

General practitioner referral guidelines for cancer genetics assessment

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In this document the terms 'male' and 'female' refer to sex assigned at birth. For more information about providing individualised care refer to [Resources to assist in the care of transgender individuals referred to a clinical genetics service or familial cancer centre](#)

Guideline

Referral to a [clinical genetics service or familial cancer centre](#) for assessment should be considered for all people meeting the categories below:

Family history of cancer

Cancer gene pathogenic variant in a blood relative (e.g. BRCA1, BRCA2, PALB2, MLH1, MSH2, MSH6, PMS2, APC, or other)

Strong family history on the same side of the family:

- THREE 1st or 2nd degree relatives with same/related cancers* **OR**
- TWO 1st or 2nd degree relatives with same/related cancers,* one diagnosed under age 50 years

For unaffected females with breast cancer family history: see [iPrevent breast cancer risk assessment tool](#)

For unaffected people with colorectal cancer family history: see [Cancer Council Australia - Colorectal cancer risk according to family history](#)

**For example, breast and ovarian are related cancers. Lynch syndrome-related cancers include colorectal, endometrial, ovarian, renal, gastric, small bowel, pancreas, brain and cholangiocarcinoma.*

Personal history of cancer

For an individual with cancer, the following features may indicate referral to a familial cancer centre: early age of diagnosis, multiple primaries, strong family history (on the same side of the family), specific pathology features. See relevant eviQ referral guideline below for further details.

Cancer type	eviQ referral guidelines
Breast	Breast cancer – referring to genetics
Colorectal or polyposis	Colorectal cancer or polyposis – referring to genetics
Endocrine	Endocrine cancer and tumours – referring to genetics
Endometrial	Endometrial cancer – referring to genetics
Gastric	Gastric cancer – referring to genetics
Kidney	Kidney cancer – referring to genetics
Ovarian (including fallopian tube and primary peritoneal)	Ovarian cancer – referring to genetics
Paediatric	Paediatric cancer and tumours – when a genetic assessment is indicated
Prostate	Prostate cancer – referring to genetics
Skin	Skin cancers and lesions – referring to genetics

Website resources

History

Version 8

Date	Summary of changes
07/06/2023	Document updated with the following changes made: <ul style="list-style-type: none"> • Minor formatting changes • Personal history of cancer: 'Prostate cancer - referring to genetics' added to the table. Version increased to V.8. Review in 2 years.
15/09/2023	Term change from "women" to "females". Terminology flag added.

Version 7

Date	Summary of changes
23/01/2023	First sentence updated to align with the new eviQ cancer genetics referral guidelines template: <ul style="list-style-type: none"> • Changed from "People who fit any of the following warrant a referral to a clinical genetics service or familial cancer centre for genetic counselling and risk management advice.." to "Referral to a clinical genetics service or familial cancer centre for assessment should be considered for all people meeting the categories below". Version changed to V.7.

Version 6

Date	Summary of changes
26/11/2021	Document updated with the following change: <ul style="list-style-type: none"> • Personal history of cancer: 'Skin cancers and lesions - referring to genetics' added to table. Version changed to V.6.

Version 5

Date	Summary of changes
12/10/2020	Discussed at the May 2019 reference committee meeting. Approved on 12/10/20 for publication with the following changes: <ul style="list-style-type: none"> • Family history of cancer table: <ul style="list-style-type: none"> ◦ Cancer gene mutation in a blood relative: PALB2 added ◦ "Strong family history" changed to "Strong family history on the same side of the family" ◦ Row added to table: For unaffected women with breast cancer family history: see iPrevent breast cancer risk assessment tool (links to iPrevent website) ◦ Row added to table: For unaffected people with colorectal cancer family history: see Cancer Council Australia - Colorectal cancer risk according to family history (links to cancer wiki guidelines) ◦ Related cancers footnote moved to sit below this table • Personal history of cancer table <ul style="list-style-type: none"> ◦ Kidney, gastric, endocrine and paediatric added to cancer type column. ◦ Specific features warranting a referral removed from cancer type. Replaced with a general statement at the top of the table "For an individual with cancer, the following features may indicate a referral to a family cancer clinic: early age of diagnosis, multiple primaries, strong family history (on the same side of the family), specific pathology features. See relevant eviQ referral guideline for details." ◦ Referral guidelines titles changed and links updated

Date	Summary of changes
	<ul style="list-style-type: none"> "Mutation" changed to "pathogenic variant" throughout document for consistency among eviQ cancer genetics protocols per agreement among the cancer genetics reference committees' chairs. Definition of "pathogenic variant" added as a pop-up. <p>Version changed to V.5. Changes published on 17/11/2020.</p>

Version 4

Date	Summary of changes
17/08/2012	Presented and discussed at April 19, 2012 reference committee meeting. Approved for publication.
15/10/2012	Placed in new format.
30/04/2013	Format updated.
26/03/2014	Referral guidelines template discussed at March 5 2014 reference committee meeting, currently in the process of being reviewed and reformatted. Review of these guidelines taking place in conjunction with this process and will be discussed again at October 23 2014 reference committee meeting.
27/05/2014	Tumour pathology characteristics - Epithelial ovarian, fallopian tube or primary peritoneal cancer <60 yrs age changed to <70 yrs as per previous reference committee discussion and in accordance with eviQ - Cancer Genetics - Ovarian and endometrial cancer referral guidelines.
14/03/2017	Presented at the October 2016 RCM and review continued via email. <ul style="list-style-type: none"> Format updated as per template Links to other referral guidelines added Content updated <p>For 2 yearly review.</p>
31/05/2017	Transferred to new eviQ website. Version number changed to V.4.

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