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Ordering Restrictions: Predictive testing, as well as carrier testing (unless excepted) 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. Tests with no restrictions can be ordered by the patient's primary care provider including nurse practitioners (NPs).

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, Provincial Medical Examiner's Service, Cardiology
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	Arrhythmogenic Right Ventricular Cardiomyopathy Panel	IWK Clinical Genomics Laboratory: CDH2-DSC2-DSG2-DSP-JUP-PKP2-RYR2-TMEM43	Diagnosis: GENETICS, Provincial Medical Examiner's Service, Cardiology

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Brugada Syndrome	Brugada Panel	IWK Clinical Genomics Laboratory: SCN5A	Diagnosis: GENETICS, Provincial Medical Examiner's Service, Cardiologists
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, Provincial Medical Examiner's Service, Cardiology
Catecholaminergic Polymorphic Ventricular tachycardia (CPVT)	Catecholaminergic Polymorphic Ventricular Tachycardia Panel	IWK Clinical Genomics Laboratory: CALM1-CASQ2-RYR2-TECRL-TRDN	Diagnosis: GENETICS, Provincial Medical Examiner's Service, Cardiology
Congenital Heart Disease (CHD)	Congenital Heart Disease Panel	IWK Clinical Genomics Laboratory: ABL1-ACTB-ACTG1-ACVR2B-ADAMTS10-AFF4-ARHGAP31-ARID1A-ARID1B-BCOR-BRAF-CBL-CDK13-CFAP53-CFC1-CHD4-CHD7-CITED2-CREBBP-CRELD1-DHCR7-DLL4-DOCK6-EFTUD2-EHMT1-ELN-EOGT-EP300-EVC-EVC2-FLNA-FLT4-FOXF1-FOXH1-FOXP1-GATA4-GATA5-GATA6-GDF1-GJA1-GPC3-HAND1-HAND2-HNRNPK-HOXA1-HRAS-JAG1-KDM6A-KMT2D-KRAS-KYNU-LEFTY2-LZTR1-MAP2K2-MED12-MED13L-MEGF8-MEIS2-MMP21-MNS1-MYRF-NAA15-NEK8-NEXN-NIPBL-NKX2-5-NKX2-6-NODAL-NONO-NOTCH1-NOTCH2-NR2F2-NRAS-NSD1-NUP188-PITX2-PKD1L1-PLD1-PPP1CB-PRDM6-PRKD1-PTPN11-PUF60-RAB23-RAF1-RBFOX2-RBM10-RERE-RIT1-SHOC2-SMAD6-SMARCB1-SMC1A-SMC3-SOS1-SOS2-STAG2-STRA6-TAB2-TBX1-TBX20-TBX5-TFAP2B-TGDS-TLL1-TMEM94-TWIST1-ZEB2-ZFPM2-ZIC3	GENETICS Only
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-	Diagnosis: GENETICS, Provincial Medical Examiner's Service, Cardiology

		TTN-VCL	
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, Provincial Medical Examiner's Service, Cardiology
Inherited dyslipidemia	Hyperlipidemia Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Provincial 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, Provincial Medical Examiner's Service, Cardiology
Long QT Syndrome (LQTS)	Long QT Panel	IWK Clinical Genomics Laboratory: ANK2-CALM1-CALM2-CALM3-CACNA1C-CAV3-KCNE1-KCNE2-KCNH2-KCNJ2-KCNQ1-SCN5A	Diagnosis: GENETICS, Provincial Medical Examiner's Service, Cardiology
Transthyretin Amyloidosis	*TTR* Single Gene Testing	IWK Clinical Genomics Laboratory: TTR	Restrictions: None

CONNECTIVE TISSUE			
Alkaptonuria	Alkaptonuria Panel (HGD Single Gene Testing)	IWK Clinical Genomics Laboratory: HGD	GENETICS Only
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, Provincial Medical Examiner's Service, Cardiology

	Aorta Extend Panel	IWK Clinical Genomics Laboratory: ABL1-ACTA2-ADAMTS6-ADAMTSL4-ARIH1-ASPH-BGN-CBS-COL1A1-COL1A2-COL3A1-COL5A1-COL5A2-EFEMP2-ELN-FBLN5-FBN1-FBN2-FKBP14-FLNA-FOXE3-HCN4-IPO8-LOX-MAT2A-MFAP5-MYH11-MYLK-NOTCH1-PLOD1-PMEPA1-PRKG1-SECISBP2-SKI-SLC2A10-SMAD2-SMAD3-SMAD4-SMAD6-TGFB2-TGFB3-TGFBR1-TGFBR2-YY1AP1 (NM_001198903:c.1079C>T only)	GENETICS Only
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 (HDCT)	Hereditary Disorder of Connective Tissue Panel	IWK Clinical Genomics Laboratory: ABL1-ACTA2-ADAMTS2-ADAMTS6-ADAMTSL2-ADAMTSL4-AEBP1-ALDH18A1-ARIH1-ASPH-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-FOXE3-GORAB-HCN4-IPO8-LOX-LTBP4-MAT2A-MED12-MFAP5-MYH11-MYLK-NOTCH1-	GENETICS Only

		PLOD1-PMEPA1-PRDM5-PRKG1-PYCR1-RIN2-SECISBP2-SKI-SLC2A10-SLC39A13-SMAD2-SMAD3-SMAD4-SMAD6-TAB2-TGFB2-TGFB3-TGFBR1-TGFBR2-TNXB-YY1AP1 (NM_139119:c.1079C>T only)-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
Vascular Malformations	Vascular Malformations Panel	IWK Clinical Genomics Laboratory: ACVRL1-ADAMTS13-AKT1-ALAS2-ATM-CCBE1-CCM2-ENG-EPHB4-F12-FECH-FLT4-FOXC2-GDF2-GLMN-KRIT1-PDCD10-PIK3CA-PIK3R2-PTEN-RASA1-SCN9A-SMAD4-SOX18-STAMBP-STING1-TEK	GENETICS Only

CONGENITAL ANOMALIES			
Congenital Diaphragmatic Hernia (CDH)	Congenital Diaphragmatic Hernia (CDH) Panel	IWK Clinical Genomics Laboratory: EFEMP2-EFNB1-FBN1-GATA6-GPC3-HCCS-KMT2D-LRP2-LTBP4-MYRF-NIPBL-NR2F2-PBX1-PIGN-PLS3-PORCN-RAD21-RARB-RLIM-SPECC1L-STRA6-WT1-ZFPM2	GENETICS Only

DERMATOLOGY			
Adams-Oliver Syndrome	Adams-Oliver Syndrome Panel	IWK Clinical Genomics Laboratory: ARHGAP31-DLL4-DOCK6-EOGT-KCTD1-NOTCH1-RBPJ-UBR1	GENETICS Only
Albinism	Albinism Panel	IWK Clinical Genomics Laboratory: AP3B1-BLOC1S3-BLOC1S5-BLOC1S6-DCT-DTNBP1-	Diagnosis: Dermatology, GENETICS, Ophthalmology

