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The IWK CGL is repatriating genetic testing. As we expand our testing menu to meet the needs of our patients in the Maritimes, referred out requests are subject to review. Ordering providers will be informed if their test of choice will be performed in-house and will no longer be referred out to an external laboratory.

Ordering Restrictions: Carrier and predictive testing, as well as are currently restricted to Maritime Medical Genetics Service (MMGS). Symptomatic testing might be restricted or allowed based on the test and the ordering provider's clinical specialty.

For any questions, please consult the lab genetic counsellor or a laboratory scientist.
For any test restricted to Maritime Medical Genetics Services (MMGS), please refer your patient to their department or contact them at 902.470.8754.

Please contact the IWK CGL Laboratory Genetic Counsellors for refer out requests for tests <100 genes to determine if your request can be offered in house. Single genes that are not included in this list can be requested on a case-by-case basis – Please contact the Laboratory GCs at clinicalgenomics.gc@iwk.nshealth.ca

Indication for Testing	Test Name	Gene Content / Methodology and Laboratory	Ordering Restrictions
BANKING – LONG TERM DNA STORAGE (25 YEARS)			
Irreplaceable samples that require long term storage	Bank – DNA Long Term Banking	N/A	Restrictions: None but must meet criteria for long-term banking* *This option is indicated for circumstances in which our 5-year retention is insufficient such as: patient being palliative and future testing for family members is likely to occur.
CARDIOLOGY			
Arrhythmias	Arrhythmia Panel	IWK Clinical Genomics Laboratory: ANK2-CACNA1C-CACNB2-CALM1-CALM2-CALM3-CASQ2-CAV3-CDH2-DSC2-DSG2-DSP-FLNC-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, Mainstreamed Cardiologists
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	Arrhythmogenic Right Ventricular Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: CDH2-DSC2-DSG2-DSP-JUP-PKP2-RYR2-TMEM43	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreamed Cardiologists
Brugada Syndrome	Brugada Panel	IWK Clinical Genomics Laboratory: SCN5A	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreamed Cardiologists

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Cardiomyopathies	Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: ABCC9-ACTC1-ACTN2-ANKRD1-BAG3-CAV3-CDH2-CSRP3-DES-DMD-DSC2-DSG2-DSP-EMD-FLNC-GLA-JUP-LAMP2-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	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreamed Cardiologists
Catecholaminergic Polymorphic Ventricular tachycardia (CPVT)	Catecholaminergic Polymorphic Ventricular Tachycardia Panel	IWK Clinical Genomics Laboratory: CALM1-CASQ2-RYR2-TECRL-TRDN	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreamed Cardiologists
Dilated Cardiomyopathy (DCM)	Dilated Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: 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, Mainstreamed Cardiologists
Familial Hypercholesterolemia (FH)	Familial Hypercholesterolemia Panel	IWK Clinical Genomics Laboratory: APOB-LDLR-LDLRAP1-PCSK9	None but must meet criteria for testing *Provide Dutch Lipid Network Diagnostic Criteria (adults) or Simone Broome Diagnostic Criteria (children) score.
Holt-Oram Syndrome (HOS)	*TBX5* Single Gene Testing	IWK Clinical Genomics Laboratory: TBX5	GENETICS Only
Hypertrophic Cardiomyopathy (HCM)	Hypertrophic Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: 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, Mainstreaming Cardiologists
Inherited dyslipidemia	Hyperlipidemia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service
Left Ventricular Non-Compaction Cardiomyopathy (LVNC)	Left Ventricular Non-Compaction Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: ACTC1-LDB3-LMNA-MYBPC3-MYH7-TAFAZZIN-TNNI3-TNNT2-TPM1-TTN	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreaming Cardiologists
Long QT Syndrome (LQTS)	Long QT Panel	IWK Clinical Genomics Laboratory: ANK2-CALM1-CALM2-CALM3-CACNA1C-CAV3-	Diagnosis: GENETICS, Nova Scotia Medical Examiner's Service, Mainstreaming Cardiologists

		KCNE1-KCNE2-KCNH2-KCNJ2-KCNQ1-SCN5A	
Structural heart defects	Syndromic Cardiac Defect Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Transthyretin Amyloidosis	*TTR* Single Gene Testing	IWK Clinical Genomics Laboratory: TTR	Restrictions: None

CONNECTIVE TISSUE			
Aortopathy	Aortopathy Panel	IWK Clinical Genomics Laboratory: 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, Mainstreamed Cardiologists
Ehlers-Danlos Syndrome (EDS)	Ehlers-Danlos Syndrome (EDS) Panel	IWK Clinical Genomics Laboratory: ACTA2-ADAMTS2-ADAMTSL2-AEBP1-ALDH18A1-ATP6V0A2-ATP6V1A-ATP6V1E1-ATP7A-B3GALT6-B3GAT3-B4GALT7-BGN-C1R-C1S-CBS-CHST14-CHST3-COL11A1-COL11A2-COL12A1-COL1A1-COL1A2-COL2A1-COL3A1-COL5A1-COL5A2-COL6A1-COL6A2-COL6A3-COL9A1-COL9A2-COL9A3-DSE-EFEMP1-EFEMP2-ELN-EMILIN1-FBLN5-FBN1-FBN2-FKBP14-FLNA-FLNB-GORAB-LOX-LTBP4-MED12-PLOD1-PRDM5-PYCR1-RIN2-SKI-SLC2A10-SLC39A13-SMAD2-SMAD3-TAB2-TGFB2-TGFB3-TGFBR1-TGFBR2-TNXB-ZNF469	GENETICS Only
Hereditary Connective Tissue Disorders	Hereditary Disorder of Connective Tissue Panel	IWK Clinical Genomics Laboratory: ACTA2-ADAMTS2-AEBP1-ATP7A-B3GALT6-B4GALT7-BGN-C1R-C1S-CBS-CHST14-COL12A1-COL1A1-COL1A2-COL3A1-COL5A1- COL5A2-DSE-EFEMP2-FBN1-FBN2-FKBP14-FLNA-FOXE3-HCN4-LOX-MAT2A-MFAP5-MYH11-MYLK-PLOD1-PRDM5-PRKG1 (c.530G>A;p.R177Q only)-SKI-	GENETICS Only

		SLC2A10-SLC39A13-SMAD2-SMAD3-TGFB2-TGFB3-TGFBR1-TGFBR2-TNXB-YY1AP1 (c.1079C>T only) and ZNF469	
Hereditary Hemorrhagic Telangiectasia (HHT)	Hereditary Hemorrhagic Telangiectasia (HHT) Panel	IWK Clinical Genomics Laboratory: ACVRL1-ENG-EPHB4-GDF2-RASA1-SMAD4	GENETICS Only
Loeys-Dietz Syndrome	Loeys-Dietz Syndrome Panel	IWK Clinical Genomics Laboratory: SMAD2-SMAD3-TGFB2-TGFB3-TGFBR1-TGFBR2	GENETICS Only
Marfan Syndrome	*FBN1* Single Gene Testing	IWK Clinical Genomics Laboratory: FBN1	GENETICS Only
Stickler Syndrome	Stickler Syndrome Panel	IWK Clinical Genomics Laboratory: BMP4-COL11A1-COL11A2-COL2A1-COL9A1-COL9A2-COL9A3-GZF1-VCAN	GENETICS Only

