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The role of genomics data on the concept of “developmental epileptic encephalopathies” in the “electroclinic syndromes”: A reflection from the national cohort study (Turkish EPISTEP-2)

Background/Objective. There is an enormous effect of genomics on the concept of "developmental epileptic encephalopathies-DEE". To define the well-known "electroclinic syndromes" in a large cohort with “DEE". Methods. A study group consisted of 655 (49%) children DEE with a genetic code labeled " gene-name DEE " from a national cohort called Turkish EPISTEP-1 (n=1336). The demographics are; the mean age at first seizure; 5.8 ±5.1 months, the mean follow-up period; 3.41±2.8 years, and the median duration of etiologic diagnosis; 20 months, genetic-molecular diagnosis with gene-based tests in 595

(90.8%) of 655 patients and remaining with chromosomal analyses. Result. According to the ILAE classification, a well-known " electroclinical syndrome " could be defined in 505 of 655 (77%) patients with " gen name DEE" in Table 1. The most common functional mutations and pathologic variants were presented in Table 2. "Electroclinical syndrome name, gene name DEE" was labeled in 317 (82.3%) of 385 patients with "gene name DEE ". The defined "gen-name DEEs" was also applied to the OMIM database (Table 3). Conclusion. This cohort study reflects that the importance of "electroclinical syndromes " has been reduced in favor of the new genetic tests. We proposed the term "gene name DEE" for the initial diagnosis of the clinical entity for still unknown electroclinic syndrome and the term " electroclinical syndrome name, gene name DEE" in order to obtain a better genotype and phenotype correlation with simple nomenclature

Keywords: developmental epileptic encephalopathies, electroclinic syndromes, gene-name , OMIM

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