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		FRMD7-GPR143-HPS1-HPS3-HPS4-HPS5-HPS6-LRMDA-LYST-MLPH-MYO5A-OCA2-RAB27A-SLC24A5-SLC38A8-SLC45A2-TYR-TYRP1	
Amelogenesis and Dentinogenesis Imperfecta	Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Epidermolysis Bullosa	Epidermolysis Bullosa Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Dermatology, GENETICS
Ichthyosis	Ichthyosis Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only

ENDOCRINOLOGY			
Bartter Syndrome	Bartter Syndrome Panel	IWK Clinical Genomics Laboratory: BSND-CLCNKA-CLCNKB-KCNJ1-MAGED2-SLC12A1	Diagnosis: Endocrinology, GENETICS, Nephrology
Congenital Adrenal Hyperplasia (CAH)	Congenital Adrenal Hyperplasia Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS

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Congenital Hyperinsulinism	Congenital Hyperinsulinism Panel	IWK Clinical Genomics Laboratory: ABCC8-GCK-GLUD1-HADH-HNF1A-HNF4A-KCNJ11-SLC16A1-UCP2	Diagnosis: Endocrinology, GENETICS
Disorders of Sex Development (DSD)	Abnormal Genitalia/Disorders of Sexual Development Panel	IWK Clinical Genomics Laboratory: AMH-AMHR2-ANOS1-AR-ARX-ATRX-CBX2-CCNQ-CDKN1C-CHD4-CHD7-CYB5A-CYP11A1-CYP11B1-CYP17A1-CYP19A1-DHCR7-DHH-DHX37-DYNC2H1-FEZF1-FGF8-FGFR1-FRAS1-FREM2-GATA4-HSD17B3-HSD3B2-LHCGR-MAMLD1-MAP3K1-MKKS-NR0B1-NR2F2-NR5A1-POR-PPP1R12A-PROK2-PROKR2-ROR2-RSPO1-SAMD9-SOX3-SOX10-SRD5A2-SRY-STAR-TOE1-TSPYL1-WNT4-WT1-WWOX-ZFPM2	Diagnosis: GENETICS
Familial Hyperaldosteronism	Primary Hyperaldosteronism NGS Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS
	Pseudohypoaldosteronism Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS, Internal Medicine, Nephrology
Hypothyroidism and Thyroid Resistance	Hyperparathyroidism Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS
	Hypothyroidism and Resistance to Thyroid Hormone Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS
Hypogonadotropic Hypogonadism/Kallmann Syndrome	Kallmann Syndrome Panel	IWK Clinical Genomics Laboratory: ANOS1-CHD7-FEZF1-FGF8-FGFR1-GNRH1-GNRHR-IL17RD-KISS1R-LHB-PROK2-PROKR2-SEMA3A-SOX10-TAC3-TACR3-WDR11	Diagnosis: Endocrinology, GENETICS

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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS, Nephrology
Lipodystrophy	Congenital and Familial Lipodystrophy Panel	IWK Clinical Genomics Laboratory: AGPAT2-AKT2-BANF1-BSCL2-CAV1-CAVIN1-CIDEC-FBN1-KCNJ6-LIPE-LMNA-PCYT1A-PLIN1-POLD1-PPARG-ZMPSTE24	GENETICS Only
Maturity 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, Pediatrics GENETICS
Monogenic Obesity	Monogenic Obesity Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Pituitary Adenoma (PITAD)	Pituitary Adenoma (PITAD) Panel	IWK Clinical Genomics Laboratory: AIP, MEN1, CDKN1B	Diagnosis: Endocrinology, GENETICS
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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Pediatric ENT
Hereditary Hearing Loss	Hereditary Hearing Loss Panel	IWK Clinical Genomics Laboratory: P461-MLPA (STRC gene) + NGS Panel sequencing: ACTG1-ADGRV1-BSND-CABP2-CDC14A-CDH23-	Diagnosis: Audiology, ENT, Medical Genetics, Pediatrics *Testing includes P461-MLPA (STRC gene) & NGS multi-

		CEACAM16-CHD7-CIB2-CLDN14-COCH-COL11A2-CRYM-DIAPH1-DSPP-ESPN-ESRRB-EYA1-EYA4-GIPC3-GJB2-GJB6-GPSM2-GRHL2-GRXCR1-GSDME-HGF-ILDR1-KCNE1-KCNQ1-KCNQ4-LHFPL5-LOXHD1-LRTOMT-MARVELD2-MSRB3-MYH14-MYH9-MYO15A-MYO3A-MYO6-MYO7A-OSBPL2-OTOA-OTOF-OTOG-OTOGL-P2RX2-PCDH15-PJVK-PNPT1-POU3F4-POU4F3-PRPS1-PTPRQ-RDX-S1PR2-SERPINB6-SIX1-SLC17A8-SLC26A4-SLITRK6-SMPX-STRC-SYNE4-TBC1D24-TECTA-TMC1-TMIE-TMPRSS3-TPRN-TRIOBP-USH2A-WFS1-WHRN	gene panel
Pendred Syndrome	Pendred Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Endocrinology, GENETICS, Pediatric ENT
Usher Syndrome (Type 1, Type 2)	Usher Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	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
Adrenoleukodystrophy	*ABCD1* Single Gene Testing	IWK Clinical Genomics Laboratory: ABCD1	Diagnosis: GENETICS, Neurology
Alagille Syndrome	*JAG1* Single Gene Testing	IWK Clinical Genomics Laboratory: JAG1	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 Screening	Ashkenazi Jewish Carrier Screening Panel	IWK Clinical Genomics Laboratory: ASPA-BLM-ELP1-FANCC-HEXA-MCOLN1-SMPD1	GENETICS Only
Bardet-Biedl Syndrome (BBS)	Bardet-Biedl Syndrome Panel	IWK Clinical Genomics Laboratory: ALMS1-ARL6-BBIP1-BBS1-BBS10-BBS12-BBS2-BBS4-BBS5- BBS7-BBS9-CFAP418-CEP19-CEP290-IFT172-IFT27-IFT74-LZTFL1-MKKS-MKS1-PNPLA6-SCAPER-SCLT1-SDCCAG8-TTC8-WDPCP	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	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Cornelia de Lange Syndrome	Cornelia de Lange Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
	Ciliopathy Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only