DERMATOLOGY			
Adams-Oliver Syndrome	Adams-Oliver Syndrome Panel	IWK Clinical Genomics Laboratory: ARHGAP31-DLL4-DOCK6-EOGT-KCTD1-NOTCH1-RBPJ-UBR1	GENETICS Only
Oculocutaneous Albinism	Albinism Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Dermatology, GENETICS, Ophthalmology
Amelogenesis and Dentinogenesis Imperfecta	Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Cutis Laxa	Cutis Laxa Panel	IWK Clinical Genomics Laboratory: ELN-ATP6V0A2-EFEMP2-FBLN5-LTBP4-PYCR1	GENETICS Only
Ectodermal Dysplasia (Hidrotic or Hypohidrotic)	Ectodermal Dysplasia Panel	Blueprint Genetics: Please check lab details for gene content	Restrictions: None
Epidermolysis Bullosa	Epidermolysis Bullosa Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Dermatology, GENETICS
Ichthyosis	Ichthyosis Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Dermatology, GENETICS
	X-Linked Ichthyosis via MLPA	IWK Clinical Genomics Laboratory: XLI-MLPA	Restrictions: None

Incontinentia Pigmenti	IKBKG (NEMO) Gene Sequencing & Common Del/Dup	GeneDx: Please check lab details	Diagnosis: Dermatology, GENETICS
Palmoplantar Keratoderma	Palmoplantar Keratoderma Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Dermatology, GENETICS
Pseudoxanthoma Elasticum (PXE)	*ABCC6* and *GGCX* Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: ABCC6-GGCX	Diagnosis: Dermatology, GENETICS, Ophthalmology
Xeroderma Pigmentosum (XP)	Xeroderma Pigmentosum Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only

ENDOCRINOLOGY			
Congenital Adrenal Hyperplasia (CAH)	Congenital Adrenal Hyperplasia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS
Disorders of Sex Development (DSD)	Abnormal Genitalia/Disorders of Sexual Development Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS
Familial Hyperaldosteronism	Primary Hyperaldosteronism NGS Panel	Prevention Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS
	Pseudohypoaldosteronism Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS
Familial Hyperparathyroidism	Hyperparathyroidism Panel	IWK Clinical Genomics Laboratory: CASR-CDC73-CDKN1B-MEN1-RET	Diagnosis: Endocrinology, GENETICS
Familial Hypocalciuric Hypercalcemia (FHH)	Familial Hypocalciuric Hypercalcemia (FHH) Panel	IWK Clinical Genomics Laboratory: AP2S1-GNA11-CASR	Diagnosis: Endocrinology, GENETICS
Hereditary Nephrogenic Diabetes Insipidus	Hereditary Nephrogenic Diabetes Insipidus Panel	IWK Clinical Genomics Laboratory: AQP2-AVP-AVPR2	Diagnosis: Endocrinology, GENETICS
Hypomagnesemia	Hypomagnesemia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS, Internal Medicine
Hypothyroidism and Thyroid Resistance	Hyperparathyroidism Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS
	Hypothyroidism and Resistance to Thyroid Hormone Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS

Hypogonadotropic Hypogonadism/Kallmann syndrome	Kallman Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS, Internal Medicine
Hypophosphatasia	*ALPL* Single Gene Testing	IWK Clinical Genomics Laboratory: ALPL	Diagnosis: Endocrinology, GENETICS
Hereditary Hypophosphatemic Rickets	Hereditary Hypophosphatemic Rickets Panel	IWK Clinical Genomics Laboratory: ALPL-CLCN5-CTNS-CYP27B1-CYP2R1-DMP1-ENPP1-FAH-FAM20C-FGF23-OCRL-PHEX-SLC34A1-SLC34A3-SLC9A3R1-VDR	GENETICS Only
Liddle Syndrome	Liddle Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS, Nephrology
Lipodystrophy	Congenital and Familial Lipodystrophy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Mature onset diabetes of the young (MODY)	Maturity-Onset Diabetes of the Young Panel	IWK Clinical Genomics Laboratory: ABCC8-GCK-HNF1A-HNF1B-HNF4A-INS-KCNJ11-PDX1	Diagnosis: Endocrinology, Internal Medicine, GENETICS
Monogenic Obesity	Monogenic Obesity Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Primary Macronodular Adrenal Hyperplasia / Primary Pigmented Nodular Adrenocortical Disease (PMAH/PPNAD)	Primary Macronodular Adrenal Hyperplasia / Primary Pigmented Nodular Adrenocortical Disease (PMAH/PPNAD) Panel	IWK Clinical Genomics Laboratory: ARMC5, KDM1A, PDE11A, PDE8B, APC, MEN1, PRKACA (duplication only), PRKAR1A	GENETICS Only

EAR, NOSE AND THROAT (ENT)			
Branchio-Oto-Renal (BOR) Syndrome	Branchio-Oto-Renal (BOR) Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Pediatric ENT
Non-Syndromic and Syndromic Hearing Loss	Non-Syndromic Hearing Loss	IWK Clinical Genomics Laboratory: GJB2 + GJB6 Gene Sequencing and Del/Dup	Restrictions: None

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	Non-Syndromic Hearing Loss Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only *In-house testing (*GJB2* + *GJB6*) must be performed first, if relevant
	Syndromic Hearing Loss Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Comprehensive Hearing Loss and Deafness Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Pendred Syndrome	Pendred Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: Endocrinology, GENETICS, Pediatric ENT
Usher Syndrome (Type 1, Type 2)	Usher Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Single Gene Testing Only	IWK Clinical Genomics Laboratory: Single genes associated with Usher Syndrome can be requested through CGL if applicable	GENETICS Only
Waardenburg Syndrome	Waardenburg Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Pediatric ENT

