Reply

Resposta

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Dear Editor,

We thank Finsterer et al.[1] for their interest in our paper. However, we think that their concerns have missed the target. The main purpose of our study was to call the attention of child neurologists and pediatricians to the difficulties in making the diagnosis of hereditary spastic paraplegia (HSP) in children for the reasons largely explained in the manuscript. This objective is explicit in the title: *Hereditary spastic paraplegia: a clinical and epidemiological study of a Brazilian pediatric population*[2].How does one make this diagnosis in patients in whom a genetic study cannot be performed? Moreover, it is known that in one-third to half of the patients, the genetic results are negative[3,4,5]. Should we exclude the diagnosis? These concerns are highly surprising when it appears that two of the authors of the letter work in an official Brazilian institution. Surely they are aware of the difficulty of performing genetic studies, most of which are still unaffordable, in our population.

It is true that extrapyramidal signs were the second most frequent abnormality in our cohort of complicated HSP. However, they were encountered in only six patients, this being the reason that they were not fully described. We agree that they could have been listed in Table 2. Treatment was beyond the scope of our study.

As for their disagreement with the statement that "*It is known that there are mutations in genes such as ATL1 (SPG3A) and BSCL2 (SPG17) responsible for either HSP or Charcot-Marie-Tooth disease*", we suggest a careful review of the following manuscripts:

- Timmerman, Vincent; Clowes, Virginia E; Reid, Evan. Overlapping molecular pathological themes link Charcot-Marie-Tooth neuropathies and hereditary spastic paraplegias. Experimental Neurology 2013; 246:14-25[6].
- Guo-hua Zhao and Xiao-min Liu. Clinical features and genotype-phenotype correlation analysis in patients with ATL1 mutations: A literature reanalysis. Translational Neurodegeneration 2017; 6:9[7].

Finally, the authors say that they do not agree with our statement: "*Over 70 distinct loci and over 50 genes have been identified...*" because according to them *"...at least 79 loci and at least 60 genes have been identified..."*. We think that this concern does not deserve a response and leave to the judgment of the readers the purpose of such a concern. The same argument may be applied to their proposition to revise our statement: "*pure HSPs are usually autosomal dominantly inherited and that complex HSPs are usually autosomal recessively transmitted",* when according to them "...*only 70-90% of the pure HSPs follow an autosomal dominant trait and about 20% of the pure HSPs follow an autosomal recessive trait.*"

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