Ordering Restrictions: Carrier and predictive testing is currently restricted to Maritime Medical Genetics (MMGS) with some exceptions. Testing for Hereditary Cancer Syndromes and Cardiomyopathies can only be ordered by MMGS. Symptomatic testing might be restricted/allowed based on the test and the ordering provider's clinical specialty.

For any questions, please consult the lab genetic counsellor at clinicalgenomics.gc@iwk.nshealth.ca

For any test restricted to Maritime Medical Genetics Services (MMGS), please refer your patient to their department or contact them at 902.470.8754.

Indication for Testing	Test Name	Ordering Restrictions/Notes
BANKING – LONG TERM DNA STORAGE (25 YEARS)		
Irreplaceable samples that require long-term storage	Bank – DNA Long-term Banking	**Please check banking storage policy for appropriate clinical indications**
CARDIOLOGY		
Arrhythmias	Arrhythmia Panel Genes: *ANK2-CACNA1C-CACNB2-CALM1-CALM2-CALM3-CASQ2-CAV3-CDH2-DSC2-DSG2-DSP-GPD1L-HCN4-JUP-KCNE1-KCNE2-KCNE3-KCNH2-KCNJ2-KCNQ1-PKP2-RYR2-SCN1B-SCN3B-SCN5A-TECRL-TMEM43-TRDN*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	Arrhythmogenic Right Ventricular Cardiomyopathy Panel Genes: *CDH2-DSC2-DSG2-DSP-JUP-PKP2-RYR2-TMEM43*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Brugada Syndrome	Brugada Panel Genes: *SCN5A*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Cardiomyopathies	Cardiomyopathy Panel Genes: *ABCC9-ACTC1-ACTN2-ANKRD1-BAG3-CAV3-CDH2-CSRP3-DES-DMD-DSC2-DSG2-DSP-EMD-FLNC-GLA-JUP-LAMP2-*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service

	LDB3-LMNA-MYBPC3-MYH6-MYH7-MYL2-MYL3-PKP2-PLN-PRKAG2-RAF1-RBM20-RYR2-SCN5A-SGCD-TAFAZZIN-TCAP-TMEM43-TMPO-TNNC1-TNNI3-TNNT2-TPM1-TTN-TTR-VCL	
Catecholaminergic Polymorphic Ventricular tachycardia (CPVT)	Catecholaminergic Polymorphic Ventricular Tachycardia Panel Genes: *CALM1-CASQ2-RYR2-TECRL-TRDN*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Dilated Cardiomyopathy (DCM)	Dilated Cardiomyopathy Panel Genes: *ABCC9-ANKRD1-ACTC1-ACTN2-BAG3-CSRP3-DES-DMD-DSG2-EMD-LAMP2-LDB3-LMNA-MYBPC3-MYH6-MYH7-PLN-RAF1-RBM20-SCN5A-SGCD-TAFAZZIN-TCAP-TNNC1-TNNI3-TNNT2-TPM1-TMPO-TTN-VCL*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Familial Hypercholesterolemia (FH)	Familial Hypercholesterolemia Panel Genes: *APOB-LDLR-LDLRAP1-PCSK9*	Diagnosis: Cardiology, Endocrinology, GENETICS, Nova Scotia Medical Examiners' Service
Holt-Oram Syndrome (HOS)	*TBX5* Single Gene Testing	GENETICS Only
Hypertrophic Cardiomyopathy (HCM)	Hypertrophic Cardiomyopathy Panel Genes: *ACTC1-ACTN2-CAV3-CSRP3-FLNC-GLA-LAMP2-MYBPC3-MYH7-MYL2-MYL3-PLN-PRKAG2-TNNC1-TNNI3-TNNT2-TPM1-TTR*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Left Ventricular Non-Compaction Cardiomyopathy (LVNC)	Left Ventricular Non-Compaction Cardiomyopathy Panel Genes: *ACTC1-LDB3-LMNA-MYBPC3-MYH7-TAFAZZIN-TNNI3-TNNT2-TPM1-TTN*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Long QT Syndrome (LQTS)	Long QT Panel Genes: *ANK2-CALM1-CALM2-CALM3-CACNA1C-CAV3-KCNE1-KCNE2-KCNH2-KCNJ2-KCNQ1-SCN5A*	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service

