and metabolic investigations in those without obvious cause after initial clinical phenomenology and magnetic resonance imaging (MRI). Methods: The study cohort consisted of 1238 patients with DEE from 16 tertiary centers between 2005 and 2021. The most likely specific etiologies were coded by two independent reviewers based on a step-based approach: step I (clinical phenomenology), step II (magnetic resonance imaging-MRI), step III (metabolic screening), and step IV (genetic tests). Etiologic diagnoses were divided into five subgroups according to the ILAE classification: structural, metabolic, genetic, immune-infectious, and unknown. Results. An etiology specific diagnosis was established 78.8% of patients (976/1238): structural, (n:335, 27.1%); genetic, (n:498, 39.8%); metabolic (n:114, 9.2%); immune-infectious, (n:34, 2.7%) (Table 1). Combined use of the first three steps (I-III) provides a specific diagnosis in 39.9% of children; clinical phenomenology (1.4%) MRI 22.3%, and

metabolic screening (16.2%) (Table 2). Since 2014 with the advent of next-generation sequencing (NGS), the diagnostic yield of step IV reached up to 60.2% with the dynamic use of genetic tests with extended variabilities. A new trend was defined for the genetic test preference in the NGS era: WES with %34.3, and epilepsy panels with 25.2% (Table 3). Conclusion. The presented step-based approachment provided more diagnostic yield and precision therapy implication (33.5%) in children with DEE.
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