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GENERAL DISORDERS			
22q11.2 Deletion and Duplication Syndrome	22q11.2 Deletion/Duplication MLPA	IWK Clinical Genomics Laboratory: 22q11.2 Del/Dup by MLPA	Restrictions: None
Adams-Oliver Syndrome	Adams-Oliver Syndrome Panel	IWK Clinical Genomics Laboratory: ARHGAP31-DLL4-DOCK6-EOGT-KCTD1-NOTCH1-RBPJ-UBR1	GENETICS Only
Adrenoleukodystrophy	*ABCD1* Single Gene Testing	IWK Clinical Genomics Laboratory: ABCD1	GENETICS Only
Alström Syndrome	*ALMS1* Single Gene Testing	IWK Clinical Genomics Laboratory: ALMS1	GENETICS Only
Angelman Syndrome	15q11 Methylation and Dosage	AS-MLPA	Restrictions: None
Ashkenazi Jewish Carrier Testing	Ashkenazi Jewish Screening Panel	SickKids: Please check lab details for gene content	GENETICS Only
Bardet-Biedl Syndrome (BBS)	Bardet-Biedl Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Beckwith-Wiedemann Syndrome	Beckwith-Wiedemann Syndrome MLPA/NGS	BWS-MLPA followed by CDNK1C sequencing	Restrictions: None *Testing includes MS-MLPA & reflex to NGS if MLPA negative
Carney Complex	*PRKAR1A* Single Gene Testing	IWK Clinical Genomics Laboratory: PRKAR1A	GENETICS Only
CHD7 Disorder	*CHD7* Single Gene Testing	IWK Clinical Genomics Laboratory: CHD7	GENETICS Only
Cleft Lip/Palate/Vanderwoude	Cleft Lip/Palate and Associated Syndromes Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Cornelia de Lange Syndrome	Cornelia de Lange Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Ciliopathies, Heterotaxy/Situs Inversus and Primary Ciliary Dyskinesia (PCD)	Ciliopathy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Heterotaxy and Situs Inversus Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Prenatal Heterotaxy Panel	IWK Clinical Genomics Laboratory: ACVR2B-ANKS6-CCDC103-CCDC39-CCDC40-CFAP298-CFAP300-CFAP53-DNAAF1-DNAAF11-	GENETICS Only

		DNAAF2-DNAAF3-DNAAF4-DNAAF5-DNAAF6-DNAH1-DNAH11-DNAH5-DNAH9-DNAI1-DNAI2-DNAL1-FOXJ1-GDF1-INVS-LRRC56-MMP21-MNS1-NKX2-5-NME8-NODAL-ODAD1-ODAD2-ODAD3-ODAD4-PKD1L1-SMAD2-SPAG1-ZIC3-ZMYND10	
	Primary Ciliary Dyskinesia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Pediatric Respirology
Cystic Fibrosis	*CFTR* Single Gene Testing	IWK Clinical Genomics Laboratory: CFTR	Restrictions: None *Must meet criteria for testing. If testing is for diagnostic purposes, request must include sweat chloride test results and symptoms.
Developmental Delay, Autism Spectrum Disorder, Multiple Congenital Anomalies	Microarray (Illumina CytoSNP-850K)	Microarray	Postnatal: None Prenatal: GENETICS only unless POC *Must meet criteria for testing
Down Syndrome (postnatal)	Down Syndrome via QF-PCR	IWK Clinical Genomics Laboratory: Rapid Aneuploidy Testing (RAD)	Restrictions: None
Fragile X Syndrome	*FMR1* Analysis (Triplet-primed PCR)	IWK Clinical Genomics Laboratory: FMR1	Restrictions: None (but must meet clinical 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 (including siblings) 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	*FMR1* Analysis (Triplet-primed PCR)	IWK Clinical Genomics Laboratory: FMR1	Diagnosis: None

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Ovarian Failure			
Fraser Syndrome	Fraser Syndrome Panel	IWK Clinical Genomics Laboratory: FRAS1-FREM1-FREM2-GRIP1	GENETICS Only
GNAS Single Gene Disorders	*GNAS* Single Gene Testing	IWK Clinical Genomics Laboratory: GNAS	GENETICS Only *Requires consultation with Lab GC / Lab Scientist prior to ordering
Hereditary Multiple Osteochondromas	Hereditary Multiple Osteochondromas Panel	IWK Clinical Genomics Laboratory: EXT1-EXT2	GENETICS Only
Joubert Syndrome	Joubert Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Kabuki Syndrome	Kabuki Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Klinefelter Syndrome	Klinefelter Syndrome via QF-PCR	IWK Clinical Genomics Laboratory: Rapid Aneuploidy Testing (RAD)	Restrictions: None
L1 Syndrome	*L1CAM* Single Gene Testing	IWK Clinical Genomics Laboratory: L1CAM	GENETICS Only
Male Infertility – Y Microdeletion	Y Microdeletion Testing	IWK Clinical Genomics Laboratory: Devyser AZF microdeletion fPCR	Restrictions: None
Male Infertility – CFTR sequencing (CBAVD)	CFTR Single Gene Testing	IWK Clinical Genomics Laboratory: CFTR	Restrictions: None
Malignant Hyperthermia Susceptibility Disorder	Malignant Hyperthermia Susceptibility Disorder Panel	IWK Clinical Genomics Laboratory: CACNA1S-RYR1-STAC3	GENETICS Only
Maternal DNA Contamination (MCC studies)	QF-PCR	IWK Clinical Genomics Laboratory:	Restrictions: None
Meckel Syndrome	Meckel Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Neurofibromatosis Type 1	*NF1* and *SPRED1* Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: NF1-SPRED1	Diagnosis: Dermatology, GENETICS, Neurology, Pediatrics *Must meet criteria for testing.
Neurofibromatosis Type 2	*NF2* Single Gene Testing	IWK Clinical Genomics Laboratory: NF2	GENETICS Only
Noonan Syndrome	Noonan Syndrome Panel	IWK Clinical Genomics Laboratory:	GENETICS Only

		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)	
Prader Willi Syndrome (PWS)	15q11 Methylation and Dosage MLPA	IWK Clinical Genomics Laboratory: PWS-MLPA	Restrictions: None
Primary Ciliary Dyskinesia	Primary Ciliary Dyskinesia Panel	IWK Clinical Genomics Laboratory: CCDC39-CCDC40-CCDC65-CCDC103-CCNO-CFAP221-CFAP298-CFAP300-CFTR-DNAAF1-DNAAF11-DNAAF2-DNAAF3-DNAAF4-DNAAF5-DNAAF6-DNAH1-DNAH11-DNAH5-DNAH8-DNAH9-DNAI1-DNAI2-DNAJB13-DNAL1-DRC1-FOXJ1-GAS2L2-GAS8-HYDIN-LRRC56-MCIDAS-NEK10-NME8-ODAD1-ODAD2-ODAD3-ODAD4-OFD1-RPGR-RSPH1-RSPH3-RSPH4A-RSPH9-SPAG1-SPEF2-STK36-TTC12-ZMYND10	Diagnosis: GENETICS, Respirology *Must meet criteria for testing
PTEN-related Macrocephaly/Autism Spectrum Disorder	*PTEN* Single Gene Testing	IWK Clinical Genomics Laboratory: PTEN	GENETICS Only
Rett Syndrome	*MECP2* Single Gene Testing	IWK Clinical Genomics Laboratory: MECP2	Diagnosis: GENETICS, Neurology, Pediatrics
Russell-Silver Syndrome (RSS)	Russell-Silver Syndrome Methylation and Dosage MLPA	IWK Clinical Genomics Laboratory: RSS-MLPA	Restrictions: None
Schwannomatosis / Multiple Schwannomas	Schwannomatosis Panel	IWK Clinical Genomics Laboratory: LZTR1-NF2-SMARCB1	GENETICS Only
Sotos Syndrome	*NSD1* Single Gene Testing	IWK Clinical Genomics Laboratory: NSD1	GENETICS Only *15-50% of probands detected via array (order array in conjunction if applicable)
Stickler Syndrome	Stickler Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Subcutaneous Panniculitis-like T-cell Lymphoma	*HAVCR2* Single Gene Test	IWK Clinical Genomics Laboratory: HAVCR2	Diagnosis: GENETICS, Hematology, Immunology *Please consult the Clinical Genomics Lab prior to ordering