Transthyretin Amyloidosis	*TTR* Single Gene Testing	Restrictions: None
CONNECTIVE TISSUE		
Aortopathy	Aortopathy Panel Genes: *ACTA2-BGN-CBS-COL1A1 (c.934C>T only)-COL3A1-EFEMP2-FBN1-FBN2-FOXE3-HCN4-LOX-MAT2A-MFAP5-MYH11-MYLK-PRKG1-SKI-SLC2A10-SMAD2-SMAD3-TGFB2-TGFB3-TGFBR1-TGFBR2-YY1AP1* (c.1079C>T only)	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Ehlers-Danlos Syndrome (EDS)	Ehlers-Danlos Syndrome (EDS) Panel Genes: *ADAMTS2-AEBP1-ATP7A-B3GALT6-B4GALT7-C1R-C1S-CHST14-COL1A1-COL1A2-COL3A1-COL5A1-COL5A2-COL12A1-DSE-FKBP14-FLNA-PLOD1-PRDM5-SLC39A13-ZNF469-TNXB*	GENETICS Only
Hereditary Connective Tissue Disorders	Hereditary Disorder of Connective Tissue Panel Genes: *ACTA2-ADAMTS2-AEBP1-ATP7A-B3GALT6-B4GALT7-BGN-C1R-C1S-CBS-CHST14-COL12A1-COL1A1-COL1A2-COL3A1-COL5A1- COL5A2-DSE-FBN1-FBN2-FKBP14-FLNA-PLOD1-PRDM5-PRKG1* (c.530G>A;p.R177Q only)*-SKI-SLC2A10-SLC39A13-SMAD2-SMAD3-TGFB2-TGFB3-TGFBR1-TGFBR2-TNXB-YY1AP1* (c.1079C>T only) *and ZNF469*	GENETICS Only
Hereditary Hemorrhagic Telangiectasia (HHT)	Hereditary Hemorrhagic Telangiectasia (HHT) Panel Genes: *ACVRL1-ENG-RASA1-SMAD4*	GENETICS Only
Loeys-Dietz Syndrome	Loeys-Dietz Syndrome Panel Genes: *SMAD2-SMAD3-TGFB2-TGFB3-TGFBR1-TGFBR2*	GENETICS Only
Marfan Syndrome	*FBN1* Single Gene Testing	GENETICS Only

DERMATOLOGY		
Cutis Laxa	Cutis Laxa Panel Genes: *ELN-ATP6V0A2-EFEMP2-FBLN5-LTBP4-PYCR1*	GENETICS Only
Pseudoxanthoma Elasticum (PXE)	*ABCC6* and *GGCX* Gene Sequencing and Del/Dup	Diagnosis: Dermatology, GENETICS, Ophthalmology
X-Linked Ichthyosis (Steroid Sulfatase Deficiency)	X-Linked Ichthyosis via MLPA	Restrictions: None
ENDOCRINOLOGY		
Familial Hypocalciuric Hypercalcemia (FHH)	Familial Hypocalciuric Hypercalcemia (FHH) Panel Genes: *AP2S1-GNA11-CASR*	Diagnosis: Endocrinology, GENETICS
Familial Hyperparathyroidism	Hyperparathyroidism Panel Genes: *CASR-CDC73-CDKN1B-MEN1-RET*	Diagnosis: Endocrinology, GENETICS
Hypophosphatasia	*ALPL* Single Gene Testing	Diagnosis: Endocrinology, GENETICS
Mature onset diabetes of the young (MODY)	Maturity-Onset Diabetes of the Young Panel Genes: *ABCC8-GCK-HNF1A-HNF1B-HNF4A-INS-KCNJ11-PDX1*	Diagnosis: Endocrinology, GENETICS
EAR, NOSE AND THROAT (ENT)		
Non-Syndromic Hearing Loss	*GJB2* + *GJB6* Gene Sequencing and Del/Dup	Restrictions: None
Usher Syndrome Type II	*USH2A* Single Gene Testing Only	GENETICS Only **Please note: *USH2A* analysis only accounts for 57-79% of USH2A**
GENERAL DISORDERS		

