

# Succinil Dehydrogenase A mutation in a 63 years old patient with wildtype GIST tumor

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**INTRODUCTION:**Gastrointestinal Stromal Tumors (GIST) are the most common soft Tissue Sarcoma of the Gastrointestinal Tract. Most of them result from KIT or PDGFRA activating mutations. Approximately 10 – 15% of GISTs have no detectable KIT or PDGFRA mutations: these tumors are called “Wild Type GISTs and are more likely to be associated with Carney Complex, the Carney-Stratakis Syndrome or NF1. Around-10-20% of wtGIST present a constitutional mutation mutation in one subunit of the SDH complex<sup>1</sup>.

**MATERIALS AND METHODS:** We present THE case re ofA 63years old male 63 with multiple subcutaneous nodular lesions, gastric wt-GIST, multiple lung nodules and peripancreatic mass. Two years earlier he had a chest benignmesenchymal tumor removed, and twenty years earlier he had a benign mass removed from the scalp. He also presented multiple skin lesions but did not fulfill NF1 diagnosis criteria. In order to orientate the diagnosis we performed sequencing and MLPA analysis of PRKAR1A, because it is altered in up to 60% of Carney’s complex cases. Providing that the result was normal, we followed with the study of mitochondrial complex, and this time we founded a SDHA mutation in heterozygosis: c.221dup (p.Leu74Phefs\*9). This mutation has been previously described in Paraganglioma type V.

**DISCUSSION:** We present a case of a SDHA germline mutation in a 63 years old patient with gastric GIST. The presence of dermal and subdermal lesions in the Neurofibromatosis spectrum warrants the need to rule out germline variations of NF1. In addition the constitutional SDHA mutation generates the need for genetic counselling and extensive follow up in first degree relatives.

1-Pantaleo ,MA et al. Eur J. Hum. Genetics, (2014),22-32-29