FAMILIAL CANCER PREDISPOSITION REQUEST FORM

Peter Mac
Peter MacCallum Cancer Centre
Victoria Australia

NATA & RCPA Accredited Laboratory
Number 2465

RCPA The Royal College of Pathologists of Australasia

NATA

PATIENT DETAILS – Place Bradma label below

Surname: ..... ☐ Male ☐ Female

First name: ..... DOB: ..... / ..... / .....

Address: .....

.....

Medicare Number: ☐☐☐☐☐ ☐☐☐☐☐ ☐ (☐)

REQUESTING CLINICIAN DETAILS

Doctor name: .....

Address: .....

Provider No: .....

Email*: .....

Phone: .....

Signature: ..... Date ..... / ..... / .....

SPECIMEN DETAILS

☐ Blood ☐ OTHER (please state): .....

☐ STORED SAMPLE Please provide lab ID.: .....

REPORT COPY TO CLINICIAN/HEALTHCARE PROVIDER

Doctor name: .....

Address: .....

Email*: .....

Phone: .....

CLINICAL NOTES

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SELECT TEST/S

See reverse for gene lists, test information, prices & specimen collection guidance

GENE PANELS

☐ SPECTRUM ONE*	☐ COLO & ENDO*	☐ MELANOMA
☐ BRCA ONLY	☐ MMR	☐ GORLIN SYNDROME
☐ BRCA PLUS	☐ POLYPS	☐ FACIAL PAPULES
☐ BOPP	☐ ENDOCRINE	☐ SCHWANNOMATOSIS
☐ OVARIAN*	☐ RENAL	☐ S ARCOMA
☐ PROSTATE*	☐ PARA, PHEO & GIST	☐ ASHKENAZI
☐ PANCREAS*	☐ PITUITARY	☐ HAEM

State additional "add on" genes (up to 5): .....

State any gene/s to be removed from selected panel(s): .....

* ☐ Add-on PMS2 exon11-15 long range testing (IHC result must be provided)

TARGETED VARIANT TESTING

Gene and Variant details: .....

A Peter Mac lab ID or external report must be attached for this testing.

PMCC Lab ID: ..... OR report emailed ☐

☐ PREDICTIVE TESTING (Known Familial LP / Pathogenic Variant)

☐ CONFIRMATORY TESTING (Known Familial LP / Pathogenic Variant)

☐ SEGREGATION TESTING (Known Familial VUS)

☐ GERMLINE VS SOMATIC ORIGIN OF VARIANT DETECTED IN TUMOUR

☐ CONFIRMATION OF RESEARCH / EXTERNAL RESULT

☐ APC Promoter 1B deletion (MLPA)

SINGLE GENE(S)

Please state single gene(s) for screening .....

.....

RE-ANALYSIS

Please state gene(s) or panel(s) for re-analysis: .....

Previous PMCC Lab ID: .....

SPECIMEN COLLECTION DECLARATION

To be completed by collector (if primary form)
I certify that the pathology specimen and request form comply with minimum labelling requirements and that the specimen was taken from the patient stated above as established by direct enquiry and/or inspection of the identification band and was labelled immediately.

Signature:

Collected by: ..... Date: ..... / ..... / ..... Time: .....

SELECT PAYMENT OPTION & PATIENT CONSENT

☐ **Medicare -** MUST BE ELIGIBLE, MUST SIGN BELOW.
PLEASE PROVIDE MEDICARE ITEM NUMBER (see reverse for guidance): .....

☐ **Other** - Please specify: .....

☐ **Hospital/Pathology Provider** (please specify): .....

☐ **Bill Patient** (must sign to acknowledge cost)
Please complete credit card form or contact Peter Mac Billing team on 8559 6655

To be completed by patient
I provide consent to have genomic testing. I have had the opportunity to discuss genomic testing and its implications with a health professional. Been given access to information about this testing. Been able to ask questions until I am satisfied with the answers. I authorise the approved pathology practitioner who will render the requested pathology services, and any further pathology services which the practitioner determines to be necessary, to submit my unpaid account to Medicare, so that Medicare can assess my claim and issue me a cheque made payable to the practitioner, for the Medicare benefit. Verbal consent was provided by patient to submit unpaid account to Medicare (no signature available). Authority for Peter Mac to submit claim on behalf of claimant. Your doctor has requested testing from Peter MacCallum Pathology. You are free to choose your own pathology provider however, if your doctor has specified a particular pathologist on clinical grounds, a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor. **Privacy Note:** The information provided will be used to assess any Medicare benefit payable for the services rendered and to facilitate the proper administration of government health programs and may be used to update enrolment records. Its collection is authorised by provision of the Health Insurance Act 1973. The information may be disclosed to the Department of Health and Ageing or to a person in the medical practice associated with this claim, or as authorised/required by law.