22q11.2 Deletion and Duplication syndrome	22q11.2 Deletion/Duplication MLPA	Restrictions: None
Adrenoleukodystrophy	*ABCD1* Single Gene Test	GENETICS Only
Alström Syndrome	*ALMS1* Single Gene Test	GENETICS Only
Angelman Syndrome	Angelman Syndrome MLPA	Restrictions: None
Beckwith-Wiedemann Syndrome	Beckwith-Wiedemann Syndrome MLPA/NGS	Restrictions: None **Testing includes MS-MLPA & reflex to NGS if MLPA negative**
Carney Complex	*PRKAR1A* Single Gene Test	GENETICS Only
Cystic Fibrosis	*CFTR* Single Gene Test	Restrictions: Depends on request (carrier/diagnostic/etc).
Developmental Delay, Autism Spectrum Disorder, Multiple Congenital Anomalies	Microarray (Illumina CytoSNP-850K)	Postnatal: None Prenatal: GENETICS only unless POC **Must meet criteria for testing**
Down Syndrome (postnatal)	Down Syndrome via QF-PCR	Restrictions: None
Fragile X	*FMR1* Analysis (Triplet-primed PCR)	Diagnosis: None (but must meet criteria – see below) Patient with GDD/ID and/or ASD AND other features such as macroorchidism, macrocephaly, facial features and/or connective tissue findings OR Patient with GDD, ID and/or ASD with a maternal family history that includes: GDD/ID/ASD, females with premature menopause or ovarian insufficiency, and

		males or females with adult-onset tremor, ataxia or parkinsonism
Fragile X Tremor-Associated Syndrome/Premature Ovarian Failure	*FMR1* Analysis (Triplet-primed PCR)	Diagnosis: None
Hereditary Multiple Osteochondromas	Hereditary Multiple Osteochondromas Panel Genes: *EXT1-EXT2*	GENETICS Only
Klinefelter Syndrome	Klinefelter Syndrome via QF-PCR	Restrictions: None
L1 Syndrome	*L1CAM* Single Gene Test	GENETICS Only
Male Infertility – Y Microdeletion	Y-chromosome AZF Microdeletion via fPCR	Restrictions: None
Malignant Hyperthermia Susceptibility Disorder	Malignant Hyperthermia Susceptibility Disorder Panel Gene: *CACNA1S-RYR1-STAC3*	DMF-Malignant Hyperthermia clinic at Maritime Medical Genetics Service
Maternal DNA Contamination (MCC studies)	QF-PCR	Restrictions: None
Neurofibromatosis Type 1	*NF1* and *SPRED1* Gene Sequencing and Del/Dup	Diagnosis: Dermatology, GENETICS, Neurology, Pediatrics
Neurofibromatosis Type 2	*NF2* Single Gene Test	GENETICS Only
Noonan Syndrome	Noonan Syndrome Panel Genes: *BRAF-CBL-HRAS-KRAS-LZTR1-MAPK1-MAP2K1-MAP2K2-NRAS-PTPN11-RAF1-RASA2-RIT1-RRAS-SHOC2* (c.4A>G only)-*SOS1-SOS2* (DH Domain: c.592_1164 only)	GENETICS Only
Prader Willi Syndrome (PWS)	15q11 Methylation and Dosage MLPA	Restrictions: None
PTEN-related Macrocephaly/Autism Spectrum Disorder	*PTEN* Single Gene Test	GENETICS Only

