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| **COLORECTAL CANCER GENE PANEL TESTING PRO FORMA** | | | | | | | |
| **Patient details** *(printed label preferred)* | | | | | | | |
| ***Forename(s):*** | | |  | | ***Sex:*** | |  |
| ***Surname:*** | | |  | | ***Patient ID:*** | |  |
| ***DOB:*** | | |  | | ***Pedigree no.*** | |  |
| ***CHI:*** | | |  | | ***Referrer:*** | |  |
| **Clinical Summary:** | | | | | | | |
| ***Clinical information***  ***(type of cancer, age of onset, family history)*** | | | | | ***Tumour information*** | | |
|  | MSI high | |
|  | MMR loss of staining (please detail): | |
|  | IHC uninformative | |
|  | *BRAF* variant detected | |
|  | *MLH1* promoter hypermethylation | |
|  | Tumour unavailable | |
| **Testing requested** (please see test directory for specific referral criteria) | | | | | | | |
| **Sequence analysis** **of the colorectal gene cancer panel is undertaken for ALL patients (see genes below):**  *APC*, *BMPR1A*, *MBD4*ǂ, *MLH1*, *MSH2*, *MSH3*ǂ, *MSH6*, *MUTYH*, *NTHL1*, *PMS2*§, *POLD1* (exons 4-12), *POLE* (exons 3-13), *PTEN*, *RNF43*, *SMAD4*, *STK11*  ǂbiallelic truncating variants only  §*PMS2* analysis includes 1-10; exons 11-15 cannot be reliably investigated by this method due to the presence of the *PMS2CL* pseudogene. These exons are analysed using long range PCR when PMS2 testing is indicated i.e., isolated loss of PMS2 staining in tumour tissue.  **Dosage analysis** **is conducted according to clinical indication as outlined below.**  **Please tick (or double click)** to select | | | | | | | |
| **Select** | **Clinical Indication** | | | **Additional test details including dosage analysis (MLPA)** | | | |
|  | **Polyposis**  Desmoid tumour \*  CHRPEs \*\* | | | *APC*, *MUTYH* (selected exons), and *GREM1* (upstream region)  \* only *APC* and *MUTYH* variants reported  \*\* only *APC* variants reported | | | |
|  | **Lynch Syndrome**  *(see overleaf for guidelines)* | | | *MLH1*, *MSH2*, *MSH6* and *EPCAM* (selected exons)  *PMS2* only if indicated i.e., isolated loss of PMS2 staining in tumour tissue | | | |
|  | **Colorectal Cancer**  patient dx <45yrs | | | **None as standard**  Lynch dosage (see above) will be undertaken for patients  dx <45yrs | | | |
|  | **Peutz-Jeghers Syndrome** | | | *STK11* | | | |
|  | **Juvenile Polyposis Syndrome** | | | *SMAD4* and *BMPR1A* | | | |
|  | **Hereditary Diffuse Gastric Cancer** | | | Patients will ONLY be analysed for *CDH1* gene (sequencing and dosage analysis) | | | |
| *Additional sequencing analysis for the following gene panels can be included if clinically indicated:* | | | | | | | |
| **Breast** | | *BRCA1¤*, *BRCA2¤*, *PALB2*, *PTEN*, *STK11*, *TP53*, *ATM*#, *CHEK2*#, *RAD51C, RAD51D*  #Reporting truncating variants only and the ATM variant c.7271T>G p.(Val2424Gly). Please note CHEK2 analysis is restricted to exons 1 to 9 plus the common c.1100delC variant due to the presence of a pseudogene | | | | | |
| **Ovarian** | | *BRCA1¤*, *BRCA2¤*, *BRIP1*, *MLH1*, *MSH2*, *MSH6*, *RAD51C*, *RAD51D*, *PALB2* | | | | | |
| *¤* Dosage analysis will be conducted separately by the Aberdeen laboratory | | | | | | | |

