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Pure or Complex Hereditary Spastic Paraplegia Type 4?

Josef Finsterer

Krankenanstalt Rudolfstiftung,
Messerli Institute,
Veterinary University of Vienna,
Vienna, Austria

Dear Editor,

We read with interest the article by Yang et al.[1] about a 45-year-old male with hereditary spastic paraplegia (HSP) due to the mutation c.1413A>G in *SPAST*. Below we present our comments and concerns about their study.

It is interesting that the reported patient did not present with muscle weakness but only isolated lower limb spasticity.[1] In addition to spasticity and muscle weakness, pure forms of HSP may also present with sensory disturbances or urinary dysfunction. Readers should therefore have been informed about whether the reported patient developed muscle weakness, bladder dysfunction, or sensory disturbances as the disease progressed during follow-up.

HSP type 4 (HSP4) may present as either the pure or complex form of HSP. Manifestations in addition to those found in pure forms of HSP4 include tremor,[2] neuropathy,[3] epilepsy,[4] upper limb spasticity, ataxia,[5] white-matter lesions,[6] cognitive impairment,[6] an abnormal rear skull pit, and recurrent stillbirths. Although the patient presented with the pure form on admission, it should also have been reported whether he developed any of these features during long-term follow-up.

HSP therapy is usually symptomatic and particularly directed toward the application of physiotherapy, use of walking aids, and administration of antispastics, anticholinergics for bladder dysfunction, and antiepileptic drugs in the case of epilepsy. The types of treatment that were recommended to the patient should have been reported, as well as their effectiveness.

Since the mother of the index case presented with a phenotype similar to that of her son and carried the same mutation, it is another item of missing information whether she developed a complex type of HSP4 during follow-up, as age at onset of the clinical manifestations was the same in both patients.

Lastly, it should be mentioned that at least SPG1 to SPG79 (i.e., not only to SPG72) have been identified thus far, and that >65 HSP proteins (i.e., not only 50) have been detected.[7,8]

Overall, this interesting case report would have been more meaningful if long-term follow-up data had been provided, as well as the therapeutic measures applied to the index case and his mother.

Conflicts of Interest

The author has no potential conflicts of interest to disclose.

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Correspondence

Josef Finsterer, MD, PhD
Krankenanstalt Rudolfstiftung,
Messerli Institute,
Veterinary University of Vienna,
Postfach 20, 1180 Vienna, Austria
Tel +43-1-71165-72085
Fax +43-1-4781711
E-mail fifigs1@yahoo.de

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