

Criteria for Referral for a Cancer Genetics Evaluation

If any of the following apply, genetic counseling is appropriate. Genetic counseling may also be appropriate for individuals who do not meet criteria but are concerned about their history. A referral for genetic counseling does not always lead to a recommendation for genetic testing.

Common Cancers:

Anyone with a personal history or a close blood relative (first-degree relative (FDR) or second-degree relative (SDR) with:

Breast Cancer:

- Breast cancer at or under age 50.
- Breast cancer, triple negative, at any age.
- Breast cancer, bilateral or multiple tumors.
- Breast cancer that is recurrent or metastatic and HER2/Neu Negative.
- Breast cancer at any age and Ashkenazi Jewish Ancestry.
- Breast cancer, male.

Colon Cancer and Polyposis:

- Colorectal cancer under age 50.
- Ten or more Cumulative Tubular Adenomas.
- Three or more cumulative Hamartomatous polyps.

Uterine Cancer:

- Uterine cancer under age 50.

Ovarian Cancer:

- Ovarian, fallopian tube, or primary peritoneal cancer at any age.

Pancreatic Cancer:

- Pancreatic cancer at any age.

Prostate Cancer:

- Prostate cancer at any age with metastatic, Intraductal/Cribiform histology, or high- or very-high-risk group.

Renal Cancer:

- Renal cancer at or under age 46.
- Renal cancer with bilateral or multifocal tumors.
- Renal cancer at any age with at least one close relative with renal cancer.

Less Common Cancers/Tumors:

Anyone with a personal history or a close blood relative (FDR or SDR) with:

- Adrenocortical carcinoma.
- Choroid plexus carcinoma.
- Diffuse gastric cancer.
- Medullary thyroid cancer.
- Myelodysplastic Syndrome or Acute Myeloid Leukemia at or under age 50.
- Paraganglioma.
- Pheochromocytoma.
- Uveal melanoma at or under age 30.
- Uveal melanoma and another primary cancer.

Family History Indications:

Three or more close relatives on the same side of the family at any age with:

- Breast, ovarian, pancreatic, and/or prostate cancer.
- Colorectal, uterine, ovarian, and/or stomach cancer.
- Cutaneous melanoma, pancreatic cancer, renal cancer, breast cancer, astrocytoma, uveal melanoma, and/or mesothelioma.

Anyone with a close blood relative with a known pathogenic variant (positive mutation).

Pediatric Cancer:

- ANY child with two malignancies, one of those with onset younger than 18 years of age.
- ANY child with cancer and congenital anomalies.

Hematologic Malignancies:

- Hypodiploid Leukemia.
- Juvenile Myelomonocytic Leukemia.
- Myelodysplastic Syndrome.
- Bone marrow failure diagnosed younger than 12 years of age.

Pediatric Cancers (Continued):

Solid Tumors:

- Adrenocortical carcinoma.
- Atypical teratoid/rhabdoid tumor.
- Cardiac rhabdomyoma.
- Childhood skin cancer.
- Choroid plexus carcinoma.
- Ciliary body medulloepithelioma.
- Ependymoma (spinal cord).
- Hepatoblastoma.
- Juvenile granulosa cell tumor.
- Medulloblastoma.
- Nasal chondromesenchymal hamartoma.
- Optic glioma.
- Osteosarcoma.
- Paraganglioma/Pheochromocytoma.
- Pineoblastoma.
- Pituitary blastoma.
- Rhabdomyosarcoma.
- Sertoli-Leydig cell tumors.
- Subependymal giant cell astrocytoma.

Recommended Germline Testing of Tumor Variants For Solid Tumors:

Germline testing for variants identified in the following genes for:

- Any tumor type arising at any age: *BMPR1A*, *BRCA1*, *BRCA2*, *BRIP1*, *MLH1*, *MSH2*, *MSH6*, *MUTYH*, *PALB2*, *PMS2*, *RAD51C*, *RAD51D*, *RET*, *SDHA*, *SDHAF2*, *SDHB*, *SDHC*, *SDHD*, *SMAD4*, *STK11*, *TSC2*, and *VHL*.
(*Exclude renal tumors for *VHL*. Include germline testing of *MUTYH* only if two pathogenic mutations identified in tumor testing).
- An associated tumor type only, arising at any age: *BAP1*, *FH*, *FLCN*, and *POLE*.
- Any tumor type arising under age 30: *APC* and *RB1*.
- Any associated tumor type only, arising under age 30: *NF1* and *TP53*.
(*For *TP53*, brain tumors should be excluded.)
- For hematologic malignancies.
- Hematologic malignancy and one or more somatic variants in *RUNX1*, *CEBPA*, or *GATA2*.

National Comprehensive Cancer Network (NCCN) Guidelines:

Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate.

<https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1503>

Genetic/Familial High-Risk Assessment: Colorectal.

<https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1436>

Kidney Cancer.

<https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1440>

Neuroendocrine and Adrenal Tumors.

<https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1448>

Melanoma: Cutaneous.

<https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1492>

Melanoma: Uveal.

<https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1488>

Myelodysplastic Syndromes.

<https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1446>

Mandelker D, Donoghue M, Talukdar S, Bandlamudi C, Srinivasan P, Vivek M, Jezdic S, Hanson H, Snape K, Kulkarni A, Haqwkles L, Douillard JY, Wallace SE, Rial-Sebbag E, Meric-Bersntam F, George A, Chubb D, Loveday C, Ladanyi M, Berger MF, Taylor BS, Tumbull C. Germline-Focussed analysis of tumour-only sequencing: recommendations from the ESMO Precision Medicine Working Group. *Ann Oncol*. 2019 Aug 1;30(8):1221-1231. Doi: 10.1093/annonc/mdz136. Erratum in: *Ann Oncol*. 2021 Aug;32(8): 1069-1071. PMID: 31050713;

PMCID: PMC6683854.

To access the contact information for all Ohio Cancer Risk Assessment sites, please visit:

<https://odh.ohio.gov/know-our-programs/genetic-services/Ohio-Cancer-Genetics-Risk-Assessment-Sites>

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NOTE: This list is NOT exhaustive.

Refer to NCCN Guidelines for Additional Referral Recommendations based on personal and family history of a combination of different cancers.