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Familial paraganglioma syndrome: a rare cause of carotid artery occlusion

Rosafio F[a,*], Vandelli L[a], Dell'Acqua ML[a], Vallone S[b], Madeo B[c], Bigliardi G[a], Picchetto L[a], Zini A[a]
[a]Stroke Unit, Department of Neuroscience, [b]Department of Neuroradiology, [c]Department of Endocrinology, S. Agostino-Estense Hospital, Modena.

CASE REPORT

- 45-year-old man; history of occasional bilateral tinnitus and scintillanting scotoma in the last 5 years.
- Positive familial history for laterocervical space-occupying masses in paternal family.
- Admitted for sudden-onset of headache and visual loss.
- Neurologic examination: inferior altitudinal visual field impairment in the left eye.

- **Neuroimaging (CTA, Angiography and brain MRI): complete occlusion of the left ICA, bilateral carotid body paragangliomas (CBPs), more extended on the left; no parenchymal lesion** *(fig. 1, 2,3)*.
- Laboratory tests: increased levels of normetanephrines.
- Whole body Ga68-DOTA-NOC-PET: tracer accumulation in the laterocervical areas, in the left adrenal gland and para-aortic lymph node.

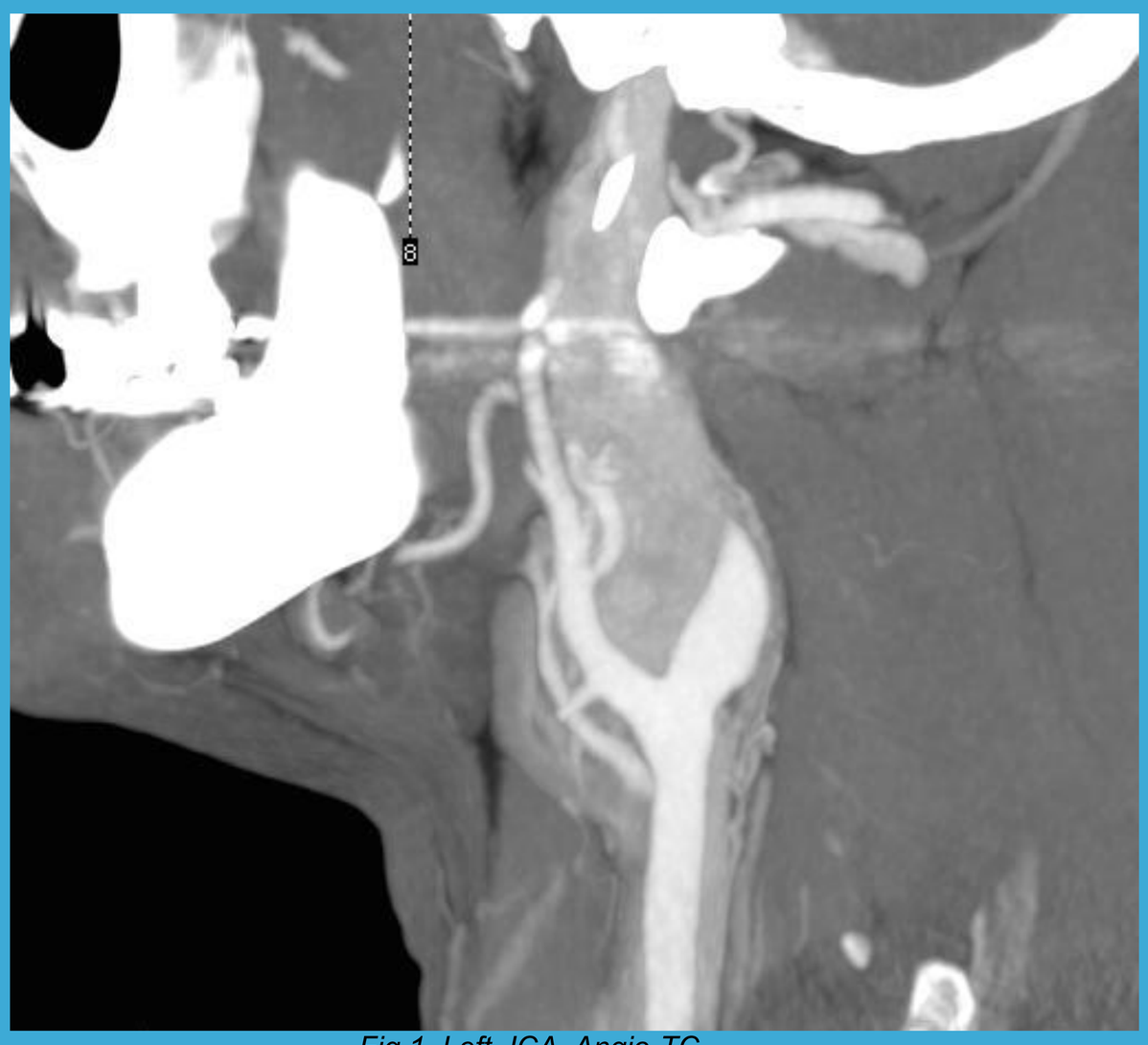

Fig 1. Left ICA. Angio-TC

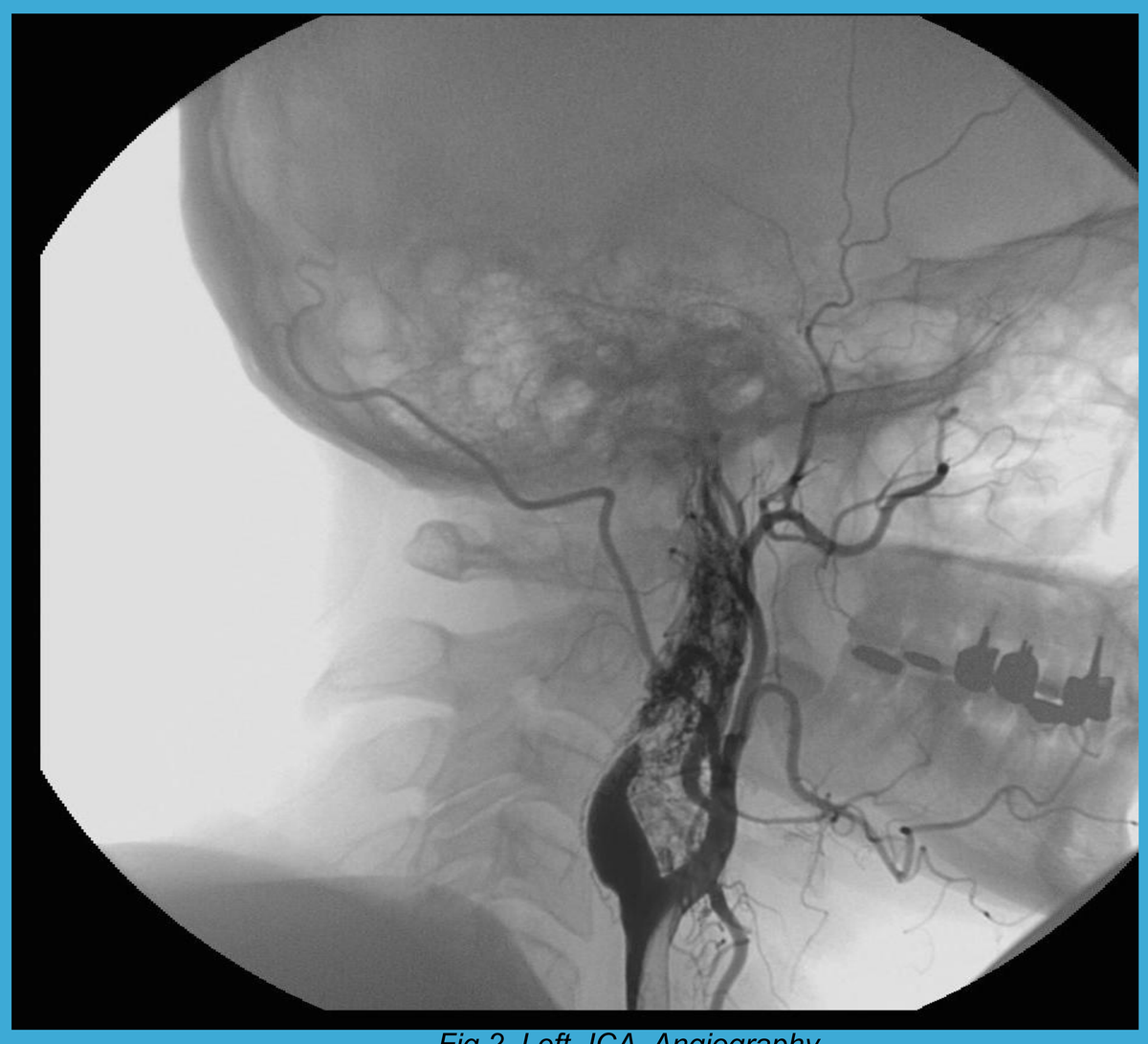

Fig 2. Left ICA. Angiography

Our patient presented with an acute retinal ischemia secondary to left ICA occlusion, likely due to paraganglioma. Later he developed hypertension and underwent left adrenectomy. The molecular analysis revealed mutation in succinate dehydrogenase subunit D (SDHD) gene, consistent with a familial pheochromocytoma paraganglioma syndrome 1.

Fig 3. Bilateral paragangliomas. MRA

DISCUSSION

Head and neck paragangliomas are rare tumors arising from chromaffin cells of the autonomic paraganglia, most frequently within the covering of the carotid artery.

Carotid body paraganglioma are often clinically silent, typically presenting as indolent and **slowly enlarging mass in lateral neck**. When symptomatic, these lesions can cause malignant hypertension.

They can be occasionally bilateral and associated with secreting thorax and abdominal paraganglioma, in pheocromocitoma-paraganglioma syndromes.

CONCLUSION

We report a case of acute cerebrovascular disease cause by an aggressive carotid paraganglioma, as the first manifestation of a familial pheochromocytoma paraganglioma syndrome 1. We suggest to follow-up CBPs, in order to avoid symptoms due to mass and secreting effects and possible thrombotic complications.