Patient's Signature:

Date ..... / ..... / .....

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GENE LISTS, TEST INFORMATION & PRICES

- All Cancer Predisposition testing is performed using Next Generation Sequencing. Screening analysis includes copy number variant (CNV) detection for GREM1 (SCG5) and EPCAM, single nucleotide variant (SNV) detection in exonuclease domains of POLE and POLD1 and **both SNV and CNV detection for all other genes.**.
- All tests listed on this form are for germline testing, refer to the Specimen Requirements section below for a list of accepted specimen types. Assays for tumour samples are available please see https://www.petermac.org/health-professionals/services-for-health-professionals/pathology-health-professionals/molecular-pathology.
- If requesting Cancer Predisposition testing on a patient with active haematological malignancy please consider sending a non-haematological specimen for testing (hair preferred). Bone marrow is not an accepted sample type for this panel. Please provide relevant pathology reports.
- PMS2 testing note: When evidence of loss of expression of MLH1/PMS2 by immunohistochemistry (IHC) is provided (IHC report) exons 11-15 long-range PCR & Sanger sequencing is performed in addition to NGS. Otherwise, all exons of the PMS2 gene are tested by NGS and if a variant is detected, it is confirmed by long-range PCR and Sanger sequencing prior to reporting.

GENE PANELS

SPECTRUM ONE (28 genes): APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, GREM1 (SCG5), MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53	**RENAL** (15 genes): BAP1, FH, FLCN, MET, MLH1, MSH2, MSH6, PTEN, SDHA, SDHB, SDHC, SDHD, TSC1, TSC2, VHL
BRCA ONLY (2 genes): BRCA1, BRCA2	**PARA, PHEO & GIST** (paraganglioma, pheochromocytoma & GIST) (18 genes): EPAS1, FH, IDH1, KIF1B, KIT, MAX, MDH2, NF1, NF2, PDGFRA, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
BRCA PLUS (10 genes): ATM, BARD1, BRCA1, BRCA2, BRIP1, CHEK2, PALB2, RAD51C, RAD51D, TP53	**PITUITARY** (8 genes): AIP, CDKN1B, MEN1, PRKAR1A, SDHA, SDHB, SDHC, SDHD
BOPP (Breast, Ovarian, Prostate & Pancreas) (20 genes): ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53	**MELANOMA** (8 genes): BAP1, BRCA2, CDK4, CDKN2A, POT1, PTEN, RB1, TP53
OVARIAN (12 genes): BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, TP53	**GORLIN SYNDROME** (2 genes): PTCH1, SUFU
PROSTATE (11 genes): ATM, BRCA1, BRCA2, CHEK2, HOXB13, EPCAM, MLH1, MSH2, MSH6, PMS2, TP53	**FACIAL PAPULE SYNDROME** (5 genes): FH, FLCN, PTEN, TSC1, TSC2
PANCREAS (12 genes): ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53	**SCHWANNOMATOSIS** (3 genes): LZTR1, NF2, SMARCB1
COLORECTAL & ENDOMETRIAL (20 genes): APC, AXIN2, BMPR1A, BUB1B, CDH1, EPCAM, GREM1 (SCG5), MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53	**SARCOMA** (5 genes): APC, EXT1, EXT2, RB1, TP53
MMR (Mismatch Repair) (7 genes): EPCAM, MLH1, MSH2, MSH6, PMS2, POLD1, POLE	**HAEM MALIGNANCY PREDISPOSITION** (10 genes): ANKRD26, CEBPA, DDX41, ETV6, GATA2, MBD4, RUNX1, SAMD9, SAMD9L, TP53
POLYPS (13 genes): APC, AXIN2, BMPR1A, BUB1B, GREM1 (SCG5), MUTYH, NTHL1, POLD1, POLE, PTEN, SMAD4, STK11, TP53	**ENDOCRINE** (8 genes): AIP, CDC73, CDKN1B, MEN1, PRKAR1A, PTEN, RET, VHL

