ONE-PAGE CHEAT SHEET

Inherited cancer syndromes

The genes behind family cancers, and the surgery that prevents them

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Free surgical teaching

RED FLAGS

- Cancer at a young age (under 50)
- Bilateral cancer or two primaries
- Several relatives on one side
- Rare cancers: male breast, medullary thyroid, adrenocortical

BREAST AND OVARY

- **BRCA1/BRCA2**: breast, ovary; BRCA2 also prostate, pancreas, male breast
- Risk-reducing mastectomy; salpingo-oophorectomy when the family is complete
- PARP inhibitors for BRCA-mutated cancers

BOWEL

- **Lynch** (MLH1, MSH2, MSH6, PMS2): right colon, endometrium, ovary, stomach, urinary tract
- Amsterdam 3-2-1; test every colorectal cancer for mismatch repair
- **FAP** (APC): thousands of polyps; prophylactic colectomy in late teens or twenties
- **MUTYH**: recessive polyposis
- **Peutz-Jeghers** (STK11): hamartomas, lip pigmentation

ENDOCRINE

- **MEN 1**: parathyroid, pancreas, pituitary
- **MEN 2A** (RET): medullary thyroid, phaeochromocytoma, parathyroid
- **MEN 2B** (RET): medullary thyroid, phaeochromocytoma, mucosal neuromas
- Prophylactic thyroidectomy timed by the RET mutation; exclude phaeo first
- **VHL**: kidney, phaeo, haemangioblastoma

OTHERS

- **Li-Fraumeni** (TP53): sarcoma, early breast, brain, adrenocortical; avoid radiation
- **CDH1**: diffuse gastric and lobular breast; prophylactic total gastrectomy
- **Cowden** (PTEN): breast, follicular thyroid, endometrium

WHAT TO DO

- Draw a three-generation family tree
- Refer to clinical genetics for counselling and testing
- Test the affected person first, then relatives for the known variant
- Most are autosomal dominant: 50% risk to each child

EXAM PEARLS

- Two-hit hypothesis: inherited cancers need only one more hit
- Germline = family; somatic = tumour only
- MSI-high tumours respond well to immunotherapy

Full page: **palsurgery.com/oncology/inherited-cancer-syndromes/** · Test yourself: **palsurgery.com/oncology/#quiz**

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