Familial Adenomatous Polyposis

Opening Declaration: I promise to do this assignment authentically

Student Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date:\_\_\_\_\_\_\_\_\_

Affected Chromsome

Familial adenomatous polyposis (FAP) is a genetic disorder. FAP is caused by a mutation in the APC gene. According to the National Center for Biotechnology Information, the cytogenic gene location of this disorder is from 5q21 to 5q22, which includes the base pairs from 112,707,505 to 112,846,239 (NCBI). This translates to the APC gene being located on the fifth autosomal chromosome's longer arm between the 21st and 22st band when stained.

Symptoms of FAP

There are numerous symptoms of FAP. Classical FAP is characterized by non cancerous growths, or polyps, appearing at an early teenage age. These polyps generally appear in the large intestine and the rectum. This may lead to blood in the feces. If left untreated, the polyps may turn cancerous. In addition to colon cancer, other associated cancers are "cancer in the stomach and/or small intestine. Other types of cancer found in families with FAP include hepatoblastoma, a type of liver cancer seen in young children; desmoid tumors/desmoid fibromatosis, a locally aggressive tumor that does not metastasize; papillary thyroid cancer; pancreatic, adrenal, and bile duct tumors; and a type of brain tumor called medulloblastoma," (American Society of Clinical Oncology). Other non-cancerous symptoms are bony growths in the jaw, abnormal amount of teeth, changes in skin, and adrenal masses.   
 FAP is an autosomal genetic disorder. Any person with mutations in the APC gene will have a fifty percent chance of passing on the disease to their child. Since the APC gene can have varying mutations, different people will be affected differently by the disease. According to American Society of Clinical Oncology, "There are three subtypes of classic FAP called attenuated FAP (AFAP), Gardner syndrome, and Turcot syndrome," (ASCO).

Current Therapy

Current therapy for treatment of classical FAP begins with careful surveillance. Mohmmad Wehbi of the Medscape website recommends "Flexible sigmoidoscopy should be performed every 1-2 years starting at age 10-12 years in patients with FAP to document the onset of polyposis... Once polyps are detected, colonoscopic surveillance is recommended to remove large polyps in patients who have not had an operation," (Wehbi). To prevent the inevitable cancer that appears mainly the colon and sometimes the rectum, removal of these organs usually follows when the patient is in their late teens or early adulthood. Mayo Clinic lists three possible surgeries: Ileal pouch-anal anastomosis (J-pouch) surgery, total colectomy, and continent ileostomy (Mayo Clinic Staff). The medicine to take after this surgery is sulindac or celecoxib, which "may be beneficial in reducing the size and the number of adenomatous polyps in the remaining rectum," (Wehbi).

Limitations of Current Therapy

Being probed by a camera once every year or two is not a big problem. However removing such a large portion of the digestive system presents an issue. Any water and salt absorbed by the colon will not be absorbed. However the small intestine does absorb a great deal of water already. Ileal puch-anal anastomosis is a procedure where part of the small intestine is curved so it can store waste. A possible side effect is problems reproducing. The limitation of a total colectomy is that it is only useful if polyps did not appear in the rectum. In a continent ileostomy, a stoma is made where a bag to collect waste is connected to the small intestine. This carries a risk of the stoma becoming infected. The surgery is then followed up by sulindac or celecoxib. Celecoxib is no longer widely used because " of the association between cyclooxygenase 2 (COX-2) inhibitors (celecoxib is a member of this drug family) and coronary artery disease," (Wehbi). These drugs reduce the amount of polyps that may appear in the rectum.   
 These treatments are largely effective in preventing future cancer in the colon and rectum. However, all these treatments involve removal of a major organ, which most people would like to keep. These treatments also do not address the previously discussed increased chance of cancers in places besides the colon, such as the thyroid. It appears that the treatment eliminates the main problem with FAP, colonic cancer, but by some unknown mechanism, other non-colon related problems can still occur.

Works Cited

"APC Adenomatous Polyposis Coli." *National Center for Biotechnology Information*. National Center for Biotechnology Information, 6 Mar. 2016. Web. 13 Mar. 2016. <http://www.ncbi.nlm.nih.gov/gene/324>.

"Familial Adenomatous Polyposis." *Cancer.Net*. Ed. Cancer.Net Editorial Board. American Society of Clinical Oncology, 25 June 2012. Web. 13 Mar. 2016. <http://www.cancer.net/cancer-types/familial-adenomatous-polyposis>.

Mayo Clinic Staff. "Familial Adenomatous Polyposis." *Treatment at Mayo Clinic*. Mayo Clinic, 20 Oct. 2015. Web. 13 Mar. 2016. <http://www.mayoclinic.org/diseases-cMonditions/familial-adenomatous-polyposis/care-at-mayo-clinic/treatment/con-20035680>.

Wehbi, Mohammad. "Familial Adenomatous Polyposis Treatment & Management." *Familial Adenomatous Polyposis Treatment & Management: Medical Care, Surgical Care, Consultations*. Medscape, 6 Mar. 2015. Web. 13 Mar. 2016. <http://emedicine.medscape.com/article/175377-treatment>.

Closing Declaration: At the close of this assignment, I can attest to having done it by my own hand. If I received help from peers or from tutors in doing it, this was purely to understand the material, and I did not knowingly transfer the information from or to other sources (my peers or otherwise) in the process of doing this work

Student Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date:\_\_\_\_\_\_\_\_\_

http://www.ncbi.nlm.nih.gov/gene/324

http://www.cancer.net/cancer-types/familial-adenomatous-polyposis  
http://emedicine.medscape.com/article/175377-treatment

http://www.mayoclinic.org/diseases-conditions/familial-adenomatous-polyposis/care-at-mayo-clinic/treatment/con-20035680