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LETTER

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Response: Sotos Syndrome and the added value of genetic workup in epilepsy surgery

To the editors:

I thank Bättig et al for their very interesting letter in response to our paper, "Seizures in Sotos syndrome: Phenotyping in 49 patients."[1] They recently described a patient with drug-resistant focal epilepsy and a diffuse astrocytoma, who continued to have seizures following resection of the lesion.[2] The patient was subsequently diagnosed with Sotos syndrome and found to carry a pathogenic variant in *NSD1*. Sotos syndrome is associated with an increased risk for malignancy, though cerebral tumors are extremely rare. Our cohort did not include any patients in whom seizures occurred due to an intracranial neoplastic process.

Interestingly, of the other two patients with Sotos syndrome and brain tumors described in the literature, one was a boy with intractable epilepsy that also continued *after* resection of a ganglioglioma.[3] The other was an adult male with a low-grade glioma identified when he presented with left face and arm paresthesia; the authors noted that he had a past history of generalized tonic-clonic seizures, but it was not clear that these were thought related to the neoplastic process.[4] Based on this admittedly very small sample, it appears that for Sotos syndrome patients with brain tumors and epilepsy, resection of the lesion often has little to no impact on seizure control.

The experience of Bättig et al highlights the importance of genetic testing as part of an epilepsy presurgical workup. Although in many cases a genetic diagnosis will not change the recommendation to proceed with surgery, the finding of an underlying genetic condition that predisposes to epilepsy will change the counseling of the individual and his/her family regarding the long-term prognosis. At present, genetic testing is performed in a relatively small fraction of patients undergoing epilepsy presurgical evaluation.[5] However, given that the cost of an epilepsy gene panel is relatively small in the context of the financial investment made in patients considered for epilepsy surgery, it is probably reasonable that genetic testing becomes a standard element of such evaluations.

CONFLICT OF INTEREST

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ETHICAL PUBLICATION STATEMENT

I confirm that I have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

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