Rett Syndrome	*MECP2* Single Gene Test	Diagnosis: GENETICS, Neurology, Pediatrics
Russell-Silver Syndrome (RSS)	Russell-Silver Syndrome Methylation and Dosage MLPA	Restrictions: None
Sotos Syndrome	*NSD1* Single Gene Test	GENETICS Only **15-50% of probands detected via array (order array in conjunction if applicable)**
Tay-Sachs Disease	*HEXA* Single Gene Test	Diagnosis: GENETICS, Neurology** **Requires HEX A enzyme activity prior to testing**
Turner Syndrome	Turner Syndrome via QF-PCR	Restrictions: None
Wilson Disease	*ATP7B* Single Gene Test	Diagnosis: GI*, GENETICS, Neurology* **Check clinical indication with Lab GC/Lab Scientist**
X-Inactivation	X-Inactivation Studies via fPCR	GENETICS Only **Requires discussion with Lab GC/Lab Scientist before initiation**
HEMATOLOGY		
Atypical Hemolytic Uremic Syndrome (aHUS)	Hemolytic Uremic Syndrome Panel Genes: *ADAMTS13-C3-CD46-CFB-CFH-CFHR5-CFI-DGKE-THBD*	Diagnosis: Hematology, GENETICS, Nephrology
Hereditary Hemochromatosis & Related Disorders	Hereditary Hemochromatosis & Related Disorders Panel Genes: *HFE-HAMP-HJV-SLC40A1-TFR2-CP-BMP6*	Diagnosis: GENETICS, Hematology, Hepatology ***HFE* common variants should be completed before ordering panel**
Shwachman-Diamond Syndrome	*SBDS* Single Gene Testing	Diagnosis: Hematology, GENETICS
Thrombocytopenia	Thrombocytopenia Panel	Diagnosis: Hematology, GENETICS

	Genes: *ANKRD26-ETV6-FLNA-GBA-RUNX1-THBD*	
IMMUNOLOGY/RHEUMATOLOGY		
Autoinflammatory Disease	Autoinflammatory Disease Panel Genes: *ACP5-ADAR-CARD14-DDX58-ELANE-IFIH1-IL1RN-IL36RN-ISG15-LPIN2-MEFV-MVK-NLRC4-NLRP1-NLRP12-NLRP3-NOD2-OTULIN-PLCG2-PRG4-PSENEN-PSMB8-PSTPIP1-RNASEH2A-RNASEH2B-RNASEH2C-SAMHD1-SLC29A3-STING1-TNFAIP3-TNFRSF1A-TREX1-TRNT1*	Diagnosis: GENETICS, Immunology, Rheumatology
Familial Mediterranean Fever	*MEFV* Single Gene Testing	Diagnosis: GENETICS, Immunology, Rheumatology
VEXAS Syndrome	*UBA-1* (c.121A>C & c.122T>C & c.121A>G)	Restrictions: None **Requires consultation with Lab Scientist prior to ordering**
METABOLIC		
Acid Sphingomyelinase Deficiency (ASMD)	*SMPD1* Single Gene Testing	GENETICS Only
Arysulfatase A Deficiency/ Metachromic Luekodystrophy (MLD)	*ARSA* Single Gene Testing	GENETICS Only
Bartter Syndrome	Bartter Syndrome Panel Genes: *AP2S1-BSND-CASR-CLCNKA-CLCNKB-GNA11-KCNJ1-MAGED2-SLC12A1-SLC12A3*	GENETICS Only
Biotinidase Deficiency	*BTD* Single Gene Test	GENETICS Only
Crigler Najjar Syndrome Type I & II	*UGT1A1* Single Gene Test	GENETICS Only
Carnitine Deficiency (Systemic Primary)	*SLC22A5* Single Gene Test	GENETICS Only

