

COLARIS[®] - A genetic test for hereditary nonpolyposis colorectal cancer (HNPCC)

SERVICE SPECIFICATION SHEET

If you have any queries regarding the test specifications outlined below please contact Lab21 Customer Service to discuss prior to sending samples for analysis.

Intended Use: To provide a service to test samples from individuals for risk of hereditary colorectal and endometrial cancer.

Technical Information: The sequencing of patient DNA is carried out following polymerase chain reaction and other molecular biological techniques.

Comprehensive COLARIS[®]

Full DNA sequence analysis for mutations in the MLH1 and MSH2 genes.

Single site COLARIS[®]

DNA sequence analysis for a specified mutation in the MLH1 and/or MSH2 genes.

MSH6

DNA sequence analysis of the MSH6 gene.

All mutations and genetic variants are named according to the convention of Beaudet and Tsui. (Beaudet AL, Tsui LC. A suggested nomenclature for designating mutations. *Hum Mut* 1993; **2**:245-248).

This test is not required to be CE marked under the European IVD directive 98/79/EC. (MHRA notification August 2007). The test is performed in a CAP & CLIA approved laboratory.

Testing Time: Turnaround: report issued in 3 weeks from receipt of sample. Twelve working days for Rapid analysis.

Specimen Requirements: Lab21 will provide suitable testing kits and packaging which should be returned for analysis to Lab21 Ltd.

Specimen Handling: See 'Advice for Healthcare Providers – Myriad Genetic Tests'.

Reporting of Results: Test results will be sent to the ordering physician to be discussed with the patient.

- References:**
1. Aarnio M *et al.* Cancer risk in mutation carriers of DNA-mismatch-repair genes. *Int J Cancer*. 1999; **81**:214-218.
 2. Jarvinen HJ *et al.* Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*. 2000; **118**:829-34.
 3. Vasen HFA *et al.* Cancer risk in families with hereditary nonpolyposis colorectal cancer diagnosed by mutation analysis. *Gastroenterology* 1996; **110**:1020-1027.
 4. Burke W *et al.* Recommendations for follow-up care of individuals with an inherited predisposition to cancer. I. Hereditary nonpolyposis colon cancer. Cancer Genetics Studies Consortium. *JAMA*. 1997; **277**:915-9.

COLARIS [®] Service Specification Sheet		Page 1 of 1
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