

Paediatric syndromes with cancer predisposition

Paediatric syndrome with cancer predisposition	Gene(s)	Expression	Key associated tumours	Hosted by	Latest update	Links to guidelines and/or additional information
Ataxia-telangiectasia	<i>ATM</i>	biallelic	85% of reported malignancy = lymphoreticular malignancy (most common B-cell non-Hodgkin lymphoma, ALL and CLL), brain, breast	AT Children's Project Ataxia-Telangiectasia Society (UK)	2008 2014 2015	atcp.org/additional-clinical-information/ atsociety.org.uk/wp-content/uploads/2017/10/A-T_Clinical_Guidance_Document_Final.pdf Bhatt et al. Eur Respir Rev 2015;24(138):565–581. doi:10.1183/16000617.0066-2015
Beckwith–Wiedemann syndrome	<i>CDKN1C</i> 11p15.5 methylation defect	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Birt–Hogg–Dubé syndrome	<i>FLCN</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Bloom syndrome	<i>BLM</i>	biallelic	Lymphoma, AML, ALL, MDS, Wilms, cutaneous SCC and BCC, oropharyngeal, upper gastrointestinal, colorectal, breast, lung	Bloom Syndrome Registry at Weill Cornell Medicine	2018	Cunniff et al. Am J Med Genet A. 2018 Sep;176(9):1872-1881. doi: 10.1002/ajmg.a.40374
Carney complex	<i>PRKAR1A</i>	monoallelic	primary pigmented nodular adrenocortical dysplasia, large cell calcifying Sertoli cell tumours, Leydig cell tumours, cutaneous myxoma, cardiac myxoma	Gene Reviews	2018	ncbi.nlm.nih.gov/books/NBK1286/
Congenital central hypoventilation syndrome	<i>PHOX2B</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au

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Constitutional mismatch repair deficiency (CMMRD)	<i>MLH1, PMS2, MSH2, MSH6</i>	biallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Costello syndrome	<i>HRAS</i>	monoallelic	rhabdomyosarcoma, neuroblastoma, bladder TCC, breast papilloma,	RASopathies Network	various	rasopathiesnet.org
Cowden syndrome (PTEN hamartoma syndrome)	<i>PTEN</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Diamond-Blackfan anemia	multiple	monoallelic X-linked (rare)	AML, MDS, osteosarcoma, colon	Diamond Blackfan Anaemia Foundation	various	dbaFoundation.org/resources/
Dyskeratosis congenita	multiple	>50% X-linked ~10% biallelic 5% monoallelic others unknown	Head and neck SCC, oesophageal SCC, AML	Team Telomere	2015	teamtelomere.org/wp-content/uploads/2018/07/DC-TBD-Diagnosis-And-Management-Guidelines.pdf
Familial adenomatous polyposis (FAP)	<i>APC</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Familial platelet disorder with predisposition to myeloid malignancy (FPDMM)	<i>RUNX1</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Fanconi anaemia	multiple	biallelic (most) X-linked (one)	AML, MDS, head & neck SCC, gynaecological SCC, other SCC	Fanconi Anaemia Research Fund	2020 (5th edition)	fanconi.org/explore/clinical-care-guidelines

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GATA2 deficiency syndrome	<i>GATA2</i>	monoallelic	See eviQ	Seattle Children's Hospital Bethesda, Maryland, USA eviQ	Unknown 2014 eviQ	seattlechildrens.org/conditions/gata2-deficiency/ Spinner, et al. Blood 2014;123:809-821 doi: 10.1182/blood-2013-07-515528 eviQ.org.au
Hereditary leiomyomatosis renal cell carcinoma (HLRCC, Reed syndrome)	<i>FH</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Howel–Evans syndrome (palmoplantar keratosis with oesophageal cancer)	<i>RHBDF2</i>	monoallelic	squamous cell carcinoma of the oesophagus	none identified	n/a	n/a
Juvenile polyposis of infancy (10q23 deletion)	<i>PTEN, BMPR1A</i>	dominant	gastrointestinal juvenile polyps and cancer	none identified	n/a	pubmed.ncbi.nlm.nih.gov/35487791/
Li–Fraumeni Syndrome	<i>TP53</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Mosaic variegated aneuploidy	<i>BUB1B</i>	biallelic	embryonal rhabdomyosarcoma, Wilms	none identified	n/a	n/a
Multiple endocrine neoplasia type 1	<i>MEN1</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Multiple endocrine neoplasia type 2	<i>RET</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au
Multiple endocrine neoplasia type 4	<i>CDKN1B</i>	monoallelic	See eviQ	eviQ	eviQ	eviQ.org.au

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Neurofibromatosis type 1	<i>NF1</i>	monoallelic	See eviQ	eviQ Children's Tumour Foundation Australia	eviQ various	eviq.org.au ctf.org.au
Neurofibromatosis type 2	<i>NF2</i>	monoallelic	See eviQ	eviQ Children's Tumour Foundation Australia	eviQ various	eviq.org.au ctf.org.au
Nevoid basal cell carcinoma syndrome (Gorlin syndrome)	<i>PTCH1, SUFU</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Nijmegen breakage syndrome	<i>NBN</i>	biallelic	lymphoid malignancy	none identified	n/a	n/a
Noonan syndrome	multiple	monoallelic (most)	juvenile myelomonocytic leukemia (<i>PTPN11</i> gene), ALL, AML	RASopathies Network	various	rasopathiesnet.org
Peutz–Jeghers syndrome	<i>STK11</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Rhabdoid tumour predisposition syndrome	<i>SMARCA4, SMARCB1</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Rothmund–Thomson syndrome	<i>ANAPC1, RECQL4</i>	biallelic	osteosarcoma, basal cell carcinoma, skin SCC	none identified	n/a	n/a

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Shwachman-Diamond syndrome	<i>SBDS</i> , <i>DNAJC21</i> , <i>EFL1</i> <i>SRP54</i>	biallelic monoallelic	AML, MDS, ALL	Schwachman Diamond Syndrome Foundation	various	shwachman-diamond.org/downloads/
Simpson–Golabi–Behmel syndrome	<i>GPC3</i>	X-linked	Wilms, neuroblastoma	none identified	n/a	n/a
Tuberous sclerosis complex	<i>TSC1</i> , <i>TSC2</i>	monoallelic	cardiac rhabdomyoma, renal angiomyolipoma	Tuberous Sclerosis Australia	Various	tsa.org.au/health-professional/
				UK Consensus Guidelines	2019	Amin et al. QJM 2019;112(3):171-182. doi: 10.1093/qjmed/hcy215
				International Tuberous Sclerosis Complex Consensus Conference	2013	Krueger et al. Pediatric Neurology 2013;49:255-265. doi: 10.1016/j.pediatrneurol.2013.08.002
Von Hippel–Lindau syndrome	<i>VHL</i>	monoallelic	See eviQ	eviQ	eviQ	eviq.org.au
Xeroderma pigmentosa	multiple	biallelic	cutaneous melanoma, BCC & SCC	Team Telomere	2015	teamtelomere.org/resources xps.org

Last updated: 06/06/2022

Review due: 22/02/2023

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