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Tay-Sachs Disease	*HEXA* Single Gene Test	IWK Clinical Genomics Laboratory: HEXA	Diagnosis: GENETICS, Neurology *Requires HEX A enzyme activity prior to testing
Turner Syndrome	Turner Syndrome via QF-PCR	IWK Clinical Genomics Laboratory: Rapid Aneuploidy Testing (RAD)	Restrictions: None
Wilson Disease	*ATP7B* Single Gene Testing	IWK Clinical Genomics Laboratory: ATP7B	Diagnosis: Gastrointestinal, GENETICS, Neurology

GI/NEPHROLOGY			
Alport Syndrome	Alport Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nephrology
Cholestasis	Cholestasis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Congenital Diarrhea	Congenital Diarrhea Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, GI
Hirschsprung Disease	Hirschsprung Disease Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, GI *Pathology report must be provided
Nephrolithiasis	Nephrolithiasis Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nephrology
Nephrotic Syndrome	Nephrotic Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nephrology
Pancreatitis	Hereditary Pancreatitis Panel	IWK Clinical Genomics Laboratory: CFTR-PRSS1-SPINK1	Diagnosis: GENETICS, Gastroenterology, Internal Medicine, Hepatology, Pediatrics *Must meet testing criteria – check with Lab GC or Lab Scientist
Polycystic Kidney Disease	Polycystic Kidney Disease Panel	IWK Clinical Genomics Laboratory: DNAJB11-DZIP1L-GANAB-HNF1B-JAG1-LRP5-NOTCH2-PKD1-PKD2-PKHD1-PRKCSH- SEC63	GENETICS Only
Polycystic Liver Disease	Polycystic Liver Disease Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, GI

Renal Malformations	Renal Malformation Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Renal Tubular Acidosis	Renal Tubular Acidosis Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nephrology

HEMATOLOGY			
Atypical Hemolytic Uremic Syndrome (aHUS)	Hemolytic Uremic Syndrome Panel	IWK Clinical Genomics Laboratory: ADAMTS13-C3-CD46-CFB-CFH-CFHR5-CFI-DGKE-THBD	Diagnosis: Hematology, GENETICS, Nephrology
Bone Marrow Failure	Bone Marrow Failure Syndrome Panel	IWK Clinical Genomics Laboratory: ACD-AK2-AP3B1-BRCA1-BRCA2-BRIP1-CSF3R-CTC1-CTLA4-CXCR4-DKC1-DNAJC21-EFL1-ELANE-ERCC4-ERCC6L2-FANCA-FANCB-FANCC-FANCD2-FANCE-FANCF-FANCG-FANCI-FANCL-FANCM-G6PC3-GATA1-GATA2-GFI1-HAX1-HOXA11-KRAS-LIG4-MDM4-MECOM-MPL-MYSM1-NAF1-NBN-NHP2-NOP10-NRAS-PALB2-PARN-PRF1-RAD51C-RBM8A-RPL11-RPL15-RPL26-RPL35A-RPL5-RPS10-RPS19-RPS24-RPS26-RTEL1-SAMD9-SAMD9L-SBDS-SLC25A38-SLX4-SRP54-SRP72-STN1-TERF2IP-TERC-TERT-TINF2-TP53-UBE2T-USB1-VPS45-WAS-WRAP53-XRCC2-ZCCHC8	Diagnosis: GENETICS, Hematology, Immunology
Coagulopathy	Rare Bleeding Disorders (including): Factor V/VII/X/XI/XIII	The Canadian National Inherited Bleeding Disorder Genotyping Laboratory – Queen's University: Please check lab details	GENETICS Only *A clotting factor level is required
	Bleeding Disorder/Coagulopathy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Hematology
Congenital Neutropenia	Congenital Neutropenia Panel	IWK Clinical Genomics Laboratory: ACTB-AP3B1-AP3D1-CD40LG-CEBPE-CLPB-CSF3R-CTSC-CXCR2-CXCR4-DNAJC21-EIF2AK3-EFL1-ELANE-G6PC3-GATA2-GFI1-GINS1-HAX1-HYOU1-	Diagnosis: GENETICS, Hematology, Immunology

		IFNGR2-JAGN1-LAMTOR2-LYST-PGM3-RAB27A-RAC2-SBDS-SLC37A4-SMARCD2-SRP54-STK4-TAFAZZIN-TCN2-USB1-VPS13B-VPS45-WAS-WDR1-WIPF1	
Diamond-Blackfan Anemia	Diamond-Blackfan Anemia Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Dyskeratosis Congenita	Telomere Length Testing	RepeatDX: Please check lab details for gene content	Diagnosis: GENETICS, Hematology, Hematology-Oncology
	Dyskeratosis Congenita Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Hematology, Hematology-Oncology *Order telomere length testing first before proceeding to panel (if applicable)
Factor V Leiden Thrombophilia	Factor V (Leiden) Test	Department of Pathology and Laboratory Medicine – Central Zone (QEII): Please check lab details	*Contact their lab for ordering instructions*
Fanconi Anemia (FA)	Fanconi Anemia (DEB, MMC)	SickKids: Please check lab details	Diagnosis: GENETICS, Hematology
	Fanconi Anemia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Hematology
Hemophilia A and B	Factor VIII and Factor IX variant Analysis	The Canadian National Inherited Bleeding Disorder Genotyping Laboratory – Queen's University: Please check lab details	Diagnosis: GENETICS, Hematology Carrier Testing: GENETICS, Hematology* *Appropriate for young girls who truly appear to have a level of bleeding disorder (i.e. not every female with heavy periods) for whom a diagnosis isn't reached through standard hematological testing. ALL positive results in these cases need to be referred to genetics for further review.
Hereditary Hemochromatosis & Related Disorders	HFE Hemochromatosis at the QEII Laboratory	HFE p.Cys282Tyr (p.C282Y) and p.His63Asp (p.H63D)	Restrictions: None *Two common HFE variants only
	Hereditary Hemochromatosis & Related Disorders Panel	IWK Clinical Genomics Laboratory: FTL-HAMP-HFE-HJV-SLC40A1-TFR2	Diagnosis: GENETICS, Hematology, Hepatology, Internal Medicine *HFE common variants should be completed before ordering panel