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Ciliopathies, Heterotaxy/Situs Inversus and Primary Ciliary Dyskinesia (PCD)	Heterotaxy and Situs Inversus Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
	Prenatal Heterotaxy Panel	IWK Clinical Genomics Laboratory: ACVR2B-ANKS6-CCDC103-CCDC39-CCDC40-CFAP298-CFAP300-CFAP53-DNAAF1-DNAAF11-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	GENETICS Only
	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 *A score of ≥5 on the Primary Ciliary Dyskinesia Rule (PICADAR) should be provided
Cystic Fibrosis	*CFTR* Single Gene Testing	IWK Clinical Genomics Laboratory: CFTR	Diagnosis: None Carrier Screening 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 Ovarian Failure	*FMR1* Analysis (Triplet-primed PCR)	IWK Clinical Genomics Laboratory: FMR1	Diagnosis: None
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	Diagnosis: Endocrinology (if the phenotype is pseudohypoparathyroidism), GENETICS -Methylation analysis: If the patient has findings suggestive of pseudohypoparathyroidism Ib (PHP-1b), -Sequence analysis: If the patient has findings suggestive of another GNAS phenotype
Hereditary Multiple Osteochondromas	Hereditary Multiple Osteochondromas Panel	IWK Clinical Genomics Laboratory: EXT1-EXT2	GENETICS Only
Interstitial Lung Disease	Interstitial Lung Disease Panel	IWK Clinical Genomics Laboratory: ABCA3-ACD-CSF2RB-CTC1-DKC1-FAM111B-HPS1-HPS4-ITGA3-NAF1-NF1-NHP2-NKX2-1-NOP10-PARN-POT1-RPA1-RTEL1-SFTPA1-SFTPA2-SFTPB-SFTPC-SLC34A2-SLC7A7-SMPD1-STING1-STN1-TERC-TERT-TINF2-WRAP53-ZCCHC8	Diagnosis: Hematology, Hepatology, GENETICS, Respirology
Joubert Syndrome	Joubert Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Kabuki Syndrome	Kabuki Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	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: 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
PIK3CA-Related Disorders	PIK3CA Single Gene Testing	IWK Clinical Genomics Laboratory: PIK3CA	GENETICS Only *Requires consultation with Lab GC / Lab Scientist prior to ordering
Prader Willi Syndrome (PWS)	15q11 Methylation and Dosage MLPA	IWK Clinical Genomics Laboratory: PWS-MLPA	Restrictions: None
PTEN-related Macrocephaly/Autism Spectrum Disorder	*PTEN* Single Gene Testing	IWK Clinical Genomics Laboratory: PTEN	GENETICS Only
Pulmonary Arterial Hypertension	Pulmonary Arterial Hypertension Panel	IWK Clinical Genomics Laboratory: ABCC8-ACVRL1-AQP1-ATP13A3-BMPR2-CAV1-	Diagnosis: GENETICS, Cardiology, Respirology

		EIF2AK4-ENG-FOXF1-GDF2-KCNK3-KDR-KLF2-SARS2-SMAD9-SOX17-TBX4	
Pulmonary Fibrosis	Telomere Length Testing	RepeatDX: Please check lab details for gene content	Diagnosis: GENETICS, Respirology
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)
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
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, Hepatology, Neurology

GI/NEPHROLOGY			
Alport Syndrome	Alport Syndrome Panel	IWK Clinical Genomics Laboratory: CD151-COL4A3-COL4A4-COL4A5-COL4A6-MYH9	Diagnosis: GENETICS, Nephrology
APOA4 Nephropathy	APOA4 Single Gene Testing	IWK Clinical Genomics Laboratory: APOA4 (c.196 C>G only)	Diagnosis: GENETICS, Nephrology *APOA4 variant belongs to a local kindred and associated with adult onset renal failure

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Cholestasis	Cholestasis Panel	IWK Clinical Genomics Laboratory: ABCB11-ABCB4-ABCC2-ADK-AKR1D1-ALDOB-AMACR-ATP7B-ATP8B1-BAAT-BCS1L-CFTR-CLDN1-COG7-CYP27A1-CYP7B1-DCDC2-DGUOK-FAH-FOCAD-GALE-GALM-GALT-GBA1-HADHA-HNF1B-HSD3B7-JAG1-KIF12-LIPA-MKS1-MPI-MPV17-MVK-MYO5B-NBAS-NOTCH2-NPC1-NPC2-NPHP1-NPHP3-NPHP4-NR1H4-PEX1-PEX10-PEX12-PEX2-PEX26-PEX5-PEX6-PKHD1-POLG-PSKH1-SCYL1-SERPINA1-SLC25A13-SMPD1-TALDO1-TJP2-TMEM216-TMEM67-TRMU-UGT1A1-UNC45A-USP53-VIPAS39-VPS33B-YARS1-ZFYVE19	GENETICS Only
Congenital Abnormalities of the Kidney and Urinary Tract (CAKUT)	Congenital Abnormalities of the Kidney and Urinary Tract (CAKUT) Panel	IWK Clinical Genomics Laboratory: ACE-ACTG2-AGT-AGTR1-ALG8-ALG9-ANKS6-ANOS1-BMP4-BNC2-CCNQ-CENPF-CEP164-CEP290-CEP55-CEP83-CHD7-CHRM3-CHRNA3-COL4A1-CRB2-CTU2-DHCR7-DNAJB11-DSTYK-DZIP1L-EYA1-FANCA-FANCC-FANCG-FGF20-FRAS1-FREM1-FREM2-GANAB-GATA3-GFRA1-GLI3-GPC3-GREB1L-GRIP1-HAAO-HNF1B-HOXA13-HPSE2-HS2ST1-IFT172-INVS-ITGA8-JAG1-KDM6A-KMT2D-KYNU-LIFR-LRIG2-LRP4-MAPKBP1-MYOCD-NADSYN1-NEK8-NIPBL-NOTCH2-NPHP1-NPHP3-NPHP4-NRIP1-OFD1-PAX2-PBX1-PKD1-PKD2-PKHD1-PUF60-REN-RET-ROBO1-ROBO2-RPGRIP1L-SALL1-SALL4-SDCCAG8-SIX1-SIX5-SOX11-SOX17-STRA6-TBC1D1-TBX18-TFAP2A-TMEM260-TMEM67-TRAP1-TSC1-TSC2-TTC21B-WBP11-WDR19-WNT4-WT1-ZIC3-ZMYM2-ZNF423	GENETICS Only
Congenital Diarrhea	Congenital Diarrhea Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, GI
Hirschsprung Disease	Hirschsprung Disease Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, GI *Pathology report must be provided

