Paraganglioma in SDHB mutation

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A 13 year old female presented with headaches. Physical examination was significant for elevated blood pressure, ranging from 150/89 to 186/105. Heart rate was also noted to be elevated, up to 135 bpm.

Family history is positive for pheochromocytoma in her maternal grandfather, who had hypertension since his late teens and died at age of 49 from complications of pheochromocytoma.

The young patient had elevated serum metanephrines (2866 pg/mL, normal < 149), urine catecholamines (927 mcg/24 hours, normal < 100), norepinephrine (906 mcg/24 hours, normal < 80), and chromogranin A (88 ng/mL, normal 1.9-15).
MRI demonstrated a 4.5 cm mass at the aortic bifurcation consistent with a paraganglioma arising from the organ of Zuckercandl.

T2W fat saturated image demonstrates a well-circumscribed lesion (arrowheads) with intermediate-to-hyperintense signal intensity just below the aortic bifurcation.
T1W fat saturated images before (A) and after contrast administration demonstrate intense arterial enhancement (B) with heterogeneous washout on venous (C) and delayed (D) images.
The lesion was surgically resected, and pathology was consistent with paraganglioma. The patient remains free of disease 5 years after resection.

The patient underwent genetic testing, which showed a heterozygous mutation in SDHB (succinate dehydrogenase subunit B), consistent with hereditary paraganglioma-pheochromocytoma syndrome.

Succinate dehydrogenase (SDH) has an essential role in the electron transport chain of mitochondria, and also plays a role in the citric acid cycle.
Paragangliomas in the skull base, neck, and upper mediastinum tend to be of parasympathetic origin and non-secretory.

Paragangliomas in the lower mediastinum, abdomen, and pelvis are typically of sympathetic origin and secretory.

Hereditary paraganglioma-pheochromocytoma syndrome is inherited in an autosomal dominant manner. In this setting, paragangliomas are more likely to become malignant, and prompt surgical resection is required. Patients should continue to have surveillance, and relatives at risk should be offered molecular genetic testing.
It has been classically thought that up to 10% of pheochromocytomas are hereditary, but increasing genetic studies demonstrate a much higher prevalence of hereditary paragangliomas and pheochromocytomas, possibly up to 41% of cases.

As such, hereditary paraganglioma-pheochromocytoma syndrome should be suspected in any patient diagnosed with paraganglioma or pheochromocytoma, and is highly suspected with early-onset, multifocal, or recurrent disease, and in patients with a family history of paraganglioma or pheochromocytoma.

https://www.ncbi.nlm.nih.gov/books/NBK1548/
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