Value of Routine Immunohistochemical (IHC) Analysis of Colorectal Cancer to Genetic Counseling Referrals

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Published In/Presented At

Galler, A., Fulmes, M., Park, J., Bub, D., Weiss, M. J., Namey, T., Augustyn, A. M., & Sinnott, R. (2012). Value of Routine Immunohistochemical (IHC) Analysis of Colorectal Cancer to Genetic Counseling Referrals. *LVHN Scholarly Works*. Retrieved from http://scholarlyworks.lvhn.org/surgery/33

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Introduction:

- Identification of patients with hereditary colorectal cancers (i.e. hereditary non-polyposis colon cancer (HNPCC)) is beneficial for implementing screening and interventions for high-risk families.
- Large scale screening for germline mutations, utilizing immunohistochemical (IHC) analysis of mismatch repair (MMR) proteins, has been shown to be feasible, efficient, and recommended for screening for all colorectal cancers.
- Referral to genetic counseling for high-risk individuals with germline mutations is recommended, but have historically been poor (6.9-21.9%).
- Lehigh Valley Health Network (LVHN) instituted routine IHC analysis of all colorectal cancers beginning in January 2011.

Purpose:

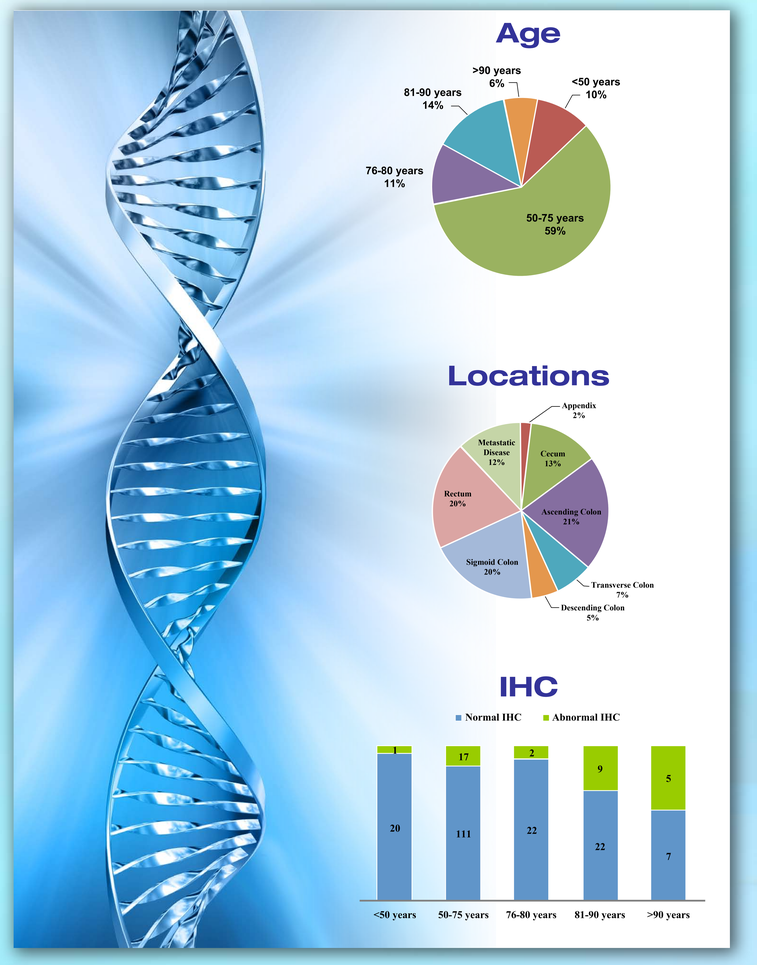
- Compare referral patterns to genetic counseling before and after initiation of routine IHC analysis.

Methods:

- All charts of patients diagnosed with colorectal cancer from July 2010 to June 2011 were retrospectively reviewed.
- Comparison of referral patterns to genetic counseling was performed for the six months prior to institution of routine IHC analysis with the six months following.

Results:

- 216 patients were diagnosed with colorectal cancer between July 2010 and July 2011.
- 12.6% (11/87) and 85.3% (110/129) of patients underwent IHC analysis before and after routine IHC testing, respectively.



Results (continued):

- 34 patients (15.7%) were found to have abnormal expression of MMR proteins.
  - Two patients were identified before and 32 patients after routine analysis
- 9.2% (8/87) of patients were referred before and 14.0% (18/129) of patients were referred to genetic counselin after routine IHC analysis (p=0.29).
- Two patients ultimately were diagnosed with HNPCC (0.9%).

	Pre-Routine IHC Analysis (%)	Post-Routine IHC Analysis (%)	Total (%)
Total Patients	87 (40.3)	129 (59.7)	216 (100)
IHC Analysis	11 (12.6)	110 (85.3)	121 (56)
Abnormal Expression MMR Proteins	2 (2.3)	32 (24.8)	34 (15.7)
Referral to Genetic Counseling	8 (9.2)	18 (14)	39 (18.5)
HNPCC	2 (2.3)	0	2 (0.9)

Conclusions:

- The initiation of routine IHC testing identified more patients with MMR protein non-expression; however, it did not improve overall referral rates for genetic counseling.
- Patient age at diagnosis, family history, and history of multiple tumors appear to be stronger factors towards genetic counseling referrals.
  - The value of routine IHC analysis for entire populations remains unclear.
- Further studies to evaluate initiatives to promote physician and patient education are needed.

References:

1. Hampel H, Frankel WL, Martin E, Arnold M, Khanduja K, et al. Screening for the Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer). NEJM 2005:352:1851-60.
2. De la Chapelle A, Hampel H. Clinical relevance of microsatellite instability in colorectal cancer. J Clin Oncol 2010:28:3380-3387.
3. Engstrom PF, Arnoletti JP, Benson AB. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines) Rectal Cancer. Version 3.2011, pg MS-4 – MS-5.
4. Sweet KM, Bradley TL, Westman JA. Identification and Referral of Families at High Risk for Cancer Susceptibility. J Clin Oncol 2002;20:528-537.
5. Hampel H, Sweet K, Westman JA, Eng C. Referral for cancer genetics consultation: a review and compliation of risk assessment criteria. J Med Genet 2004;41:81-91.
6. Wong C, Gibbs P, Johns J, Jones I, Faragher I, et al. Value of database linkage: are patients at risk of familial colorectal cancer being referred for genetic counseling and testing? Int Med J 2008:38;328-333.