University Hospitals Bristol NHS Foundation Trust

Bristol Regional Genetics Service Referral Guidelines
Colorectal Cancer Family History

These guidelines have been designed to direct appropriate referrals for Colorectal Cancer (CRC) family history to Clinical Genetics and/or Gastroenterology and are based on published guidelines (Cairns et al. Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups (update from 2002). GUT 2010;59:666-690). These guidelines only apply to asymptomatic individuals.

Clinical Genetics referral: For discussion about future surveillance recommendations and/or possible genetic investigations/predictive genetic testing.

Gastroenterology referral: For discussion about current surveillance recommendations. They can then refer to Clinical Genetics if indicated.

**Amsterdam Criteria: (used to identify HNPCC/Lynch syndrome families)**
- Associated cancers: Colorectal (CRC), endometrial, small bowel, ureter
- Three or more affected relatives in 1° kinship (At least one must be a 1° relative of your patient)
- Two or more generations affected
- One or more diagnosis <50yrs of age
- Familial Adenomatous Polyposis (FAP) excluded

**START:** Known cancer syndrome/gene mutation in family?

- Yes
  - Refer to Clinical Genetics
  - Refer to Gastroenterology for surveillance and Clinical Genetics

- No
  - Fulfil Amsterdam Criteria? (in box above)
    - Yes = Potentially High Risk
      - Age 25 years or over?
        - Yes
          - Potentially High Risk
            - One relative = First degree relative i.e. parents, child, brothers, sisters
            - Two relative = Second degree relative i.e. grandparents, grandchildren, aunts/uncles, nieces/nephews
            - 1° kinship = First degree relatives of each other

    - No
      - Potentially Moderate or Population risk
        - Two relatives with CRC at any age (1° kinship to patient and each other incl. both parents) ?
          - Yes
            - Age 50 years or over?
              - No
                - Refer to Gastroenterology for surveillance
                - Refer to Clinical Genetics
              - Yes
                - Refer to Clinical Genetics
          - No
            - Refer to Gastroenterology for surveillance
            - Refer to Clinical Genetics

If your patient’s family history does not fulfil these criteria, but there are other concerns e.g. a family history of polyps, then please contact the department.

For queries for the on-duty Genetic Counsellor St Michael’s Hospital in Bristol tel: 0117 342 5115

Please address Clinical Genetics referrals to Consultant Geneticist:
Dr Alan Donaldson (for Bath and Gloucestershire)
Dr Susan Tomkins (for Bristol and Taunton)

**Other cancer family history:** A referral to Clinical Genetics may be worth considering if your patient has a family history of:
- Multiple family members with tumours at the same site
- Unusually young ages of onset of tumours
- A history of an individual with multiple primary tumours
- Recognised associations: colon/womb; breast/ovarian
- Other tumours types that may indicate a genetic syndrome: Gastric cancer, sarcomas, neuroendocrine tumours

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