



Dr Papaspyrou Konstantinos, MD, Otorhinolaryngologist, Athens, Greece

BACKGROUND: Paragangliomas of the head and neck are rare, mostly benign tumors. Approximately 10% to 15% of



paragangliomas are caused by mutations in the succinate dehydrogenase (SDH) genes B, C, or D. These are often multifocal as part of paraganglioma syndromes and hormone secreting, and malignant particularly associated with mutations in SDHB.

METHODS AND RESULTS: A 29-year-old man was seen with recurrent paraganglioma. The patient's father reportedly suffered from bilateral carotid body tumors. Imaging studies showed metastases in both lungs and the liver. There was no increased hormone production by the tumor. Sequence analysis of the SDH genes revealed a novel C to T nonsense mutation in the first exon of the SDHD gene (R17X).

CONCLUSIONS: A novel mutation in the SDHD gene associated with malignant paraganglioma is reported. This case underscores the relevance of family history and genetic analysis, thus permitting early detection of unaffected carriers. These have to be monitored clinically, biochemically and by imaging techniques.

Head Neck. 2008 Jul;30(7):964-9. Malignant paraganglioma caused by a novel germline mutation of the succinate dehydrogenase D-gene--a case report. Papaspyrou K, Rossmann H,

Malignant paraganglioma and novel germline mutation of the succinate dehydrogenase D-gene--a

Written by Dr Papaspyrou Konstantinos, Otorhinolaryngologist, Athens - Last Updated Saturday, 14 June 2014 23:32

Fottner C, Weber MM, Mann W, Lackner KJ, Helling K. Department of Otorhinolaryngology, Johannes Gutenberg-University Medical School, Mainz, Germany. papaspyrou@hno.klinik.uni-mainz.de

Papaspyrou K, Rossmann H, Fottner C, Weber MM, Mann W, Lackner KJ, Helling K. Malignant paraganglioma caused by a novel germline mutation of the succinate dehydrogenase D-gene--a case report. Head Neck. 2008 Jul;30(7):964-9.