

COLARIS AP[®] - A genetic test for adenomatous polyposis syndromes

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 polyps and cancer.

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

Comprehensive COLARIS AP[®]

Full DNA sequence analysis for mutations in the APC and MYH genes.

Single site COLARIS[®]

DNA sequence analysis for a specified mutation in the APC 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.

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. Burt RW *et al.* Genetic testing and phenotype in a large kindred with attenuated familial polyposis. *Gastroenterology*. 2004; **127**:444-451
 2. Giardiello FM *et al* technical review on hereditary colorectal cancer and genetic testing. *Gastroenterology*. 2001; **121**:198-213.
 3. Sieber OM *et al.* Multiple colorectal adenomas, classic adenomatous polyposis, and germ-line mutations in MYH. *N Engl J Med*. 2003; **348**:791-799.
 4. Wang L *et al.* MYH mutations in patients with attenuated and classical polyposis and young onset colorectal cancer without polyps. *Gastroenterology*. 2004; **127**:9-16