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Nephrocalcinosis Nephrolithiasis	Nephrocalcinosis/Nephrolithiasis Panel	IWK Clinical Genomics Laboratory: ADCY10-AGXT-ALPL-APRT-ATP6V0A4-ATP6V1B1-ATP7B-BSND-CA2-CASR-CLCN5-CLCNKB-CLDN16-CLDN19-CYP24A1-FAM20A-FOXI1-GRHPR-HNF4A-HOGA1-HPRT1-KCNJ1-MAGED2-MOCOS-NHERF1-OCRL-PHEX-PRPS1-RRAGD-SLC12A1-SLC22A12-SLC26A1-SLC2A9-SLC34A1-SLC34A3-SLC3A1-SLC4A1-SLC7A9-STRADA-UMOD-VDR-VIPAS39-VPS33B-WDR72-XDH	Diagnosis: GENETICS, Nephrology
Nephrotic Syndrome	Nephrotic Syndrome Panel	IWK Clinical Genomics Laboratory: ACTN4-ANLN-APOL1-ARHGAP24-ARHGDIA-AVIL-CD2AP-CLCN5-COL4A3-COL4A4-COL4A5-COQ2-COQ6-COQ8B-CRB2-CUBN-DAAM2-DGKE-EMP2-FAN1-FN1-GLA-GON7-INF2-ITGA3-KANK1-KANK2-KIRREL1-LAGE3-LAMA5-LAMB2-LMX1B-LYZ-MAGI2-MYH9-MYO1E-NPHP1-NPHS1-NPHS2-NUP107-NUP133-NUP205-NUP85-NUP93-OSGEP-PAX2-PDSS2-PLCE1-PTPRO-SCARB2-SGPL1-SMARCAL1-TBC1D8B-TP53RK-TPRKB-TRIM8-TRPC6-TTC21B-TTR-WDR4-WDR73-WT1-YRDC	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	Diagnosis: GENETICS, Nephrology
Polycystic Liver Disease	Polycystic Liver Disease Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, GI
Renal Malformations	Renal Malformation Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only

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Renal Tubulopathies	Inherited Renal Tubulopathies	IWK Clinical Genomics Laboratory: AP2S1-AQP2-ATP1A1-ATP6V0A4-ATP6V1B1-AVPR2-BSND-CA2-CASR-CLCN5-CLCNKB-CLDN10-CLDN16-CLDN19-CNNM2-CTNS-CUL3-CYP24A1-EHHADH-FAH-GATM-GNA11-HNF1B-HNF4A-HSD11B2-KCNJ1-KCNJ10-KCNJ16-KLHL3-MAGED2-NDUFAF6-NR3C2-OCRL-REN-RMND1-RRAGD-SARS2-SCNN1A-SCNN1B-SCNN1G-SEC61A1-SLC12A1-SLC12A3-SLC22A12-SLC2A2-SLC2A9-SLC34A1-SLC34A3-SLC4A1-SLC4A4-SLC5A2-TRPM6-UMOD-VIPAS39-VPS33B-WDR72-WNK1-WNK4	Diagnosis: GENETICS, Nephrology
Renal Tubular Dysgenesis	Renal Tubular Dysgenesis Panel	IWK Clinical Genomics Laboratory: ACE-AGT-AGTR1-REN	Diagnosis: GENETICS, Pathology
Wilms Tumor	Wilms Tumor Panel	IWK Clinical Genomics Laboratory: *BLM-BUB1B-CDC73-CDKN1C-CHEK2-CTR9-DICER1-DIS3L2-GPC3-REST-TP53-TRIM28-TRIP13-WT1*	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-POT1-RAD51C-RBM8A-RPA1-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-	Diagnosis: GENETICS, Hematology, Immunology

		WRAP53-XRCC2-ZCCHC8	
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	Not currently available at the IWK CGL. Please contact the lab for options	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-IFNGR2-JAGN1-LAMTOR2-LYST-PGM3-RAB27A-RAC2-SBDS-SLC37A4-SMARCD2-SRP54-STK4-TAFAZZIN-TCN2-USB1-VPS13B-VPS45-WAS-WDR1-WIPF1	Diagnosis: GENETICS, Hematology, Immunology
Diamond-Blackfan Anemia	Diamond-Blackfan Anemia Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Dyskeratosis Congenita/Telomere Biology Disorder	Telomere Length Testing	RepeatDX: Please check lab details for gene content	Diagnosis: GENETICS, Hematology, Hematology-Oncology
	Dyskeratosis Congenita/Telomere Biology Disorder Panel	IWK Clinical Genomics Laboratory: ACD-CTC1-DCLRE1B-DKC1-NAF1-NHP2-NOP10-PARN-POT1-RPA1-RTEL1-STN1-TERC-TERT-TINF2-USB1-WRAP53-ZCCHC8	Diagnosis: GENETICS, Hematology, Hematology-Oncology
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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Hematology
Fibrinogen Disorders	Fibrinogen Disorders Panel	IWK Clinical Genomics Laboratory: FGA-FGB-FGG	Diagnosis: GENETICS, Hematology

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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 OR young girls with no symptoms and a positive family history (pre menarche). Note: ALL positive results in these cases need to be referred to genetics for further review.
Hereditary Erythrocytosis	Hereditary Erythrocytosis Panel	IWK Clinical Genomics Laboratory: BPGM-EGLN1-EPAS1-EPO-EPOR-JAK2	Diagnosis: GENETICS, Hematology
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
Hereditary Leukemia	Hereditary Leukemia Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: Dr. Trottier, GENETICS
Hermansky-Pudlak Syndrome	Hermansky-Pudlak Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Hemoglobinopathies	Sickle Cell Disease/Trait and other known HBB variants	IWK Clinical Genomics Laboratory: HBB	Diagnosis: Hematology, GENETICS Prenatal Diagnosis: GENETICS Carrier Testing (molecular confirmation): Fertility providers for the purpose of PGT planning *CBC, Hemoglobin electrophoresis and ferritin results are required *Variants will be reviewed to determine if they can be done in-house. Unknown variants and Newborn Screening cases must be performed at the Hamilton Health Sciences Lab

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	Thalassemias	Hamilton Health Sciences: Please check lab details	Diagnosis: Hematology, GENETICS 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, Internal Medicine
Schwachman-Diamond Syndrome	SBDS Single Gene Testing	IWK Clinical Genomics Laboratory: SBDS	Diagnosis: Hematology, GENETICS
Thrombocytopenia	Thrombocytopenia Panel	IWK Clinical Genomics Laboratory: ABCG5-ABCG8-ACTN1-ADAMTS13-ANKRD26-ARPC1B-CD36-CYCS-EFL1-ETV6-FLI1-FLNA-FYB1-GATA1-GBA1-GFI1B-GP1BA-GP6-GP9-HOXA11-IKZF5-ITGA2B-ITGB3-MASTL-MECOM-MPIG6B-MPL-MYH9-NBEAL2-PRKACG-PTPRJ-RBM8A-RUNX1-SLFN14-SRC-THPO-TPM4-TUBB1-VWF-WAS	Diagnosis: Hematology, GENETICS
Von Willebrand Disease (VWD)	Analysis for Type 2N VWD, 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, GENETICS *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	IWK Clinical Genomics Laboratory: C1QA-C1QB-C1QBP-C1QC-C1R-C1S-C2-C3-C3AR1-C4BPA-C4BPB-C5-C5AR1-C5AR2-C6-C7-C8A-C8B-C8G-C9-CD46-CD55-CD59-CD93-CFI-CFB-CFD-CFH-CFHR5-CFP-COLEC11-DGKE-FCN1-FCN2-FCN3-MASP1-MASP2-MAT2A-MBL2-SERPING1-THBD-VTN	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-IL10-IL10RA-IL10RB-IL21-IL21R-IL2RA-IL2RB-ITGB2-JAK1-	Diagnosis: Immunology, GENETICS

