



Corrigendum: Parental Somatic Mosaicism Uncovers Inheritance of an Apparently De Novo GFAP Mutation

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A Corrigendum on

Parental Somatic Mosaicism Uncovers Inheritance of an Apparently De Novo GFAP Mutation
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In the original article, there was an error in the **Introduction** section. The phrase “siblings of parents” seems to imply the aunts and uncles of the affected child, rather than the siblings of the child, as it should be. A correction has been made in the **Introduction** section:

Alexander disease (AxD) is an extremely rare, untreatable, and usually fatal neurodegenerative disorder (OMIM #203450), classified among leukodystrophies due to white matter deficits (Messing and Brenner, 2020). It is estimated to affect 1:2.7 million people in Japan (Yoshida et al., 2011). The disease presents at different ages of onset, with distinct symptoms and prognosis: in neonates and early childhood (type I) and later, though not restricted to adulthood (type II) (Prust et al., 2011). AxD is caused by heterozygous mutations of glial fibrillary acidic protein (*GFAP*) gene, which eventually lead to the formation of aggregates, also containing alphaB-crystallin, HSP27, ubiquitin, and proteasome components (Quinlan et al., 2007). To date, a broad spectrum of pathogenic *GFAP* variants accounts for more than 90% of patients. Mutations occur either *de novo* or through transmission from the parental generation. A recurrent occurrence of the same disease-causing *GFAP* mutation in siblings from parents who tested negative for the variant strongly suggests the presence of a germline mosaicism (Melchionda et al., 2013) (two affected siblings were also reported by Namekawa et al. (2002), but the parents were not examined). Indirect evidence for germline mosaicism in *de novo* AxD cases has also been provided by studies finding that the *de novo* mutations predominantly arise on the paternal chromosome (Li et al., 2006; Zang et al., 2013). Such a condition may be associated with somatic mosaicism, a circumstance nevertheless unproven so far (Messing, 2018). In the case of AxD, the risk of transmitting a *GFAP* mutation to a second child by germline mosaicism has been estimated as less than 1% (Messing, 2018); however, when significant somatic mosaicism is observed in a parent, the risk of recurrence could be substantially higher.

Also, a reference “**Li et al., 2006**” was cited but was not included in the reference section. A correction has been made to the **Reference** list.

The authors apologize for this error and state that this does not change the scientific conclusions of the article in any way. The original article has been updated.

REFERENCE

Li, R., Johnson, A. B., Salomons, G. S., van der Knaap, M. S., Rodriguez, D., Boespflug-Tanguy, O., et al. (2006). Propensity for Paternal Inheritance of *De Novo* Mutations in Alexander Disease. *Hum. Genet.* 119 (1–2), 137–144. doi:10.1007/s00439-005-0116-7

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