Diagnostic criteria for Lynch syndrome (Hereditary non-polyposis colorectal cancer, HNPCC)

▪ **Revised Bethesda Diagnostic criteria**
(Umar A. et al., J Natl Cancer Inst. 2004;96(4):261-8)

- CRC diagnosed at younger than 50 years,
- Presence of synchronous or metachronous CRC or other Lynch syndrome-associated tumors[1],
- CRC with MSI-high pathologic-associated features (Crohn-like lymphocytic reaction, mucinous/signet cell differentiation, or medullary growth pattern) diagnosed in an individual younger than 60 years old,
- Patient with CRC and CRC or Lynch syndrome-associated tumor[1] diagnosed in at least one first-degree relative younger than 50 years old,
- Patient with CRC and CRC or Lynch syndrome-associated tumor[1] at any age in two first-degree or second-degree relatives.

CRC, colorectal cancer
MSI, microsatellite instability
[1] Lynch syndrome-associated tumors include tumor of the colorectum, endometrium, stomach, ovary, pancreas, ureter, renal pelvis, biliary tract, brain, small bowel, sebaceous glands, and kerotoacanthomas.

Note
Additional useful reference discussing the use of the above diagnostic criteria:
Giardiello FM et al., US Multi-Society Task Force on Colorectal Cancer. Guidelines on genetic evaluation and management of Lynch syndrome: a consensus statement by the US Multi-Society Task Force on colorectal cancer. Gastroenterology 2014;147(2):502-26.
Accessible at http://www.gastrojournal.org/article/S0016-5085%2814%2900448-X/fulltext

▪ Amsterdam I and Amsterdam II criteria

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Table 1. Amsterdam and Amsterdam II Criteria for the Clinical Diagnosis of HNPCC

Amsterdam Criteria [1]	Amsterdam II Criteria [2]
• **Three** or more family members, one of whom is a first-degree [3] relative of the other two, with a confirmed diagnosis of colorectal cancer • **Two** successive affected generations • **One** or more colon cancers diagnosed before age 50 years Exclusion of familial adenomatous polyposis (FAP)	• **Three** or more family members (one of whom is a first-degree relative [3] of the other two) with HNPCC-related cancers [4] • **Two** successive affected generations • **One** or more of the HNPCC-related cancers diagnosed before age 50 years Exclusion of familial adenomatous polyposis (FAP)

1. Vasen et al [1991]

2. Vasen et al [1999]

3. Parent, child, or sibling

4. Colorectal, endometrial, stomach, small intestinal, hepatobiliary, renal pelvic, or ureteral

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Pagon RA, Adam MP, Ardinger HH, et al., editors.
Seattle (WA): University of Washington, Seattle; 1993-2014.

1. Amsterdam Criteria: Vasen HF, *et al*. The International Collaborative Group on hereditary non-polyposis colorectal cancer (ICG-HNPCC). Dis Colon Rectum. 1991;34:424–5.
2. Amsterdam II Criteria: Vasen HF, *et al.* New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch syndrome) proposed by the International Collaborative group on HNPCC. Gastroenterology. 1999;116:1453–6.