Abstract

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Aberrant Kit-Mutated gastrointestinal stromal tumor in suspected Carney-Stratakis syndrome

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Abstract

We present the case of a 33-year Philippines patient with a synchronous presentation of paranganglioma and Gastrointestinal Stromal Tumor (GIST). Germline mutations in SDH subunits are known to predispose to both of these relatively rare tumors called the Carney-Stratakis Syndrome (CSS). Investigations revealed that patient have a germline heterozygous pathogenic SDHB-inactivating mutation. While his paraganglioma did show IHC absence of SDHB, his GIST displayed normal SDHB staining and instead was positive for an exon 9 mutation of KIT. This was confirmed by DNA sequencing. Additionally, prior large series have shown only gastric GISTs in CSS, while this patient's GIST was in the small intestine. The finding of a KIT-mutated GIST in the setting of a germline SDHB-inactivating mutation in what would otherwise be consistent with Carney-Stratakis Syndrome has not been described previously and highlights the need for better understanding of the function of SDH and its interplay with other tumor suppressors and oncogenes. Specifically, this case demonstrates the need for somatic mutation analysis of tumors even in the presence of known germline SDH mutations since additional actionable mutations such as KIT or PDGFRA can improve prognosis given the high therapeutic index of imatinib mesylate.

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