

Turcot Syndrome

This section has been reviewed and approved by the [Cancer.Net Editorial Board \[1\]](#), December / 2011

Overview

What is Turcot syndrome?

Turcot syndrome is a condition in which cells become abnormal and form masses called polyps. A polyp is benign (noncancerous) but can eventually turn malignant (cancerous, meaning it can spread to other parts of the body). Turcot syndrome is rare and is considered to be an alternative form of two more common syndromes associated with polyp formations: [hereditary non-polyposis colorectal cancer \[2\]](#) (HNPCC) and [familial adenomatous polyposis \[3\]](#) (FAP).

People with Turcot syndrome have multiple adenomatous colon polyps (polyps in the colon made up of cells that form mucous), an increased risk of [colorectal cancer \[4\]](#), and an increased risk of [brain cancer \[5\]](#). The type of brain cancer generally depends on whether the Turcot syndrome is more similar to HNPCC or FAP. The two most common types of brain tumors in Turcot syndrome are:

- [Glioblastoma \[5\]](#). This type of brain tumor is a very aggressive form of [astrocytoma \[6\]](#) that is commonly found in families who have features of HNPCC.
- [Medulloblastoma \[7\]](#). This type of brain tumor begins in granular cells in the cerebellum (back of the brain). Medulloblastoma most often occurs in children and is commonly found in families who have features of FAP.

What causes Turcot syndrome?

Turcot syndrome is a genetic condition. This means that the risk of Turcot syndrome can be passed from generation to generation in a family. In families with glioblastoma and other features of HNPCC, mutations (alterations) have been found in two genes: *MLH1* and *PMS2*. In families with medulloblastoma and other features of FAP, mutations have been found in the *APC* gene.

How is Turcot syndrome inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. Turcot syndrome follows an autosomal dominant inheritance pattern, in which a mutation needs to happen in only one copy of the gene for the person to have an increased risk of getting that disease. This means that a parent with a gene mutation may pass along a copy of their normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having the same mutation.

How common is Turcot syndrome?

Turcot syndrome is considered to be rare.

How is Turcot syndrome diagnosed?

A person with multiple adenomatous colon polyps and/or [colorectal cancer \[4\]](#), along with either [glioblastoma \[5\]](#) or [medulloblastoma \[7\]](#), may have Turcot syndrome. Blood tests are available to look for the three genes linked to Turcot syndrome. People who may have Turcot syndrome can have a blood test to look for a mutation in the *APC* gene associated with FAP or the *MLH1* gene associated with HNPCC. They may also have testing for the mutation in *PMS2* if Turcot syndrome is suspected. If a specific gene mutation is found, other family members may also be diagnosed with Turcot syndrome if they are tested and have the same gene mutation. However, some families that appear to have Turcot syndrome may not have a detectable gene mutation.

What are the estimated cancer risks associated with Turcot syndrome?

The risks of cancer and other features depend on whether the Turcot syndrome appears to be more similar to [HNPCC \[2\]](#) or [FAP \[3\]](#); visit the sections for those conditions for a summary of cancer risks and other features.

What are the screening options for Turcot syndrome?

The screening options for Turcot syndrome are considered to be similar to those for [HNPCC \[2\]](#) or [FAP \[3\]](#), with the addition of screening for [brain cancer](#)

[5]. See those conditions' sections for a summary of screening recommendations.

There are no specific guidelines for the frequency and method of screening for [brain cancer](#) [5]. Individuals from families diagnosed with Turcot syndrome are encouraged to talk with a neurologist (a doctor who specializes in problems with the brain and central nervous system) about screening options.

Screening options may change over time as new technologies are developed and more is learned about Turcot syndrome. It is important to talk with your doctor about appropriate screening tests.

Learn more about [what to expect when having common tests, procedures, and scans](#) [8].

Questions to ask the doctor

If you are concerned about your risk of [colorectal cancer](#) [4] or [brain cancer](#) [5], talk with your doctor. Consider asking the following questions of your doctor:

- How many colon polyps have I had in total?
- What type of colon polyps have I had? (the two most common kinds are hyperplastic and adenomatous)
- What is my risk of developing [colorectal cancer](#) [4]?
- What is my risk of developing [brain cancer](#) [5]?
- What can I do to reduce my risk of cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think your family may have Turcot syndrome, consider asking the following questions:

- Does my family history increase my risk of [colorectal cancer](#) [4] or [brain cancer](#) [5]?
- Should I meet with a genetic counselor?
- Should I consider [genetic testing](#) [9]?

Additional resources

[Guide to Lynch syndrome](#) [2]

[Guide to Familial Adenomatous Polyposis](#) [3] (FAP)

[Guide to Colorectal Cancer](#) [4]

[The Genetics of Colorectal Cancer](#) [10]

[Guide to Brain Tumor](#) [5]

[What to Expect When You Meet With a Genetic Counselor](#) [11]

Colon Cancer Alliance

www.ccalliance.org [12]

National Cancer Institute

www.cancer.gov [13]

American Cancer Society

www.cancer.org [14]

CancerCare

www.cancercares.org [15]

To find a genetic counselor in your area, ask your doctor or visit the following websites:

National Society of Genetic Counselors

www.nsgc.org [16]

National Cancer Institute: Cancer Genetics Services Directory

www.cancer.gov/cancertopics/genetics/directory [17]

Links:

- [1] <http://www.cancer.net/about-us>
- [2] <http://www.cancer.net/node/19223>
- [3] <http://www.cancer.net/node/18852>
- [4] <http://www.cancer.net/node/18701>
- [5] <http://www.cancer.net/node/18562>
- [6] <http://www.cancer.net/node/18488>
- [7] <http://www.cancer.net/node/19237>
- [8] <http://www.cancer.net/node/24959>
- [9] <http://www.cancer.net/node/24895>
- [10] <http://www.cancer.net/node/24898>
- [11] <http://www.cancer.net/node/24907>
- [12] <http://www.ccalliance.org/>
- [13] <http://www.cancer.gov/>
- [14] <http://www.genetests.org/servlet/access?db=genestar&id=8888891&fcn=e&fcn2=refer&refer=http%3A%2F%2Fwww.cancer.org&type=resource&key=rqddkjp16MpW>
- [15] <http://www.genetests.org/servlet/access?db=genestar&id=8888891&fcn=e&fcn2=refer&refer=http%3A%2F%2Fwww.cancercare.org&type=resource&key=rqddkjp16MpW>
- [16] <http://www.nsgc.org/>
- [17] <http://www.cancer.gov/cancertopics/genetics/directory>