LETTER

Paraganglioma syndrome type 4 presenting as hypertensive encephalopathy in an 8-year-old boy

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Dear Editor,

Paragangliomas (PGL) are rare catecholamine-secreting tumors deriving from the paravertebral ganglia[1,2]. They are often associated with genetic mutations in the succinate dehydrogenase (SDH) complex (*SDHB> SDHD> SDHC* genes)[3]. There are only a few *SDHB* mutation-related cases described in children. We report the case of an 8-year-old boy with PGL type 4 syndrome presenting as an emergency with status epilepticus due to hypertensive encephalopathy, which is an unusual presentation of catecholamine excess. The disease progress was typical with recurring tumors and metastases. This case contributes to physicians' awareness of the non-specific symptoms of catecholamine secreting tumors and points out the severity of the disease in patients with *SDHB* mutations and the importance of genetic screening.

The boy presented with generalized seizures and hypertension and, on admission, went over to status epilepticus. His symptoms had started ten months before; he developed mild headaches, dizziness, and lately nocturnal enuresis. Symptoms of a catecholamine-producing tumor in children can occasionally be non-specific (such as blurred vision, polyuria, polydipsia, and behavioral problems). No information regarding family history was available, as the patient was adopted.

Laboratory investigations showed massively elevated 24-hour urine normetanephrine and dopamine. A magnetic resonance imaging of the abdomen revealed a 4.0 x 4.5 x 3.0 cm extra-adrenal paraganglioma, which was laparoscopically excised. Eighteen months later, symptoms recurred, and three new paragangliomas were excised laparoscopically. DNA analysis showed that the patient was heterozygote of the P254fsX255 germline mutation of the *SDHB*, a previously described mutation[1,3]. Because of the high incidence of hereditary PGL, genetic counseling and screening for mutations are mandatory to improve the follow-up of the patients and detect asymptomatic mutation carriers among relatives.

Three years after the second operation, a tumor on the left adrenal was excised by open abdominal surgery, showing malignant transformation and metastatic lesions in two thoracic vertebrae. It is typical for *SDHB*-related PGL to recur often[2,3] and have a high malignant potential (35-70 %)[1-3]. Size of the primary tumor more than 4.5 cm is related to earlier development of metastases, whereas patients diagnosed at a younger age seem to have improved survival[3]. The patient showed a mild disease progression over the next nine years following the diagnosis of metastatic paraganglioma.

In conclusion, this is a case report of a boy with PGL type 4 carrying an *SDHB* gene mutation. The patient presented with signs of hypertensive encephalopathy, which is unusual and should be included in the differential diagnosis of children presenting with seizures. Early diagnosis in children is important, as catecholamine hypersecretion in these tumors is related to high cardiovascular morbidity and mortality. Furthermore, patients have an increased risk for recurrence and malignant transformation and require lifelong monitoring. Genetic counseling and testing of family members to detect early asymptomatic disease carriers among relatives are mandatory.

Keywords: Paraganglioma, succinate dehydrogenase B mutation, child, catecholamines

Conflict of interest
Authors declared no conflict of interest.

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