Review article

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Idiopathic focal epilepsies: the "lost tribe"*

Deb K Pal [1,2], Colin Ferrie [3], Laura Addis [1], Tomoyuki Akiyama [4], Giuseppe Capovilla [5], Roberto Caraballo [6], Anne de Saint-Martin [7], Natalio Fejerman [6], Renzo Guerrini [8], Khalid Hamandi [9], Ingo Helbig [10], Andreas A. Ioannides [11], Katsuhiro Kobayashi [4], Dennis Lal [12], Gaetan Lesca [13], Hiltrud Muhle [14], Bernd A. Neubauer [15], Tiziana Pisano [8], Gabrielle Rudolf [16], Caroline Seegmuller [17], Takashi Shibata [4], Anna Smith [18], Pasquale Striano [19], Lisa J. Strug [20], Pierre Szepetowski [21], Thalia Valeta [22], Harumi Yoshinaga [4], Michalis Koutroumanidis [23]

Epileptic Disorders

Idiopathic Focal Epilepsies
From Phenotype to Genotype

- The idiopathic focal epilepsies are a group of childhood epilepsies that share clinical and electrographic similarities as well as some common genetic aetiology.
- The group includes Rolandic Epilepsy, Panayiotopoulos syndrome, Landau-Kleffner syndrome and Continuous Spikes in Slow-Wave Sleep, as well as some less commonly described proposed syndromes; the grouping is not currently recognised in the 2010 ILAE Classification.
- Distinctive semiology of these syndromes can also be mimicked by symptomatic epilepsies, which remain an important differential in atypical cases.
- The fascinating involvement of selected functional brain circuits lends support to the recent hypothesis of “system epilepsies”
- There is a large overlap between IFEs and both comorbid and complicating neurodevelopmental disorders including those involving language, attention and behaviour.

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- Advanced electrophysiological methods such as the measurement of high frequency oscillation (HFOs) and magnetoencephalography (MEG) offer potential for prognostic biomarkers and for localising generators of epileptic activity.
- Speech, language and literacy impairments are specific accompaniments of rolandic epilepsy and are important for academic achievement.
- Attentional difficulties are common and may be independent of seizures and EEG activity in RE; they may also underpin reading difficulties.
- The long-held observation of a spectrum or continuum in the IFEs with presumed shared aetiology may be explained in part by mutations in the glutamate receptor subunit *GRIN2A*, found in 5-30% of patients in the IFE group, more commonly in the more severe epilepsies eg CSWS.
- Much of the genetic basis of IFE remains to be discovered, including the role of possible non-coding variants in *ELP4-PAX6* for the autosomal dominant trait of centrotemporal spikes, as well as copy number variation in the rest of the genome.