Dihydropyrimidine Dehydrogenase Deficiency	*DPYD* Sanger (c.1905+1G>A, c.1679T>G, c.2846A>T, c.1236G>A, and c.1129-5923C>G)	Diagnosis: Oncology
Fabry Disease	*GLA* Single Gene Test	Diagnosis: GENETICS, Neurology, Internal Medicine, Nephrology
Galactosemia	*GALT* Single Gene Test	GENETICS Only
Gaucher Disease	*GBA* Single Gene Test	GENETICS Only
Glucose Transporter Type I Deficiency Syndrome	*SLC2A1* Single Gene Test	GENETICS Only
Medium Chain acyl-CoA Dehydrogenase Deficiency	*ACADM* Single Gene Test	GENETICS Only
Mucopolysaccharidosis Type I (MPSI)/Hurler Syndrome	*IDUA* Single Gene Test	GENETICS Only
Nieman-Pick Disease Type C	*NPC1* and *NPC2* Gene Sequencing and Del/Dup	GENETICS Only
Phenylketonuria (PKU)	*PAH* Single Gene Test	Diagnosis: GENETICS, New Brunswick PKU Program
Very Long-Chain acyl-CoA Dehydrogenase Deficiency	*ACADVL* Single Gene Test	GENETICS Only
GI/NEPHROLOGY		
Familial Wilms Tumor	Familial Wilms Tumor via fPCR (Linkage study)	GENETICS Only
Polycystic Kidney Disease	Polycystic Kidney Disease Panel Genes: *DNAJB11-DZIP1L-GANAB-HNF1B-JAG1-LRP5-NOTCH2-PKD1-PKD2-PKHD1-PRKCSH- SEC63*	GENETICS Only
NEUROLOGY		

Amyotrophic Lateral Sclerosis	*SOD1* and *C9Orf72* Analysis	Diagnosis: GENETICS, Neurology, Physiatry **Testing includes both *SOD1* by NGS and *C9Orf72* by fPCR**
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS* (c.6594delT & c.5254C>T only)	GENETICS Only
Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)	*NOTCH3* Single Gene Test	Diagnosis: GENETICS, Neurology
Charcot Marie Tooth 1A (CMT1A) and Hereditary Neuropathy with Pressure Palsies (HNPP)	*PMP22* dosage via MLPA	Diagnosis: None
Duchenne Muscular Dystrophy/Becker Muscular Dystrophy	*DMD* Single Gene Test	Diagnosis: GENETICS, Neurology, Pediatrics - Pediatrics can order *only* for symptomatic young males (<13 yo)
Episodic Ataxia Type 2 (EA2) and Spinocerebellar Ataxia Type 6 (SCA6)	*CACNA1A* Single Gene Test	Diagnosis: GENETICS, Neurology **CGL cannot detect clinically relevant repeat expansions in this gene** - If request is due to EA2: Complete in-house test and refer-out if negative - If request is due to SCA6: Proceed to refer-out
Familial Hemiplegic Migraine (FHM)	*CACNA1A* and *SCN1A* Gene Sequencing and Del/Dup	Diagnosis: GENETICS, Neurology *Analysis only accounts for ~15% of FHM**
Friedreich Ataxia (FRDA)	FRDA kit and Triplet-primed PCR repeat amplification	Diagnosis: GENETICS, Neurology
Huntington Disease (HD)	HD via fPCR and Triplet-primed PCR combined assay	Diagnosis: GENETICS, Neurology, Psychiatry

