

SDH-deficient renal cell carcinoma: A case report associated with a novel germline mutation

Vassilis Milionis¹, Dimitrios Goutas², Dimitrios Vlachodimitropoulos³, Andreas C. Lazaris², Iason Kyriazis⁴, E Liatsikos⁵, Nikolaos Marinakis⁶, Joanne Traeger-Synodinos⁷, and Nikolaos Goutas³

¹Istomedica S.A

²First Department of Pathology, School of Medicine, The National and Kapodistrian University of Athens

³2. Laboratory of Forensic Medicine and Toxicology, The National and Kapodistrian University of Athens

⁴Univ Patras

⁵University of Patras

⁶5. Laboratory of Medical Genetics, National and Kapodistrian University of Athens, St. Sophia Children's Hospital, 11527, Athens, Greece

⁷Aghia Sofia Childrens Hosp

April 6, 2021

Abstract

Routine examination of an asymptomatic 40-year-old female patient revealed a right unilateral and unifocal renal mass. The patient underwent a partial nephrectomy, and the renal specimen was sent for histopathologic examination. Molecular testing revealed a heterozygous variant NM_003000.3:c.412G>T, p.(Asp138Tyr), in SDHB gene.

Hosted file

SDHB.pdf available at <https://authorea.com/users/406154/articles/516928-sdh-deficient-renal-cell-carcinoma-a-case-report-associated-with-a-novel-germline-mutation>