Spectrum One = $550 **All other Cancer Predisposition panels = $450** **Add on genes = $50 per gene** (up to 5 additional genes may be added to a panel)

SINGLE GENE(S)

• Any of the genes listed above as well as the following genes are available to request as standalone gene screens: CSDE1, DICER1, EGLN1, EGLN2, MLH3, MSH3, RNF43, SMARCA4, SMARCE1

Single gene = $350 **Each additional gene = $50 per gene (up to 6 genes may be requested)**

TARGETED VARIANT TESTING

• Any of the genes listed are available to request for targeted testing of a known variant(s) – either familial (predictive, confirmatory or segregation testing) or based on prior testing (confirmation of research/external finding or germline testing of variant detected in tumour sample). A Peter Mac Lab ID or external report containing the variant details must be provided for this testing.

• Testing of two independent sample collections is recommended. Please submit a request form for each sample.

Targeted variant testing = $250

RE-ANALYSIS

• Any of the genes/panels listed above are available to request for re-analysis of a sample previously tested. Gene/panel availability is subject to the methodology used at the time of testing.

Re-analysis is provided free of charge for up to 4 genes **More than 4 genes = $150 (flat cost regardless of number of genes)**

MEDICARE ITEM NUMBER REFERENCE TABLE

See below applicable Medicare item numbers available for various categories of testing. If selecting Medicare in the Payment section on page 1 of this form, please provide the relevant item number using the information below to avoid billing errors and unnecessary charges to the patient. Further details: http://www.mbsonline.gov.au

73295	HG serous or HG epithelial ovarian, fallopian tube or primary peritoneal ca. Panel must include BRCA1 / BRCA2	Gene Panels / Singles Gene(s)
73296	Breast, ovarian, fallopian tube or primary peritoneal ca at >10% risk of germline variant. Panel must include BRCA1, BRCA2 and STK11 / PTEN / CDH1 / PALB2 / TP53	Gene Panels / Singles Gene(s)
73297	BRCA1, BRCA2, STK11, PTEN, CDH1, PALB2, TP53 predictive testing (or in genes genes associated with breast, ovarian, fallopian tube or primary peritoneal cancer)	Targeted Variant Testing
73302	BRCA1 or BRCA2 variant detected in tumour, for germline vs somatic origin testing	Targeted Variant Testing
73304	Metastatic castration-resistant prostate ca. Panel must include BRCA1/ BRCA2	Gene Panels / Singles Gene(s)
73333	Clinical diagnosis of Von Hippel-Lindau. Panel must include VHL	Gene Panels / Singles Gene(s)
73334	VHL predictive testing	Targeted Variant Testing
73339	Clinical diagnosis of Multiple Endocrine Neoplasia type 2. Panel must include RET	Gene Panels / Singles Gene(s)
73340	RET predictive testing	Targeted Variant Testing
73354	Suspected Lynch syndrome. Panel must include MLH1 / MSH2 / MSH6 / PMS2 / EPCAM	Gene Panels / Singles Gene(s)
73355	Adenomatous polyposis at >10% risk of germline variant. Panel must include APC, MUTYH	Gene Panels / Singles Gene(s)
73356	Non-adenomatous polyposis at >10% risk of germline variant. Panel must include SMAD4 / BMPR1A / STK11 / GREM1	Gene Panels / Singles Gene(s)
73357	APC, MUTYH, MLH1, MSH2, MSH6, PMS2, EPCAM, SMAD4, BMPR1A, STK11, GREM1 predictive testing	Targeted Variant Testing

SPECIMEN REQUIREMENTS

ACCEPTED TYPES:	REQUIREMENTS:
BLOOD	4ml peripheral blood (EDTA)
HAIR FOLLICLES	>20 hair follicles (eyebrow hair preferred) in sterile container
SALIVA	Must use a commercial kit
TISSUE (FRESH)	Send in sterile container, with saline preferred
GENOMIC DNA	≥10µl at >50ng/µl extracted from any of the above sample types
FFPE TISSUE	Please contact laboratory, only accepted in certain settings. NOTE: Normal (non-tumour) tissue required

ADDRESS & CONTACT DETAILS

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