



**NeuroEndocrine  
Cancer** Australia

# **Associated Conditions**

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## **Von Hippel–Lindau syndrome (VHL) (a syndrome is a group of symptoms that commonly occur together)**

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Von Hippel Lindau (VHL) is an inherited cancer syndrome caused by mutations of the VHL gene. It affects both sexes equally, About one in every 30,000 to 40,000 people get this syndrome.

VHL causes retinal (eye) haemangiomas, cerebellar and spinal cord haemangioblastomas, renal cell carcinomas (RCC) and pheochromocytomas.

Cystic masses in the pancreas are common and 10–15% of VHL patients develop islet cell neuroendocrine tumours.

## **Neurofibromatosis type 1**

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Neurofibromatosis type 1 is a relatively common inherited disorder that affects 1 in every 3000 people.

Patients usually have a particular skin pigmentation ('café au lait spots'), neurofibromas (nodules on the skin) and bone deformities, including scoliosis of the spine, and are at risk of bone fractures due to osteoporosis.

Patients with neurofibromatosis type 1 are prone to developing both benign and malignant tumours. These tumours include:

- brain and eye tumours (glioma)
- tumours of the nerves
- gastrointestinal stromal tumours (GIST)
- pheochromocytoma
- small bowel (duodenal) NET
- breast cancer, leukaemia, sarcomas.
- pancreatic tumours

## Tuberous sclerosis

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Tuberous sclerosis (TS) is an inherited condition characterised by benign growths in the skin, brain, kidneys, lungs and heart, which can affect how these organs function.

People with TS may be at risk of developing insulinomas—a neuroendocrine tumour of the pancreas.

## Multiple endocrine neoplasia (MEN syndrome)

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In Multiple Endocrine Neoplasia (MEN) there are tumours in two or more of the endocrine glands. There are four major forms of MEN. They may be inherited (autosomal dominant) or sporadic.

- **MEN 1:** Most people with MEN 1 develop parathyroid tumours (hyperparathyroidism; others develop pancreatic NETs or tumours in the pituitary gland. Other MEN 1 tumours include adrenocortical tumours, thymic NETs and gastric NETs. Patients and their families are advised to have genetic testing (MEN1 gene).
- **MEN 2** is a rare genetic (RET gene) syndrome that has three categories: 2A, MEN2B and medullary thyroid carcinoma (MTC).
  - **MEN2A** is characterised by the development of medullary thyroid carcinoma (MTC), pheochromocytoma and parathyroid adenomas.
  - **MEN 2B** patients develop MTC earlier in life, develop pheochromocytomas and neuromas of the skin and intestine. It is an aggressive form of MEN.
  - Familial medullary thyroid carcinoma (MTC) does not have the other tumours that are associated with MEN 2.



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