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Hereditary Leukemia	Hereditary Leukemia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS
Hermansky-Pudlak Syndrome	Hermansky-Pudlak Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Hemoglobinopathies	Hemoglobin Variant	Hamilton Health Sciences: Please check lab details	Diagnosis: Hematology, MMGS *CBC, Hemoglobin electrophoresis and ferritin results are required for coordinating testing
	Sickle Cell Disease	Hamilton Health Sciences: Please check lab details	Diagnosis: Hematology, MMGS *CBC, Hemoglobin electrophoresis and ferritin results are required for coordinating testing
	Thalassemia	Hamilton Health Sciences: Please check lab details	Diagnosis: Hematology, MMGS Carrier Testing: Hematology *CBC, Hemoglobin electrophoresis and ferritin results are required for coordinating testing
Prothrombin Thrombophilia	Prothrombin/FII (G20210A) Test	Department of Pathology and Laboratory Medicine – Central Zone (QEII): Please check lab details	*Contact their lab for ordering instructions*
Red Cell Membrane Disorders	Red Cell Membrane Disorders Panel	IWK Clinical Genomics Laboratory: ANK1-EPB41-EPB42-KCNN4-PIEZO1-RHAG-SLC4A1-SPTA1-SPTB	Diagnosis: Hematology, Hematopathology, GENETICS
Schwachman-Diamond Syndrome	SBDS Single Gene Testing	IWK Clinical Genomics Laboratory: SBDS	Diagnosis: Hematology, GENETICS
Thrombocytopenia	Thrombocytopenia Panel	IWK Clinical Genomics Laboratory: ANKRD26-ETV6-FLNA-GBA-RUNX1-THBD	Diagnosis: Hematology, GENETICS
Von Willebrand Disease (VWD)	Analysis for Type 2N VWD B) Confirmation of type 1C, 2A, 2B or 2M VWD Variant analysis and prenatal testing for type 3 VWD	The Canadian National Inherited Bleeding Disorder Genotyping Laboratory – Queen's University: Please check lab details for gene content	Diagnosis: Hematology, MMGS *Please check Laboratory's website for studies required to complete testing *This test reduces the chance but does not eliminate VWD as a diagnosis

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IMMUNOLOGY/RHEUMATOLOGY			
Autoinflammatory Disease	Autoinflammatory Disease Panel	IWK Clinical Genomics Laboratory: ACP5-ADAR-CARD14-ELANE-IFIH1-IL1RN-IL36RN-ISG15-LPIN2-MEFV-MVK-NLRC4-NLRP1-NLRP12-NLRP3-NOD2-OTULIN-PLCG2-PRG4-PSENEN-PSMB8-PSTPIP1-RIGI(DDX58)-RNASEH2A-RNASEH2B-RNASEH2C-SAMHD1-SLC29A3-STING1-TNFAIP3-TNFRSF1A-TREX1-TRNT1	Diagnosis: Hematology, Immunology, GENETICS, Rheumatology
Complement System Disorder	Complement System Disorder Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Immunology
Familial Mediterranean Fever	MEFV Single Gene Testing	IWK Clinical Genomics Laboratory: MEFV	Diagnosis: Immunology, GENETICS, Rheumatology
Hemophagocytic Lymphohistiocytosis	Hemophagocytic Lymphohistiocytosis (HLH) Panel	IWK Clinical Genomics Laboratory: AP3B1-AP3D1-CARMIL2-CD27-CD70-CDC42-CTPS1-FAAP24-GATA2-IL2RB-ITK-LYST-MAGT1-MCM4-NLRC4-PIK3CD-PIK3R1-PRF1-PRKCD-RAB27A-RASGRP1-SH2D1A-SLC7A7-STX11-STXBP2-TNFRSF9-UNC13D-XIAP	Diagnosis: Hematology, Immunology, GENETICS
Hereditary Angioedema	Hereditary Angioedema Panel	F12, PLG, SERPING1	Diagnosis: Dermatology, Immunology, GENETICS
Hyper IgE Syndrome	Hyper IgE Syndrome Panel	IWK Clinical Genomics Laboratory: CARD11-DOCK8-PGM3-SPINK5-STAT3-IL6R-IL6ST-ZNF341-STAT6-STAT5B	Diagnosis: Immunology, GENETICS
Monogenic Inflammatory Bowel Disease	Monogenic Inflammatory Bowel Disease Panel	IWK Clinical Genomics Laboratory: ADAM17-AICDA-ALPI-ARPC1B-BACH2-CARD11-CARD8-CARMIL2-CASP8-CD3G-CD40LG-CD55-COL7A1-CTLA4-CYBA-CYBB-CYBC1-DCLRE1C-DEF6-DGAT1-DOCK8-ELF4-EPCAM-FCHO1-FERMT1-FOXP3-G6PC3-GUCY2C-HPS1-HPS4-HPS6-ICOS-IFIH1-IKBKG-IL10-IL10RA-IL10RB-IL21-IL21R-IL2RA-IL2RB-ITGB2-JAK1-LCT-LIG4-LRBA-MASP2-MEFV-MVK-MYO5B-NCF1-NCF2-NCF4-NEUROG3-NFAT5-NFKBIA-NLRC4-OTULIN-PCSK1-PIK3CD-PIK3R1-PLCG2-PLVAP-POLA1-RAG1-RAG2-RIPK1-SAMD9-	Diagnosis: Immunology, GENETICS

		SKIC2-SKIC3-SLC10A2-SLC26A3-SLC37A4-SLC39A4-SLC51B-SLC5A1-SLC9A3-SPINT2-STAT1-STAT3-STX3-STXBP2-TGFB1-TLR3-TNFAIP3-TRIM22-TTC7A-UNC45A-XIAP-ZAP70-ZBTB24	
Primary Immunodeficiency	Primary Immunodeficiency Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Immunology
Severe Combined Immunodeficiency	Severe Combined Immunodeficiency Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Immunology
VEXAS Syndrome	UBA-1 (c.121A>C & c.122T>C & c.121A>G)	IWK Clinical Genomics Laboratory: UBA-1 (c.121A>C & c.122T>C & c.121A>G)	Restrictions: None *Requires consultation with Lab prior to ordering

