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MITF-related melanoma and renal cell carcinoma predisposition syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. MITF-related melanoma and renal cell carcinoma predisposition syndrome. ORPHA:293822

MITF-related melanoma and renal cell carcinoma predisposition syndrome is an inherited cancer-predisposing syndrome due to a gain-of-function germline mutation in the MITF gene, associated with a higher incidence of amelanotic and nodular melanoma, multiple primary melanomas and increase in nevus number and size. It may also predispose to co-occurring melanoma and renal cell carcinoma and to pancreatic cancer.