		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-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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Immunology
Severe Combined Immunodeficiency	Severe Combined Immunodeficiency Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Immunology
VEXAS Syndrome	UBA1	Department of Pathology and Laboratory Medicine – Central Zone (QEII): Please check lab details	Restrictions: None

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METABOLIC			
Acid Sphingomyelinase Deficiency (ASMD)	SMPD1 Single Gene Testing	IWK Clinical Genomics Laboratory: SMPD1	GENETICS Only
Arylsulfatase A Deficiency/ Metachromatic Leukodystrophy (MLD)	ARSA Single Gene Testing	IWK Clinical Genomics Laboratory: ARSA	GENETICS Only
Bartter Syndrome	Bartter Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	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 Common Variant Panel	IWK Clinical Genomics Laboratory: 19 DPYD variants via the Insight DPYD kit (Yourgene Diagnostics)	Diagnosis: GENETICS, Oncology
	DPYD Single Gene Testing	IWK Clinical Genomics Laboratory: DPYD	Diagnosis: GENETICS, Oncology *The DPYD common variant panel must be performed first. If the patient had no causative variants on this initial test but experienced adverse reactions, ordering provider can consider DPYD gene sequencing
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	Not currently available at the IWK CGL. Please contact the lab for options	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

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Glutaric Acidemia Type I	*GCDH* Single Gene Test	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Glutaric Acidemia Type II	Glutaric Acidemia Type II Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Glycogen Storage Disorders (GSD)	Glycogen Storage Disorder Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Hereditary Acrodermatitis Enteropathica	Hereditary Acrodermatitis Enteropathica Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Hyperammonemia	Hyperammonemia and Urea Cycle Disorder Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Hypoglycemia	Hypoglycemia Panel	IWK Clinical Genomics Laboratory: ABCC8-ACAD9-ACADL-ACADM-ACADS-ACADSB-ACADVL-ACAT1-ACSF3-AGL-AKT2-ALDH5A1-ALDOA-ALDOB-CA5A-CPT1A-CPT2-DECR1-DGUOK-DLD-ECHS1-ENO3-EPM2A-ETFA-ETFB-ETFDH-FBP1-FLAD1-G6PC1-GAA-GALT-GBE1-GCK-GK-GLUD1-GYG1-GYS1-GYS2-HADH-HADHA-HADHB-HMGCL-HMGCS2-HNF1A-HNF4A-HSD17B10-INSR-KCNJ11-LAMP2-LDHA-LPIN1-MLYCD-MPV17-NHLRC1-NNT-OXCT1-PC-PCK1-PCK2-PDX1-PFKM-PGAM2-PGK1-PGM1-PHKA1-PHKA2-PHKB-PHKG2-PRKAG2-PRKAG3-PTF1A-PYGL-PYGM-RBCK1-SLC16A1-SLC22A5-SLC25A20-SLC2A2-SLC37A4-TANGO2-TAFAZZIN-TBC1D4	GENETICS Only
Leukodystrophy	Leukodystrophy and Leukoencephalopathy Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neurology
Lysosomal Storage Disorders (and Mucopolysaccharidosis disorders, MPS)	Lysosomal Storage Disorders and Mucopolysaccharidosis Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
	MPS 1: IDUA Single Gene Testing	IWK Clinical Genomics Laboratory: IDUA	GENETICS Only

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Maple Syrup Urine Disease	Maple Syrup Urine Disease Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neurology
Mitochondrial Diseases	Mitochondrial Genome Test	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
	Mitochondrial DNA Depletion Syndrome Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Niemann-Pick Disease Type C	NPC1 and NPC2 Gene Sequencing and Del/Dup	IWK Clinical Genomics Laboratory: NPC1-NPC2	Diagnosis: GENETICS, Neurology
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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neurology
Primary Hyperoxaluria	Primary Hyperoxaluria Panel	IWK Clinical Genomics Laboratory: AGXT-GRHPR-HOGA1	Diagnosis: GENETICS, Nephrology, Urology
Tyrosinemia	*FAH* Single Gene Test	IWK Clinical Genomics Laboratory: FAH	GENETICS Only
	Tyrosinemia Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	IWK Clinical Genomics Laboratory: ALS2-ANG-ANXA11-CHCHD10-CHMP2B-DCTN1-ERBB4-FIG4-FUS-HNRNPA1-MATR3-KIF5A-NEFH-NEK1-OPTN-PFN1-PRPH-SETX-SIGMAR1-SOD1-SPG11-SQSTM1-TARDBP-TBK1-TUBA4A-UBQLN2-VAPB-VCP	Diagnosis: GENETICS, Neurology, Physical Medicine and Rehabilitation (Physiatry)
Ataxia	Ataxia Panel	Not currently available at the IWK CGL. Please contact the lab for options	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	Familial Cerebral Cavernous Malformation (FCCM) Panel	IWK Clinical Genomics Laboratory: CCM2-KRIT1-PDCD10-RASA1	Diagnosis: GENETICS, Hematology, Oncology
	Lissencephaly Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
	Comprehensive Brain Malformation Panel	Not currently available at the IWK CGL. Please contact the lab for options	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