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METABOLIC			
Acid Sphingomyelinase Deficiency (ASMD)	SMPD1 Single Gene Testing	IWK Clinical Genomics Laboratory: SMPD1	GENETICS Only
Arysulfatase A Deficiency/ Metachromatic Leukodystrophy (MLD)	ARSA Single Gene Testing	IWK Clinical Genomics Laboratory: ARSA	GENETICS Only
Bartter Syndrome	Bartter Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Biotinidase Deficiency	BTD Single Gene Testing	IWK Clinical Genomics Laboratory: BTD	GENETICS Only
Crigler Najjar Syndrome Type I & II	UGT1A1 Single Gene Testing	IWK Clinical Genomics Laboratory: UGT1A1	GENETICS Only
Carnitine Deficiency (Systemic Primary)	SLC22A5 Single Gene Testing	IWK Clinical Genomics Laboratory: SLC22A5	GENETICS Only
Dihydropyrimidine Dehydrogenase Deficiency	DPYD Single Gene Testing	IWK Clinical Genomics Laboratory: DPYD via NGS (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 Testing	IWK Clinical Genomics Laboratory: GLA	Diagnosis: GENETICS, Internal Medicine, Nephrology, Neurology
Fatty Acid Oxidation Syndromes	Fatty Acid Oxidation Syndrome Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Galactosemia	GALT Single Gene Testing	IWK Clinical Genomics Laboratory: GALT	GENETICS Only
Gaucher Disease	GBA Single Gene Testing	IWK Clinical Genomics Laboratory: GBA	GENETICS Only
Glucose Transporter Type I Deficiency Syndrome	SLC2A1 Single Gene Testing	IWK Clinical Genomics Laboratory: SLC2A1	GENETICS Only
Glutaric Acidemia Type I	*GCDH* Single Gene Test	If STAT: Prevention Genetics (Please check lab details for gene content) If Routine: Blueprint Genetics (Please check lab details for gene content)	GENETICS Only

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Glutaric Acidemia Type II	Glutaric Acidemia Type II Panel	Prevention Genetics: Please check lab details for gene content	GENETICS Only
Glycogen Storage Disorders (GSD)	Glycogen Storage Disorder Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Hereditary Acrodermatitis Enteropathica	Hereditary Acrodermatitis Enteropathica Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Hyperammonemia	Hyperammonemia and Urea Cycle Disorder Panel	If STAT: Prevention Genetics (Please check lab details for gene content) If Routine: Blueprint Genetics (Please check lab details for gene content)	GENETICS Only
Hypoglycemia	Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Hypoglycemia MMGS Custom Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Leukodystrophy	Leukodystrophy and Leukoencephalopathy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Lysosomal Storage Disorders	Lysosomal Storage Disorders and Mucopolysaccharidosis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Maple Syrup Urine Disease	Maple Syrup Urine Disease Panel	Prevention Genetics: Please check lab details for gene content	GENETICS Only
Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD)	ACADM Single Gene Testing	IWK Clinical Genomics Laboratory: ACADM	GENETICS Only
Metabolic Myopathy and Rhabdomyolysis	Metabolic Myopathy and Rhabdomyolysis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Mitochondrial Diseases	Mitochondrial Genome Test	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Mitochondrial DNA Depletion Syndrome Panel	Blueprint/LHSC: Please check lab details for gene content	GENETICS Only

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Mucopolysaccharidosis Type I (MPSI)/Hurler Syndrome	IDUA Single Gene Testing	IWK Clinical Genomics Laboratory: IDUA	GENETICS Only
Niemann-Pick Disease Type C	NPC1 and NPC2 Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: NPC1-NPC2	GENETICS Only
Phenylketonuria (PKU)	PAH Single Gene Testing	IWK Clinical Genomics Laboratory: PAH	Diagnosis: GENETICS, New Brunswick PKU Program - NB PKU
Pompe Disease	*GAA* Single Gene Test	Blueprint Genetics: Please check lab details	Diagnosis: GENETICS, Neurology
Primary Hyperoxaluria	Primary Hyperoxaluria Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Nephrology
Tyrosinemia	*FAH* Single Gene Test	If STAT: Prevention Genetics (Please check lab details) If Routine: Blueprint Genetics (Please check lab details)	GENETICS Only
	Tyrosinemia Panel	If STAT: Prevention Genetics (Please check lab details for gene content) If Routine: Blueprint Genetics (Please check lab details for gene content)	GENETICS Only
Very Long-Chain acyl-CoA Dehydrogenase Deficiency (VLCAD)	ACADVL Single Gene Test	IWK Clinical Genomics Laboratory: ACADVL	GENETICS Only

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NEUROLOGY			
Amyotrophic Lateral Sclerosis (ALS)	SOD1 and C9Orf72 Analysis	IWK Clinical Genomics Laboratory: SOD1 whole gene sequencing, c9orf72 repeat expansion fPCR	Diagnosis: GENETICS, Neurology, Physical Medicine and Rehabilitation (Physiatry)
	Amyotrophic Lateral Sclerosis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only *In-house testing (*SOD1* and *C9orf72*) must be performed first, if appropriate
Ataxia	Ataxia Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS (c.6594delT & c.5254C>T only	IWK Clinical Genomics Laboratory: SACS (c.6594delT & c.5254C>T only	GENETICS Only
Brain Malformations	Cerebral Cavernous Malformation Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Lissencephaly Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
	Comprehensive Brain Malformation Panel	Prevention Genetics: Please check lab details for gene content	GENETICS Only
Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)	NOTCH3 Single Gene Testing	IWK Clinical Genomics Laboratory: NOTCH3	Diagnosis: GENETICS, Neurology
Charcot Marie Tooth	Charcot Marie Tooth 1A (CMT1A) and Hereditary Neuropathy with Pressure Palsies (HNPP): PMP22 dosage	IWK Clinical Genomics Laboratory: PMP22-MLPA	Diagnosis: None
	Charcot-Marie-Tooth Neuropathy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology *Consider In-house testing first

Congenital Myasthenic Syndrome	Congenital Myasthenic Syndromes Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Dementia	Dementia Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Dentatorubral-Pallidoluysian Atrophy (DRLPA)	Dentatorubral-Pallidoluysian Atrophy (DRPLA) via the ATN1 CAG Repeat	Prevention Genetics: Please check lab details	Diagnosis: GENETICS, Neurology
Duchenne Muscular Dystrophy/Becker Muscular Dystrophy	DMD Single Gene Testing	IWK Clinical Genomics Laboratory: DMD	Diagnosis: GENETICS, Neurology, Pediatrics - Pediatrics can order only for symptomatic young males (<13yo) *Must meet criteria for testing: Diagnostic/Symptomatic: -All symptomatic male requests (regardless of age) need to include: CK levels and pertinent Clinical Information -All symptomatic female requests need to be seen by Neurology or Genetics for consideration of testing. -Test requests need to include: CK levels and pertinent Clinical Information Carrier Testing -For all carrier testing requests, the patient needs to be seen by Genetics for consideration of testing.
Dystonia	Dystonia Panel	IWK Clinical Genomics Laboratory: ADCY5-AFG3L2-ANO3-ARSA-ATP1A2-ATP1A3-ATP7B-BCAP31-CACNA1A-CACNA1B-CP-CSF1R-CYP27A1-DCAF17-DDC-DLAT-DNAJC12-ECHS1-FA2H-FITM2-FTL-GCH1-GNAL-HPCA-KCNMA1-KCTD17-MYORG-KMT2B-MECR-NKX2-1-PANK2-PDE10A-PDE2A- PDGFB-PDGFRB-PINK1-PLA2G6-PNKD-PRKRA-PRRT2-PTS-SGCE-SLC20A2-SLC2A1-SLC30A10-SLC39A14-SPR-TAF1-TH-THAP1-TIMM8A-	Diagnosis: GENETICS, Neurology

