LETTER TO THE EDITOR

Reply: Autosomal recessive epilepsy associated with contactin 2 mutation is different from familial cortical tremor, myoclonus and epilepsy

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Sir, We would like to thank Striano et al. (2013) for their remarks on our paper and would like to comment on some of the mentioned points. We agree that it is important to pay close attention to the correct classification of new syndromes such as familial cortical myoclonic tremor with epilepsy (FCMTE) as any inconsistencies will hamper the discovery of the underlying pathophysiology. Summing up, FCMTE is at present the recommended heading of a heterogeneous syndrome characterized by an autosomal dominant inheritance, cortical myoclonus and seizures. Different families exhibiting these symptoms to a various extent have previously been named FCMTE, ADCME, FAME or BAFME in the literature (Striano et al., 2010). The family described by Stogman et al. (2013) displays the characteristic clinical features of this syndrome, i.e. cortical myoclonic tremor and focal epilepsy with the marked difference of being inherited in an autosomal recessive manner. Striano et al. (2013) raise the concern that the tremor in the Egyptian family was not cortical in origin and part of the syndrome but might have been caused by treatment with carbamazepine. Although we could not perform testing of premyoclonic cortical spikes and C reflexes, which obviously would have strengthened the diagnosis, the short burst duration of the tremor (<50 ms) and the additional occurrence of myoclonic jerks is indicative of a cortical origin (Kojovic et al., 2011). Performed somatosensory evoked potentials were normal, which is not in contradiction with this syndrome (Crompton et al., 2012). There was no temporal association of the tremor, neither with the start of carbamazepine treatment nor with changes in carbamazepine dosages in any of the patients (maximal dosages were in the range of 200 mg to 800 mg daily, i.e. not particularly high). Moreover, carbamazepine-induced tremor is not very common and if one postulates that the tremor is not part of the genetic syndrome then the occurrence in all affected family members would be highly unusual (Morgan et al., 2005; Zeuner et al., 2012).

Striano et al. (2013) further question the phenotypic classification of the described Egyptian family as FCMTE based on atypical features in this family as compared to previously published kindreds. However, it can be argued that all the unusual features mentioned in the Egyptian patients have been described before in other families, in particular the occurrence of focal seizures and the later appearance of the tremor. Bilateral mesial temporal sclerosis (Patient V-7) has not been observed before, but can either be explained as a consequence of focal seizures in this particular patient or alternatively could be viewed as a new extension of a developing, heterogeneous phenotype (Thom, 2009). Differences in the inheritance pattern alone are—in our opinion—insufficient to alter the syndrome classification (Corti et al., 2011). The occurrence of unusual or even new features should be expected in any syndrome discovered only relatively recently. This applies particularly to diseases with a multigenic complex aetiology, as in FCMTE. However, having raised the above arguments in favour of the view that the Egyptian family conforms to the core criteria of FCMTE, we, nevertheless have to acknowledge the uncertainty on whether a separate classification would be justified given the unusual features. In the manuscript we tried to express these remaining doubts on the classification by only naming the phenotype in the Egyptian family as similar to FCMTE, which we are happy to restate here.

References


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