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	Neuropathy and Charcot-Marie-Tooth Disease (CMT)	AARS1-ABCA1-ABHD12-AGTPBP1-AIFM1-APOA1-APTX-ARHGAP19-ARSA-ATL1-ATL3-ATP1A1-ATP7A-B4GALNT1-BAG3-BICD2-BSCL2-CADM3-CD59-CHCHD10-CNTNAP1-COA7-COX6A1-CPOX-CTDP1-CYP27A1-DARS2-DCTN1-DEGS1-DNAJB2-DNAJC3-DNM2-DNMT1-DST-DYNC1H1-EGR2-ELP1-ERCC6-ERCC8-FAH-FBLN5-FBXO38-FGD4-FIG4-FLVCR1-FXN-GALC-GAN-GARS1-GBA2-GBF1-GDAP1-GJB1-GJC2-GLA-GNB4-GSN-HADHA-HADHB-HARS1-HINT1-HK1-HMBS-HSPB1-HSPB8-HYCC1-IARS2-IGHMBP2-INF2-ITPR3-KIF1A-KIF5A-LITAF-LMNA-LRSAM1-LYST-MCM3AP-MFN2-MME-MORC2-MPV17-MPZ-MTMR2-MTRFR-MTTP-NAGA-NDC1-NDRG1-NEFH-NEFL-NGF-NTRK1-OPA1-PEX10-PEX7-PHYH-PLEKHG5-PMM2-PMP2-PMP22-PNKP-POLG-POLR3A-PPOX-PRDM12-PRNP-PRPS1-PRX-RAB7A-RCC1-REEP1-RETREG1-SACS-SBF1-SBF2-SCN10A-SCN11A-SCN9A-SEPTIN9-SETX-SH3TC2-SIGMAR1-SLC12A6-SLC25A19-SLC25A46-SLC52A2-SLC52A3-SLC5A7-SORD-SOX10-SPG11-SPTAN1-SPTBN4-SPTLC1-SPTLC2-SURF1-SYT2-TFG-TRIM2-TRPV4-TTPA-TTR-TUBB3-TYMP-VPS13A-VRK1-WARS1-WNK1-XK-YARS1-ZFYVE26	Diagnosis: GENETICS, Neurology, Physiatry
Congenital Myasthenic Syndrome	Congenital Myasthenic Syndromes Panel	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neurology
Dementia	Dementia Panel	IWK Clinical Genomics Laboratory: APOE-APP-CHCHD10-CHMP2B-CSF1R-CDTN1-FUS-GRN-ITM2B-MAPT-NOTCH3-PRNP-PSEN1-PSEN2-SNCA-SQSTM1-TARDBP-TBK1-TREM2-TYROBP-UBQLN2-VCP	Diagnosis: GENETICS, Neurology
Dentatorubral-Pallidoluysian Atrophy (DRPLA)	Dentatorubral-Pallidoluysian Atrophy (DRPLA) via the ATN1 CAG Repeat	North York General Hospital: Please check lab details	Diagnosis: GENETICS, Neurology

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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-TOR1A-TUBB4A-UBTF-VAC14-VPS13A-VPS16-WDR45-XPR1	Diagnosis: GENETICS, Neurology
Epilepsy	Childhood Onset Epilepsy Panel	IWK Clinical Genomics Laboratory: ADSL-ARX-ATP1A3-ATRX-CDKL5-CHD2-CLCN4-CNTNAP2-DEPDC5-DNAJC5-DYRK1A-EHMT1-FOXG1-GABBR2-GABRB2-GABRG2-GRIN2A-GRIN2D-KANSL1-KCNJ10-KCNMA1-KCNQ3-KDM5C-MBD5-MECP2-MEF2C-NEXMIF-NGLY1-NRXN1-PAK3-PCDH19-PHF6-PIGA-PIGN-PIGO-PNKP-POLG-PRRT2-RAB39B-ROGDI-SCN1A-SCN1B-SCN2A-SLC2A1-SLC6A1-SLC6A8-SLC9A6-SMARCA2-STX1B-SYN1-SYNGAP1-TBC1D24-	Diagnosis: GENETICS, Neurology

		TCF4-TRPM3-TSC1-TSC2-UBE3A-WDR45-ZEB2	
	Comprehensive Epilepsy Panel	IWK Clinical Genomics Laboratory: ABAT-ACTB-ACTG1-ADGRG1-ADSL-AFG2A-AKT3-ALDH7A1-ALG13-AMT-AP3B2-ARFGEF2-ARHGEF9-ARV1-ARX-ASAH1-ASNS-ATP1A2-ATP1A3-ATP6V0A2-ATP7A-ATRX-B3GALNT2-B4GAT1-CACNA1A-CACNA1E-CAD-CDKL5-CHD2-CHRNA4-CHRNB2-CLCN4-CLN3-CLN5-CLN6-CLN8-CNTNAP2-CSTB-CTSD-CTSF-DCX-DEPDC5-DNAJC5-DNM1-DOCK7-DYNC1H1-DYRK1A-EEF1A2-EHMT1-EPM2A-FGF12-FKRP-FKTN-FLNA-FOLR1-FOXG1-FRRS1L-GABBR2-GABRA1-GABRB2-GABRB3-GABRG2-GAMT-GLDC-GMPPB-GNAO1-GOSR2-GPSM2-GRIN1-GRIN2A-GRIN2B-GRIN2D-GRN-HCN1-HNRNPU-ITPA-KANSL1-KATNB1-KCNA1-KCNA2-KCNB1-KCNC1-KCNH5-KCNJ10-KCNMA1-KCNQ2-KCNQ3-KCNT1-KCTD7-KDM5C-KIF2A-KIFBP-LAMA2-LARGE1-LGI1-MBD5-MDH2-MECP2-MEF2C-MFSD8-MOCS1-NDE1-NEU1-NEXMIF-NGLY1-NHLRC1-NPRL2-NPRL3-NRXN1-OCLN-PAFAH1B1-PAK3-PCDH19-PHF6-PHGDH-PIGA-PIGG-PIGN-PIGO-PIGT-PIGV-PLCB1-PLPBP-PNKP-PNPO-POLG-POMGNT1-POMGNT2-POMK-POMT1-POMT2-PPT1-PRRT2-PSAT1-PSPH-PURA-RAB18-RAB39B-RAB3GAP1-RAB3GAP2-RELN-ROGDI-RTTN-SCARB2-SCN1A-SCN1B-SCN2A-SCN3A-SCN8A-SERPINI1-SGCE-SLC12A5-SLC13A5-SLC19A3-SLC25A12-SLC25A22-SLC2A1-SLC35A2-SLC6A1-SLC6A8-SLC9A6-SMARCA2-SNAP29-SPTAN1-SRD5A3-ST3GAL5-STX1B-STXBP1-SUOX-SYN1-SYNGAP1-SYNJ1-SZT2-TBC1D24-TCF4-TPP1-TRPM3-TSC1-TSC2-TUBA1A-TUBB-TUBB2A-TUBB2B-TUBB3-UBA5-UBE3A-VLDLR-WDR45-WDR62-WWOX-YWHAG-ZEB2	Diagnosis: GENETICS, Neurology
	Early Infantile Epilepsy Panel	IWK Clinical Genomics Laboratory: ABAT-ADSL-AFG2A-ALDH7A1-ALG13-AP3B2-ARHGEF9-ARV1-ARX-CACNA1A-CACNA1E-CAD-	Diagnosis: GENETICS, Neurology