Myotonic Dystrophy Type 1 (DM1)	DM1 via fPCR and Triplet-primed PCR	Diagnosis: GENETICS, Neurology, Pediatrics
Periodic Paralysis	Periodic Paralysis Panel Genes: *CACNA1S-CLCN1-KCNJ2-SCN4A*	Diagnosis: GENETICS, Neurology
SCN1A Seizure Disorder	*SCN1A* Single Gene Test	Diagnosis: GENETICS, Neurology
SCN9A Neuropathic Pain Syndromes	*SCN9A* Single Gene Test	Diagnosis: GENETICS, Neurology, Rheumatology
SGCE Myoclonus Dystonia	*SGCE* Single Gene Test	Diagnosis: GENETICS, Neurology
Spinal Muscular Atrophy (SMA)	SMN Gene Dosage via MLPA	Diagnosis: GENETICS, Neurology, Pediatrics
ONCOLOGY		
BRCA1 & BRCA2	*BRCA1* and *BRCA2* Gene Sequencing and Del/Dup	Diagnosis: GENETICS, MedOncPilot Project **This includes Breast Cancer patients from New Brunswick, Nova Scotia and PEI**
Colorectal Cancer	Colorectal Cancer Panel Genes: *APC-BMPR1A-EPCAM* (3' del only)-*MLH1-MSH2-MSH6-MUTYH-PMS2-PTEN-SMAD4-STK11-TP53*	GENETICS Only
Hereditary Breast and Ovarian Cancer (HBOC)	HBOC Panel Genes: *ATM-ATM c.5763-1050A>G-BARD1-BRCA1-BRCA2-BRIP1-CDH1-CHEK2-EPCAM* (3' del only)-*MLH1-MSH2-MSH6-NBN* c.657_661del5-*PALB2-PMS2-PTEN-RAD51C-RAD51D-STK11-TP53*	GENETICS Only
Hereditary Breast Cancer	Hereditary Breast Cancer Panel Genes: *ATM-ATM c.5763-1050A>G-BARD1-BRCA1-BRCA2-CDH1-CHEK2-NBN* c.657_661del5-*PALB2-PTEN-STK11-TP53*	GENETICS Only

Hereditary Cancers	Hereditary Cancers Panel Genes: *AIP-APC-ATM-ATM* c.5763-1050A>G-*BAP1-BARD1-BMPR1A-BRCA1-BRCA2-BRIP1-CASR-CDC73-CDH1-CDK4-CDKN1B-CDKN2A-CHEK2-DICER1-EPCAM* (3' del only)-*FH-FLCN-HOXB13-MAX-MEN1-MET-MLH1-MSH2-MSH6-MUTYH-NBN* c.657_661del5-*PALB2-NF1-PALB2-PMS2-PTCH1-PTEN-RAD51C-RAD51D-RET-SDHA-SDHAF2-SDHB-SDHC-SDHD-SMAD4-STK11-SUFU-TMEM127-TP53-TSC1-TSC2 & VHL*	GENETICS Only
Hereditary Ovarian Cancer	Hereditary Ovarian Cancer Panel Genes: *BRCA1-BRCA2-BRIP1-EPCAM* (3' del only)-*MLH1-MSH2-MSH6-PMS2-RAD51C-RAD51D*	GENETICS Only
Lymphoid Neoplasms	Lymphoid Neoplasms Panel Genes: AC*D-ARID1A-ATM-ATM* c.5763-1050A>G-*BLM-BRCA1-BRCA2-BRIP1-CHEK2-CSF3R-DDX41-DICER1-DIS3-ETV6-FAS-IKZF1-MLH1-MSH2-MSH6-NBN-NF1-PALB2-PAX5-PMS2-PTEN-POT1-RAD51C-RAD51D-RUNX1-SDHB-SDHC-SDHD-TERF2IP-TET2-TP53-USP45*	Diagnosis: Dr. Trottier, GENETICS
Lynch Syndrome	Lynch Syndrome Panel Genes*: EPCAM* (3' del only)-*MLH1-MSH2-MSH6-PMS2*	GENETICS Only
Melanoma	Melanoma Panel Genes: *BAP1-BRCA2-CDK4-CDKN2A-PTEN-TP53*	GENETICS Only
Multiple Endocrine Neoplasia Type 1 (MEN1)	*MEN1* Single Gene Test	GENETICS Only
Multiple Endocrine Neoplasia Type 2 (MEN2)	*MEN1* Single Gene Test	GENETICS Only

