

THE DIVISION OF MEDICAL GENETICS

Royal University Hospital, Saskatoon, Saskatchewan

Cancer Genetic Services

Cancer is a common disease affecting approximately 1 in 2 men and 1 in 3 women in their lifetime. 5-10% of cancer is thought to be caused by an inherited predisposition. Many different types of cancer may have a hereditary basis. Most cancers are sporadic and are caused by a combination of many factors including age, diet and environmental exposures.

A genetic assessment can help to determine the likelihood of an inherited predisposition to cancer and whether molecular testing for a specific cancer gene would be helpful. **For those with an increased likelihood of an inherited predisposition, testing must begin with a living affected family member.** Often we are able to reassure families that they are unlikely to be at increased risk for a cancer predisposition syndrome. For those families who are identified to have a mutation, surveillance recommendations have been published for known and possible carriers.

Once a mutation is identified in the family, testing can be offered to all at-risk family members. An individual who does not yet have cancer and participates in genetic testing for a known mutation is undergoing predictive testing. We follow a strict protocol for predictive testing which includes having the patient attend two separate appointments. We highly recommend that patients attend the appointments with a trusted support person, preferably someone who is not a blood relative. The purpose of the first appointment is to review the testing procedure as well as possible outcomes in order to discuss the risks, benefits, and choices regarding testing. If the person chooses to have the testing, a second appointment is arranged for disclosure if and when the patient chooses to receive them. Results will only be given in person.

It has been well documented that patients have strong emotional reactions to both positive and negative results. The Genetic Counsellor will help clarify the implications of the results, discuss future screening or treatments available and how other family members may benefit from genetic counselling and/or testing.

Here are some guidelines to help you decide which patients may benefit by a referral for Cancer Genetic Services. **If you are uncertain, please refer.**

CLUES SUGGESTIVE OF A HEREDITARY CANCER SYNDROME:

1. Early age of onset
2. Multiple primary tumours
3. Bilateral/Multifocal cancer
4. Multiple close (first or second degree) relatives with related cancers
5. Multiple generations affected – don't forget to ask about dad's side of the family!

Examples of some known hereditary cancer syndromes and associated cancers are listed below. This list is not exhaustive. We encourage you to refer a patient if you have any concerns that their personal or family history may be suggestive of a hereditary cancer syndrome.

Cancer Syndromes

BRCA1 and BRCA2

Hereditary Non-polyposis Colorectal Cancer

Familial Adenomatous Polyposis

Multiple Endocrine Neoplasias (MEN)

Familial Medullary Thyroid Carcinoma (FMTC)

Familial Malignant Melanoma

Prostate Cancer

Familial Lymphoma and Leukemia

Li-Fraumeni

von Hippel Lindau (VHL)

Retinoblastoma

Hereditary Diffuse Gastric Cancer

Examples of associated high risk cancers

Breast cancer, ovarian cancer, pancreatic cancer, prostate cancer

Colon cancer, endometrial cancer, ovarian cancer, gastric cancer, small bowel cancer, kidney/urinary tract cancer, biliary tract cancer, gallbladder cancer, CNS cancer

Colon cancer, CNS cancer, thyroid cancer, hepatoblastoma

Parathyroid cancer, pituitary cancer, thyroid cancer, pheochromocytoma

Medullary thyroid cancer

Melanoma, pancreatic cancer, breast cancer

Prostate cancer

Lymphoma and leukemia

Osteosarcoma, brain tumours, breast cancer, adrenal cortical tumours, lung cancer, leukemia, soft tissue sarcoma, pancreatic cancer, colon cancer, melanoma

Clear cell renal carcinoma, pheochromocytoma, retinal and CNS hemangioblastomas

Unilateral and bilateral retinoblastoma, pineal gland tumours

Diffuse gastric cancer, lobular breast cancer

*****Misinformation regarding pathology of a cancer diagnosis in families is very common. Confirmation of all relevant cases is required before any testing can proceed.**

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