		CDKL5-CHD2-DCX-DNM1-DOCK7-DYRK1A-EEF1A2-FGF12-FOLR1-FOXG1-FRRS1L-GABBR2-GABRA1-GABRB2-GABRB3-GABRG2-GAMT-GLDC-GNAO1-GRIN2A-GRIN2B-GRIN2D-HCN1-HNRNPU-ITPA-KANSL1-KCNA1-KCNA2-KCNB1-KCNH5-KCNQ2-KCNQ3-KCNT1-MDH2-MECP2-MEF2C-NGLY1-PCDH19-PIGA-PIGG-PIGN-PIGO-PIGT-PIGV-PLCB1-PNKP-PNPO-POLG-PRRT2-PURA-ROGDI-SCN1A-SCN1B-SCN2A-SCN8A-SLC12A5-SLC13A5-SLC2A1-SLC25A12-SLC25A22--SLC35A2-SLC6A8-SPTAN1-ST3GAL5-STX1B-STXBP1-SYNGAP1-SYNJ1-SZT2-TBC1D24-UBA5-WDR45-WWOX-YWHAG	
	Focal Epilepsy Panel	IWK Clinical Genomics Laboratory: CHRNA4-CHRNB2-DEPDC5-GRIN2A-KANSL1-KCNT1-LGI1-NPRL2-NPRL3-PRRT2-SCN1A-SCN1B-SLC2A1	Diagnosis: GENETICS, Neurology
	Focal Epilepsy & Malformations Panel	IWK Clinical Genomics Laboratory: ACTB-ACTG1-ADGRG1-AKT3-ARFGEF2-ARX-ASNS-ATP6V0A2-B3GALNT2-B4GAT1-DCX-DYNC1H1-FKRP-FKTN-FLNA-GMPPB-GPSM2-POMGNT2-KATNB1-KIF1BP-KIF2A-LAMA2-LARGE1-NDE1-OCLN-PAFAH1B1-POMGNT1-POMT1-POMT2-RAB18-RAB3GAP1-RAB3GAP2-RELN-RTTN-POMK-SNAP29-SRD5A3-TUBA1A-TUBB-TUBB2A-TUBB2B-TUBB3-VLDLR-WDR62	Diagnosis: GENETICS, Neurology
	Neonatal / Infantile Actionable Gene Epilepsy Panel	IWK Clinical Genomics Laboratory: ALDH7A1-AMT-ATP7A-CAD-FOLR1-GAMT-GLDC-KCNQ2-KCNT1-MOCS1-PHGDH-PLPBP-PNPO-POLG-PSAT1-PSPH-SCN1A-SLC19A3-SLC2A1-SLC6A8-SUOX-TPP1-TRPM3-TSC1-TSC2	Diagnosis: GENETICS, Neurology
	Progressive Myoclonic Epilepsy Panel	IWK Clinical Genomics Laboratory: ASAH1-CLN3-CLN5-CLN6-CLN8-CSTB-CTSD-CTSF-EPM2A-GOSR2-GRN-KCNC1-KCTD7-MFSD8-NEU1-NHLRC1-PPT1-SCARB2-SERPINI1-SGCE-TPP1	Diagnosis: GENETICS, Neurology
Episodic Ataxia Type 2 (EA2) and Spinocerebellar	CACNA1A Single Gene Testing	IWK Clinical Genomics Laboratory: CACNA1A	Diagnosis: GENETICS, Neurology

Ataxia Type 6 (SCA6)			*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	Not currently available at the IWK CGL. Please contact the lab for options	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	Not currently available at the IWK CGL. Please contact the lab for options	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, Pediatrics
Hereditary Sensory and Autonomic Neuropathy (HSAN)	Hereditary Sensory and Autonomic Neuropathy Panel	*ATL1-ATL3-DNMT1-DST-ELP1-KIF1A-NGF-NTRK1-PRDM12-RAB7A-RETREG1-SCN11A-SCN9A-SPTLC1-SPTLC2-WNK1*	Diagnosis: GENETICS, Neurology, Physiatry
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-SPG7-TECPR2-TUBB4A-UBAP1-VPS37A-WASHC5-ZFYVE26	Diagnosis: GENETICS, Neurology, Physical Medicine and Rehabilitation (Physiatry)

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Huntington Disease (HD)	HD via Trinucleotide Repeat Expansion fPCR Kit	IWK Clinical Genomics Laboratory: HD-fPCR	Diagnosis: GENETICS, Neurology, Psychiatry, Geriatric Medicine
Microcephaly and Pontocerebellar Hypoplasia	Microcephaly and Pontocerebellar Hypoplasia Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Comprehensive Muscular Dystrophy and Myopathy (CMDM)	Comprehensive Muscular Dystrophy/Myopathy Panel	IWK Clinical Genomics Laboratory: ABHD5-ACAD9-ACADM-ACADVL-ACTA1-ACTN2-ADSS1-AGL-ALDOA-AMPD1-ANO5-ASCC1-ASCC3-B3GALNT2-B4GAT1-BAG3-BET1-BIN1-CACNA1S-CAPN3-CAV3-CAVIN1-CFL2-CHKB-COL12A1-COL13A1-COL25A1-COL6A1-COL6A2-COL6A3-COQ4-COQ8A-COX6A2-CPT2-CRPPA-CRYAB-DAG1-DES-DGUOK-DMD-DNAJB4-DNAJB6-DNM2-DOK7-DOLK-DPM2-DPM3-DTNA-DYSF-ECEL1-EMD-ENO3-EPG5-ETFA-ETFB-ETFDH-FDX2-FHL1-FKBP14-FKRP-FKTN-FLAD1-FLNC-FXR1-GAA-GBE1-GFER-GGPS1-GMPPB-GNE-GOLGA2-GOSR2-GYG1-GYS1-HACD1-HADHA-HADHB-HMGCR-HNRNPA2B1-HNRNPDL-INPP5K-ISCU-ITGA7-JAG2-KBTBD13-KLHL40-KLHL41-KY-LAMA2-LAMP2-LARGE1-LDHA-LETM1-LMNA-LMOD3-LPIN1-MAP3K20-MEGF10-MICU1-MLIP-MSTO1-MTM1-MYBPC1-MYH2-MYH3-MYH7-MYL1-MYL2-MYMK-MYO18B-MYOD1-MYOT-MYPN-NEB-OBSCN-ORAI1-PAX7-PFKM-PGAM2-PGK1-PGM1-PHKA1-PIEZO2-PLEC-PNPLA2-POGLUT1-POLG-POLG2-POMGNT1-POMGNT2-POMK-POMT1-POMT2-POPDC1-POPDC3-PRKAG2-PYGM-PYROXD1-RBCK1-RRM2B-RXYLT1-RYR1-RYR3-SCN4A-SELENON-SGCA-SGCB-SGCD-SGCG-SIL1-SLC22A5-SLC25A4-SMCHD1-SPEG-SPTBN4-STAC3-STIM1-SUCLA2-SYNE1-SYNE2-TAMM41-TANGO2-TCAP-TK2-TNNC2-TNNI2-TNNT1-TNNT3-TNPO3-TOR1AIP1-TPM2-TPM3-TRAPPC11-TRDN-TRIM32-TRIP4-TSFM-TTN-TYMP-UNC45B-VCP-VMA21-ZC4H2	Diagnosis: GENETICS, Neurology, Physiatry

