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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  |

