A DEDICATED COLORECTAL CANCER GENETICS SERVICE IMPROVES ADHERENCE WITH MOLECULAR TESTING FOR LYCH SYNDROME

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Introduction:
Lynch Syndrome (LS) accounts for 2-3% of colorectal cancer (CRC); ~1000 cases of CRC in the United Kingdom annually. It occurs as a result of mutations in DNA repair genes, limiting DNA repair and causing Microsatellite Instability (MSI). Previous studies have demonstrated that in current practise less than 10% of these cases are identified as LS due to a lack of appropriate testing with immunohistochemistry or MSI analysis. The international Revised Bethesda Criteria were devised in 2004 to help identify such cases; these criteria include all individuals diagnosed <50 years of age, those with synchronous or metachronous CRC or LS-related cancer, those with significant family history of CRC or a LS-related cancer, or individuals diagnosed <60 years of age with MSI-type histology.

Methods:
We identified all new cases of colorectal cancer over a 1 year period prior to and subsequent to the establishment of a dedicated 'Family History of Bowel Cancer Service'. Adherence to the Revised Bethesda Criteria was determined by examination of medical records and UK National Bowel Cancer Audit Programme (NBOCAP) data. Pathology reports were studied in patients aged under 60 years of age at diagnosis looking for features consistent with MSI-H histology. We used Chi-squared testing to calculate significance for binary variables.

Results:
Over the two year period 198 cases of colorectal cancer were discussed at the CRC multidisciplinary meeting. 41 patients fulfilled the Revised Bethesda Criteria for screening for Lynch Syndrome; 12 individuals were diagnosed under the age of 50 years (~6%); 4 patients were diagnosed under 60 years of age and had MSI-H type histology and 25 patients had a significant family history of CRC or a LS-related cancer. In the year prior to the introduction of the clinic, we identified 18 cases meeting the Revised Bethesda Criteria for screening for LS; however, only 1 patient had been tested (5.6%). In contrast following the introduction of the clinic 19 of 23 identified cases (82.6%) were tested . Chi-squared testing demonstrated clinical significance when comparing the screening prior to and subsequent to the introduction of the clinic, p value = 9.7x 10-7 (Chi=23.9956). 6 of the screened cases demonstrated molecular features with MSI and abnormal Immunohistochemistry, and are undergoing further germline genetic testing.

Conclusion: The establishment of a dedicated ‘Family History of Bowel Cancer Service’ resulted in a significant improvement in the screening for Lynch Syndrome in accordance with the Revised Bethesda Criteria, 2004. We would recommend that this service should be extended throughout the United Kingdom to help aid early diagnoses and improve long-term outcomes.

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