		TOR1A-TUBB4A-UBTF-VAC14-VPS13A-VPS16-WDR45-XPR1	
Epilepsy	Comprehensive Epilepsy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Episodic Ataxia Type 2 (EA2) and Spinocerebellar Ataxia Type 6 (SCA6)	CACNA1A Single Gene Testing	IWK Clinical Genomics Laboratory: CACNA1A	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
Emery-Dreifuss Muscular Dystrophy	Emery-Dreifuss Muscular Dystrophy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Facioscapulohumeral Muscular Dystrophy (FSHD)	Facioscapulohumeral Muscular Dystrophy (FSHD)	CHEO Genetics Diagnostic Laboratory: Please check lab details	Diagnosis: GENETICS, Neurology
Familial Hemiplegic Migraine (FHM)	CACNA1A and SCN1A Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: ATP1A2-CACNA1A-SCN1A	Diagnosis: GENETICS, Neurology
	Migraine Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology *Consider In-house testing first
Friedreich Ataxia (FRDA)	Friedreich Ataxia (FRDA) testing	North York General Hospital: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Hereditary Spastic Paraplegia (HSP)	Hereditary Spastic Paraplegia Panel	IWK Clinical Genomics Laboratory: ABCD1-ABHD16A-ALDH18A1-ALS2-AP4B1-AP4E1-AP4M1-AP4S1-AP5Z1-ATL1-ATP13A2-B4GALNT1-BSCL2-C19ORF12-CAPN1-CYP27A1-CYP2U1-CYP7B1-DARS1-DDHD1-DDHD2-ENTPD1-ERLIN1-ERLIN2-FA2H-GBA2-HEXA-HSPD1-IBA57-KIF1A-KIF1C-KIF5A-L1CAM-MAG-MTRFR-NIPA1-NKX6-2-NT5C2-PCYT2-PGAP1-PLP1-PNPLA6-REEP1-RTN2-SACS-SETX-SLC16A2-SLC33A1-SPART-SPAST-SPG11-SPG21-	Diagnosis: GENETICS, Neurology, Physical Medicine and Rehabilitation (Physiatry)

		SPG7-TECPR2-TUBB4A-UBAP1-VPS37A-WASHC5-ZFYVE26	
Huntington Disease (HD)	HD via Trinucleotide Repeat Expansion fPCR Kit	IWK Clinical Genomics Laboratory: HD-fPCR	Diagnosis: GENETICS, Neurology, Psychiatry
Microcephaly and Pontocerebellar Hypoplasia	Microcephaly and Pontocerebellar Hypoplasia Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Muscular Dystrophy/Myopathy	Comprehensive Muscular Dystrophy/Myopathy Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Myotonic Dystrophy Type 1 (DM1)	DM1 via Trinucleotide Repeat Expansion fPCR Kit	IWK Clinical Genomics Laboratory: DM1-fPCR	Diagnosis: GENETICS, Neurology, Pediatrics
Myotonic Dystrophy Type 2 (DM2)	Myotonic Dystrophy Type II (DM2) via PCR and repeat-primed PCRs	CHEO Genetics Diagnostic Laboratory: Please check lab details	Diagnosis: GENETICS, Neurology
Neuronal Migration Disorders	Neuronal Migration Disorder Panel	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Oculopharyngeal Muscular Dystrophy (OPMD)	Oculopharyngeal Muscular Dystrophy via the PABPN1 (GCN) Repeat Expansion	Prevention Genetics: Please check lab details	Diagnosis: GENETICS, Neurology
Parkinson Disease/Parkinsonism	Parkinson Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Paroxysmal Dyskinesia (PD)	PD Panel	IWK Clinical Genomics Laboratory: ADCY5-CACNA1A-DEPDC5-KCNA1-KCNMA1-PNKD-PRRT2-SLC2A1-SCN8A	Diagnosis: GENETICS, Neurology
Periodic Paralysis	Periodic Paralysis Panel	IWK Clinical Genomics Laboratory: CACNA1S-CLCN1-KCNJ2-SCN4A	Diagnosis: GENETICS, Neurology
Polymicrogyria	Polymicrogyria Panel	IWK Clinical Genomics Laboratory: ADGRG1-AKT3-FH-GPSM2-KIF1BP-LAMC3-NDE1-NSDHL-OCLN-PI4KA-RAB18-SNAP29-TBC1D20-	Diagnosis: GENETICS, Neurology

		TUBA8-TUBB2A-TUBB2B-TUBB3-WDR62	
SCN1A Seizure Disorder	SCN1A Single Gene Testing	IWK Clinical Genomics Laboratory: SCN1A	Diagnosis: GENETICS, Neurology
SCN9A Neuropathic Pain Syndromes	SCN9A Single Gene Testing	IWK Clinical Genomics Laboratory: SCN9A	Diagnosis: GENETICS, Neurology, Rheumatology
SGCE Myoclonus Dystonia	SGCE Single Gene Testing	IWK Clinical Genomics Laboratory: SGCE	Diagnosis: GENETICS, Neurology
Spinal and Bulbar Muscular Atrophy (SBMA)	X-linked Spinal and Bulbar Muscular Atrophy (Kennedy Disease) via the AR Gene CAG Repeat Expansion	Prevention Genetics: Please check lab details	Diagnosis: GENETICS, Neurology
Spinal Muscular Atrophy (SMA)	SMN Gene Dosage via MLPA	IWK Clinical Genomics Laboratory: SMA-MLPA	Diagnosis: GENETICS, Neurology, Pediatrics
Spinocerebellar Ataxia (SCA)	Spinocerebellar Ataxia (SCA) Panel	North York General Hospital: Please check lab details for gene content	Diagnosis: GENETICS, Neurology

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ONCOLOGY			
BRCA1, BRCA2	BRCA1, BRCA2Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: BRCA1-BRCA2	Diagnosis: GENETICS, Oncology* *For treatment purposes in breast cancer patients
Colorectal Cancer	Colorectal Cancer Panel	IWK Clinical Genomics Laboratory: APC-AXIN2-BMPR1A-EPCAM (3' del only)-GREM1-MBD4-MLH1-MLH3-MSH2-MSH3-MSH6-MUTYH-NTHL1-PMS2-POLD1-POLE-PTEN-RNF43-RPS20-SMAD4-STK11-TP53	GENETICS Only
Endometrial Cancer	Endometrial Cancer Panel	IWK Clinical Genomics Laboratory: EPCAM (3' del only)-MLH1-MSH2-MSH6-PMS2-POLD1-POLE-PTEN	GENETICS Only
Hereditary Breast and Ovarian Cancer (HBOC)	HBOC Panel	IWK Clinical Genomics Laboratory: 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	IWK Clinical Genomics Laboratory: 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	IWK Clinical Genomics Laboratory: 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	IWK Clinical Genomics Laboratory: BRCA1-BRCA2-BRIP1-EPCAM (3' del only)-MLH1-MSH2-MSH6-PMS2-RAD51C-RAD51D	GENETICS Only

