Turcot Syndrome

Turcot Syndrome is a disease that is characterized by Colonic Polyposis and a Malignant Central Nervous System (CNS) Tumor. Due to this there is an increased risk of Colorectal Cancer, and an increased risk of Brain Cancer. Turcot Syndrome demonstrates two types of inheritance patterns. An autosomal dominant inheritance is strongly associated with Familial Adenomatous Polyposis (FAP), but an autosomal recessive pattern is associated with Lynch Syndrome (HNPCC). If the Turcot Syndrome is more like Lynch Syndrome, the patient can develop a Glioblastoma Multiforme, but if it is more like Familial Adenomatous Polyposis (FAP), the patient can develop a Medulloblastoma.



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Pathophysiology

Autosomal Dominant or Autosomal Recessive
Domino and Recessive-chocolate Sign
When Turcot syndrome demonstrates an autosomal dominant inheritance pattern, it means an affected parent has a 50% chance of passing on the mutated gene (the disease) to their children. When Turcot syndrome demonstrates an autosomal recessive inheritance pattern, it means two mutated genes, one from each parent has been passed on to the affected individual. Two carrier parents have a 50 % chance of having an unaffected child who also is a carrier, and a 25% chance of having an affected child with two recessive genes.

Colonic Polyposis + CNS Tumor
Polyp-guy with Colon on Trench-coat and Malignant-man with Tumor-guy on Central-nervous-system
Turcot syndrome is characterized by colonic polyp growth that occur in association with a primary brain tumor.

Associations

Increased Risk of Colorectal Cancer
Up-arrow-risk and Car-gnome wearing Colon-on-hat
Patients with Turcot syndrome have an increased risk of developing colorectal cancer. The colonic growths associated in Turcot syndrome can cause bleeding from the rectum, diarrhea, constipation, abdominal pain, and/or weight loss.

Increased Risk of Brain Cancer
Up-arrow-risk and Tumor-guy on Brain
Patients with Turcot syndrome have an increased risk of developing brain cancer, commonly in the form of glioblastoma multiforme (GBM) or a medulloblastoma.

Lynch Syndrome
Lunch-lady
Turcot syndrome may be associated with Lynch syndrome, or hereditary nonpolyposis colorectal cancer (HNPCC). This is a hereditary cancer syndrome resulting from defective DNA mismatch repair genes, causing a predisposition to cancer growth via microsatellite instability.

Glioblastoma Multiforme
Glitter-blast Gladiator
Patients with Turcot syndrome who display an association with or additional diagnosis of Lynch syndrome commonly also have a glioma. GBM is the most common malignant tumor of glial cells in the brain and is derived from astrocytes.

Familial Adenomatous Polyposis (FAP)
Fat-guy
Turcot syndrome may be associated with familial adenomatous polyposis (FAP). FAP is an autosomal dominant disorder that results from a mutation in the adenomatous polyposis coli (APC) gene on chromosome 5 that also undergoes a “second hit” or “two-hit” deletion of the other allele, resulting in higher

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prevalence of colonic polyp development and associated colorectal carcinoma (CRC).

Medulloblastoma

Medusa-blast Sign

The type of brain cancer that a patient with Turcot syndrome develops when associated with FAP is a medulloblastoma. Medulloblastomas are considered one of the most common primary central nervous system (CNS) tumors in children. They tend to occur in the cerebellum and can present with multiple signs and symptoms including hydrocephalus and truncal ataxia.