MUTYH Associated Polyposis	*MUTYH* Single Gene Analysis	GENETICS Only
Myeloid Malignancy	Myeloid Malignancy Panel Genes: *ANKRD26-ATM-BLM-BRCA1-BRCA2-BRIP1-CBL-CEBPA-CHEK2-CSF3R-DDX41-DKC1* (including 5'UTR)-*ETV6-FANCA-GATA2* (including intron 4)-*IKZF1-MLH1-MSH2-MSH6-NBN-NF1-NHP2-PALB2-PARN-PAX5-PMS2-PTPN11-RAD51C-RTEL1-RUNX1-SAMD9-SAMD9L-SBDS-SRP72-TERC-TERT-TET2-TINF2-TP53*	Diagnosis: GENETICS, Dr. Trottier
Neuroendocrine Tumors	Neuroendocrine Tumor Panel Genes: *CDKN1B-MEN1-NF1-TSC1-TSC2-VHL*	GENETICS Only
Nevoid Basal Cell Carcinoma Syndrome	*PTCH1* and *SUFU* Gene Sequencing and Del/Dup	GENETICS Only
Pancreatic Cancer	Pancreatic Cancer Panel Genes: *ATM-ATM* c.5763-1050A>G-*BRCA1-BRCA2-CDKN2A-EPCAM* (3' del only)-*MLH1-MSH2-MSH6-PALB2-PMS2-STK11*	GENETICS Only
Pheochromocytoma & Paraganglioma	Pheochromocytoma & Paraganglioma Panel Genes: *FH-MAX-NF1-RET-SDHA-SDHB-SDHC-SDHD-SDHAF2-TMEM127-VHL*	GENETICS Only
Polyposis	Polyposis Panel Genes: *APC-BMPR1A-MUTYH-PTEN-SMAD4-STK11*	GENETICS Only
Prostate Cancer	Prostate Cancer Panel Genes: *ATM-ATM c.5763-1050A>G-BRCA1-BRCA2-CHEK2-EPCAM* (3' del only)-*HOXB13-MLH1-MSH2-MSH6-PMS2*	GENETICS Only
Renal Cell Carcinoma (RCC)	Renal Cell Carcinoma Panel	GENETICS Only

	Genes: *BAP1-FH-FLCN-MET-PTEN-SDHB-SDHC-SDHD-TMEM127-TSC1-TSC2-VHL*	
Retinoblastoma	*RB1* Single Gene Analysis	GENETICS Only **Refer out to Impact Genetics unless germline *RB1* sequence analysis alone is needed**
Tuberous Sclerosis Complex (TSC)	*TSC1* and *TSC2* Gene Sequencing and Del/Dup	Diagnosis: Dermatology, GENETICS, Neurology
Von Hippel-Lindau (VHL) Syndrome	*VHL* Single Gene Analysis	GENETICS Only
OPHTHALMOLOGY		
Aniridia	Aniridia Panel Genes: *PAX6* (SNV and dosage)-*WT1* (dosage only)	GENETICS Only
SKELETAL		
Achondroplasia (common mutations only)	*FGFR3* c.1138G>A and c.1138G>G via Sanger sequencing	Restrictions: None
Apert Syndrome (common mutations only)	*FGFR2* c.775C>G and c.758C>G via Sanger sequencing	Restrictions: None
Cleidocranial Dysplasia (CCD) Spectrum Disorder	*RUNX2* Single Gene Analysis	GENETICS Only

****Single Genes that are not included in this list can be requested on a case-by-case basis – Please contact the Clinical Genomics Laboratory about your specific case****