Lymphoid Neoplasms	Lymphoid Neoplasms Panel	IWK Clinical Genomics Laboratory: ACD-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	GENETICS Only
Lynch Syndrome	Lynch Syndrome Panel	IWK Clinical Genomics Laboratory: EPCAM (3’ del only)-MLH1-MSH2-MSH6-PMS2	GENETICS Only
Melanoma	Melanoma Panel	IWK Clinical Genomics Laboratory: BAP1-BRCA2-CDK4-CDKN2A-PTEN-TP53	GENETICS Only
Multiple Endocrine Neoplasia Type 1 (MEN1)	MEN1 Single Gene Testing	IWK Clinical Genomics Laboratory: MEN1	GENETICS Only
Multiple Endocrine Neoplasia Type 2 (MEN2)	MEN2 Single Gene Testing	IWK Clinical Genomics Laboratory: RET	GENETICS Only
MUTYH Associated Polyposis	MUTYH Single Gene Testing	IWK Clinical Genomics Laboratory: MUTYH	GENETICS Only
Myeloid Malignancy	Myeloid Malignancy Panel	IWK Clinical Genomics Laboratory: ANKRD26 (including 5'UTR)-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
Neuroendocrine Tumors	Neuroendocrine Tumor Panel	IWK Clinical Genomics Laboratory: CDKN1B-MEN1-NF1-TSC1-TSC2-VHL	GENETICS Only
Nevoid Basal Cell Carcinoma Syndrome	PTCH1 and SUFU Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: PTCH1-SUFU	GENETICS Only
Pancreatic Cancer	Pancreatic Cancer Panel	IWK Clinical Genomics Laboratory: ATM-ATM c.5763-1050A>G-BRCA1-BRCA2-CDKN2A-EPCAM (3’ del only)-MLH1-MSH2-MSH6-PALB2-PMS2-STK11	GENETICS Only
Pheochromocytoma &	Pheochromocytoma &	IWK Clinical Genomics Laboratory:	GENETICS Only

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Paraganglioma	Paraganglioma Panel	FH-MAX-NF1-RET-SDHA-SDHB-SDHC-SDHD-SDHAF2-TMEM127-VHL	
Polyposis	Polyposis Panel	IWK Clinical Genomics Laboratory: APC-BMPR1A-MUTYH-PTEN-SMAD4-STK11	GENETICS Only
Prostate Cancer	Prostate Cancer Panel	IWK Clinical Genomics Laboratory: ATM-ATM c.5763-1050A>G-BRCA1-BRCA2-CHEK2-EPCAM (3' del only)-HOXB13-MLH1-MSH2-MSH6-PALB2-PMS2-RAD51D-TP53	Diagnosis: Genetics, Mainstreamed physicians
Renal Cell Carcinoma Panel	Renal Cell Carcinoma Panel	IWK Clinical Genomics Laboratory: BAP1-FH-FLCN-MET-PTEN-SDHB-SDHC-SDHD-TMEM127-TSC1-TSC2-VHL	GENETICS Only
Retinoblastoma	RB1 Single Gene Analysis	IWK Clinical Genomics Laboratory: RB1	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	IWK Clinical Genomics Laboratory: TSC1-TSC2	Diagnosis: Dermatology, GENETICS, Neurology
Von Hippel-Lindau (VHL) Syndrome	VHL Single Gene Testing	IWK Clinical Genomics Laboratory: VHL	GENETICS Only

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OPHTHALMOLOGY			
Achromptasia	Achromptasia Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Aniridia	Aniridia Panel	IWK Clinical Genomics Laboratory: PAX6 (SNV and dosage)-WT1 (dosage only)	GENETICS Only
Cone Rod Dystrophy	Cone Rod Dystrophy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Ectopia Lentis	Ectopia Lentis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Leber Congenital Amaurosis	Leber Congenital Amaurosis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Macular Dystrophy	Macular Dystrophy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Optic Atrophy	Optic Atrophy	Blueprint Genetics: Please check lab details for gene content	Diagnosis: GENETICS, Neuro-Ophthalmology
Retinal Dystrophy	Retinal Dystrophy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Retinitis Pigmentosa (RP)	Retinitis Pigmentosa Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Septo-Optic Dysplasia	Septo-Optic Dysplasia Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Stargardt/Macular Dystrophy	Stargardt/Macular Dystrophy Panel	IWK Clinical Genomics Laboratory: ABCA4-BEST1-CDH3-DRAM2-EFEMP1-ELOVL4-IMPG1-IMPG2-PROM1-PRPH2-RP1L1-TIMP3-TTLL5	GENETICS Only
Vitreoretinopathy	Vitreoretinopathy Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only

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SKELETAL			
Achondroplasia (common mutations only)	FGFR3 c.1138G>A and c.1138G>G via Sanger sequencing	IWK Clinical Genomics Laboratory: 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	IWK Clinical Genomics Laboratory: FGFR2 c.775C>G and c.758C>G via Sanger sequencing	Restrictions: None
Arthrogryposis	Arthrogryposes Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Cleidocranial Dysplasia (CCD) Spectrum Disorder	RUNX2 Single Gene Testing	IWK Clinical Genomics Laboratory: RUNX2	GENETICS Only
Craniosynostosis	Craniosynostosis Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Limb Malformations	Limb Malformations Panel	Blueprint Genetics: Please check lab details for gene content	GENETICS Only
Osteogenesis Imperfecta (OI)	Osteogenesis Imperfecta Panel	IWK Clinical Genomics Laboratory: ALPL-ANO5-B3GALT6-B3GAT3-BMP1-COL1A1-COL1A2-COPB2-CREB3L1-CRTAP-FKBP10-GORAB-IFITM5-KDELR2-LRP5-MBTPS2-MESD-P3H1-P4HB-PLOD2-PLS3-PPIB-SEC24D-SERPINF1-SERPINH1-SGMS2-SP7-SPARC-TAPT1-TENT5A-TMEM38B-TNFRSF11B-WNT1-XYLT2	Diagnosis: GENETICS, IWK Suspected Trauma and Abuse Response Team (START)
Skeletal Dysplasia	Comprehensive Growth Disorders/Skeletal Dysplasias and Disorders Panel (or sub-panel)	Blueprint Genetics: Please check lab details for gene content	GENETICS Only