

**Clinical practice guidelines**

**Familial aspects of cancer:  
a guide to clinical practice**

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**NHMRC**

National Health and Medical Research Council

## APPENDIX C

### KNOWN CANCER PREDISPOSITION GENES

Hereditary syndrome	Commonest cancers	Mode of inheritance	Gene	Chromosome
Familial adenomatous polyposis	Colon, duodenum, periampullary	Dominant	APC	5q
Hereditary nonpolyposis colorectal cancer	Colon, endometrium, ovary, stomach, small bowel, renal tract, pancreas, biliary tract	Dominant	hMSH2 hMLH1 hMSH6 hPMS2	2p 3p 2p 7p
Peutz–Jeghers syndrome	Gastrointestinal tract, pancreas, ovary, testis, breast, uterus	Dominant	STK11	19p
Breast cancer, breast/ovarian cancer	Breast, ovary, prostate	Dominant	BRCA1 BRCA2	17q 13q
Melanoma	Melanoma, pancreas	Dominant	CDKN2A/ p16 <sup>INK4A</sup> CDK4	9p 12q
Li–Fraumeni	Sarcoma, breast, brain, leukaemia, adrenocortical	Dominant	p53	17p
Neurofibromatosis I	Neurofibrosarcoma, pheochromocytoma, optic glioma	Dominant	NF-1	17q
Von Hippel–Lindau syndrome	Haemangioblastoma of retina and central nervous system, renal cell carcinoma, pheochromocytoma	Dominant	VHL	3p
Multiple endocrine neoplasia type 1	Pancreatic islet, pituitary adenoma	Dominant	MEN1	11q
Multiple endocrine neoplasia type 2A and 2B	Medullary carcinoma of the thyroid, pheochromocytoma	Dominant	MEN 2A/ <i>RET</i> <i>RET</i>	10q 10q
Retinoblastoma	Retinoblastoma, osteosarcoma	Dominant	RB1	13q
Naevoid basal cell carcinoma syndrome (Gorlin syndrome)	Basal cell carcinomas, gliomas	Dominant	PTCH	9q
Wilms' tumour	Nephroblastoma	Dominant	WT1	11p
Ataxia telangiectasia	Leukaemia, lymphoma, breast, brain	Recessive	ATM	11q
Xeroderma pigmentosum	Skin, melanoma, leukaemia	Recessive	Various	Various
Tuberous sclerosis	Angiomyolipoma	Recessive	TSC2	16p
Cowden syndrome	Breast, thyroid, other	Dominant	PTEN	10q

<b>Hereditary syndrome</b>	<b>Commonest cancers</b>	<b>Mode of inheritance</b>	<b>Gene</b>	<b>Chromosome</b>
Bloom syndrome	Leukaemia, tongue, oesophagus, nephroblastoma, colon	Recessive	BLM	15q
Fanconi anaemia	Leukaemia, oesophagus, skin, hepatoma	Recessive	FAA, FAC	Various

This table is current at May 1999.