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LETTER TO THE EDITOR

'Comment on the Paper "SLC20A2 and THAP1 deletion in familial basal ganglia calcification with dystonia" by Baker et al.'

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Dear Editor,

I read with great interest the recent article by Baker et al., 2013, reporting a patient with dystonia, bilateral brain calcifications, and carrying a remarkable heterozygous deletion (~500 Kb), covering various interesting genes [1].

The authors highlight SLC20A2 and THAP1, both associated with familial brain calcification and dystonia, respectively. Curiously, patients with brain calcification can also exhibit dystonia. In this case, it is impossible to be sure if this deletion had an additive effect or not. However, I would like to call your attention to a small mistake in Figure 3. The gene *DKK3* belongs to chromosome 11, not 8. *DKK4* is located on chromosome 8 and should appear in this article.

A growing number of reports are identifying subjects with brain calcifications bearing mutations in the SLC20A2 gene. Besides the recent discovery of two other genes (*PDGFB* and *PDGFRB*), most cases are linked to SLC20A2 which encodes a highly and widely expressed inorganic phosphate transporter [2].

Fifty percent of families tested so far were not associated with these three genes, reinforcing the fact that this condition is polygenic, clinically heterogeneous, and likely underdiagnosed. This highlights the need to increase clinical and neuroimaging awareness in order to increase detection.

References

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2. Lemos RR, Ferreira JB, Keasey MP, Oliveira JR (2013) An update on primary familial brain calcification. Int Rev Neurobiol 110C:349–371. doi:10.1016/B978-0-12-410502-7.00015-6

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