Author’s response to reviews

Title: Mutations in Danish patients with long QT syndrome and identification of a large founder family with p.F29L in KCNH2

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Author’s response to reviews: see over
Copenhagen, 10.2.2014.

Dear Editor,

Re: BMC Medical Genetics manuscript #7450516811098268, “Mutations in Danish patients with Long QT syndrome...”

We thank for reviewer comments and hereby resubmit an amended version of our paper.

The reviewer Elijah Behr had no further comments.

The reviewer Britt Maria Beckmann had the comment that we should add a sentence clarifying the consequence of the variable clinical data quality. We agree, and have added a sentence on p. 14, underlined in the section of the manuscript below. The sentence is nearly identical to the suggestion made by the reviewer.

P. 14:

“The patients described here were studied and collected over a long span of years, from 1996 – 2010. In this period the clinical picture of LQTS was better defined, the possibility of referral for genetic analysis increased and the indication for genetic analysis in LQTS cases was established in 2006 [6]. Furthermore, detailed clinical information was variable in quality, therefore it can not be excluded that, in some cases, other factors, e.g. structural heart disease or mixed phenotypes, might not have been identified and taken into account. Consequently, it is very difficult to establish a success rate for the genetic screening across this period. But it is probably comparable to the 70% reported from Norway in a much smaller collection of mutations [33].”

We find that the manuscript has been improved through the review process and hope that it is now acceptable for publication.

Kind regards

Michael Christiansen