DISEASE:
BAP1-related tumor predisposition syndrome

NAME:	BAP1-related tumor predisposition syndrome
DESCRIPTION:	BAP1-related tumor predisposition syndrome (TPDS) is an inherited cancer-predisposing syndrome, associated with germline mutations in BAP1 tumor suppressor gene. The most commonly observed cancer types include uveal melanoma, malignant mesothelioma, renal cell carcinoma, lung, ovarian, pancreatic, breast cancer and meningioma, with variable age of onset. Common cutaneous manifestations include malignant melanoma, basal cell carcinoma and benign melanocytic BAP1-mutated atypical intradermal tumors (MBAIT) presenting as multiple skin-coloured to reddish-brown dome-shaped to pedunculated, well-circumscribed papules with an average size of 5 mm, histologically predominantly composed of epithelioid melanocytes with abundant amphophilic cytoplasm, prominent nucleoli and large, vesicular nuclei that vary substantially in size and shape.
ORPHACODE:	289539
SYNONYMS:	Tumor susceptibility linked to germline BAP1 mutations
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	BAP1
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- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- BRCA1 associated protein 1

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- Hereditary predisposition to cancer (47 genes) - IPG
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