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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	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neurology
Oculopharyngeal Muscular Dystrophy (OPMD)	Oculopharyngeal Muscular Dystrophy via the PABPN1 (GCN) Repeat Expansion	North York General Hospital: Please check lab details	Diagnosis: GENETICS, Neurology, Ophthalmology
Parkinson Disease/Parkinsonism	Parkinson Panel	IWK Clinical Genomics Laboratory: ATP13A2-ATP1A3-C19Orf12-CHCHD2-CP-CSF1R-DCTN1-DNAJC6-FBXO7-FTL-GBA1-GCH1-GRN-LRRK2-LYST-MAPT-PANK2-PARK7-PDGFB-PINK1-PLA2G6-PRKN-PRKRA-PTRHD1-RAB39B-SLC30A10-SLC39A14-SLC6A3-SNCA-SPG11-SPR-SYNJ1-TH-VPS13A-VPS13C-VPS35-WDR45	Diagnosis: GENETICS, Neurology
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-TUBA8-TUBB2A-TUBB2B-TUBB3-WDR62	Diagnosis: GENETICS, Neurology
Primary Familial Brain Calcification	Primary Familial Brain Calcification Panel	IWK Clinical Genomics Laboratory: CMPK2-JAM2-MYORG-PDGFB-PDGFRB-SLC20A2-XPR1	Diagnosis: GENETICS, Neurology
PRNP-Related Disorders	PRNP-Related Disorders Panel	PRNP (including c.385A>G)* *c.385A>G is classified as a benign/likely benign	Diagnosis: GENETICS, Neurology, Psychiatry, Physical Rehabilitation and Physiatry, Geriatrics

		modifier variant and will be reported in the context of a relevant pathogenic/likely pathogenic variant or VUS.	*Ordering provider must indicate that the patient's clinical presentation is highly indicative of a PRNP-related disorder (Genetic Creutzfeld-Jacob Disease (CJD), Fatal Familial Insomnia (FFI) and/or Gerstmann-Sträussler-Scheinker syndrome (GSS).
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	North York General Hospital: Please check lab details for gene content	Diagnosis: GENETICS, Neurology
Spinal Muscular Atrophy (SMA)	SMN Gene Dosage via MLPA	IWK Clinical Genomics Laboratory: SMA-MLPA	Diagnosis: GENETICS, Neurology, Pediatrics, Physical Medicine and Rehabilitation (Physiatry)
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
Gastric Cancer	Gastric Cancer Panel	APC(protein coding sequence exon 1 only)-ATM-BMPR1A-BRCA1-BRCA2-CDH1-CHEK2-CTNNA1-EPCAM-MLH1-MSH2-MSH6-PALB2-PMS2-SMAD4-STK11-TP53	GENETICS Only
Hereditary Breast and Ovarian Cancer (HBOC)	HBOC Panel	IWK Clinical Genomics Laboratory: ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN (common mutation only), PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11 and TP53	Diagnosis: GENETICS, Oncology
Hereditary Cancers	Hereditary Cancers Panel	IWK Clinical Genomics Laboratory: AIP-AKT1-APC-ATM-AXIN2-BAP1-BARD1-BMPR1A-BRCA1-BRCA2-BRIP1-CASR-CDC73-CDH1-CDK4-CDKN1B-CDKN2A-CHEK2-CTNNA1-DICER1-EPCAM-FH-FLCN-GREM1-HOXB13-MAX-MBD4-MEN1-MET-MLH1-MLH3-MSH2-MSH3-MSH6-MUTYH-NBN-NF1-NTHL1-PALB2-PMS2-POLD1-POLE-PRKAR1A-PTCH1-PTEN-RAD51C-RAD51D-RET-RNF43-RPS20-SDHA-SDHAF2-SDHB-SDHC-SDHD-SMAD4-STK11-SUFU-TMEM127-TP53-TSC1-TSC2-VHL-WRN	GENETICS Only
Hereditary Thyroid Cancer	Hereditary Thyroid Cancer Panel	IWK Clinical Genomics Laboratory: AKT1-APC-DICER1-PRKAR1A-PTEN-RET-TP53-WRN	GENETICS Only
Lymphoid Neoplasms	Lymphoid Neoplasms Panel	IWK Clinical Genomics Laboratory:	Diagnosis: GENETICS, Hematology-Oncology

		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	
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	Diagnosis: Endocrinology, GENETICS
Multiple Endocrine Neoplasia Type 2 (MEN2)	MEN2 Single Gene Testing	IWK Clinical Genomics Laboratory: RET	Diagnosis: Endocrinology, GENETICS
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)-ERCC6L2-ETV6-FANCA-GATA2 (including intron 4)-IKZF1-MBD4-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, Hematology-Oncology
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	Diagnosis: GENETICS, Oncology
Pheochromocytoma & Paraganglioma	Pheochromocytoma & Paraganglioma Panel	IWK Clinical Genomics Laboratory: FH-MAX-NF1-RET-SDHA-SDHB-SDHC-SDHD-SDHAF2-	Diagnosis: Endocrinology, GENETICS

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		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, Oncology
Renal Cell Carcinoma Panel	Renal Cell Carcinoma Panel	IWK Clinical Genomics Laboratory: BAP1-CHEK2-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 *****If in-house testing on a peripheral blood sample is non-diagnostic (due to possible somatic mosaicism) and/or tumor testing is necessary, test will have to be referred out to Impact Genetics.
Succinate Dehydrogenase Complex (SDHx)	Succinate Dehydrogenase Complex (SDHx) Panel	IWK Clinical Genomics Laboratory: SDHA-SDHAF2-SDHB-SDHC-SDHD	GENETICS Only
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			
Achromatopsia	Achromatopsia Panel	IWK Clinical Genomics Laboratory: ATF6-CNGA3-CNGB3-GNAT2-PDE6C-PDE6H-RGS9-RGS9BP	Diagnosis: GENETICS, Ophthalmology
Aniridia	Aniridia Panel	IWK Clinical Genomics Laboratory: PAX6 (SNV and dosage)-WT1 (dosage only)	GENETICS Only
Cone Rod Dystrophy	Cone Rod Dystrophy Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Duane Syndrome	Duane Syndrome Panel	IWK Clinical Genomics Laboratory: CHN1-MAFB-SALL4	Diagnosis: GENETICS, Ophthalmology
Ectopia Lentis	Ectopia Lentis Panel	IWK Clinical Genomics Laboratory: AASS-ADAMTS10-ADAMTS17-ADAMTSL4-ASPH-BCOR-CBS-COL18A1-FBN1-LTBP2-P3H2-PORCN-SUOX-VSX2	GENETICS Only
Leber Congenital Amaurosis	Leber Congenital Amaurosis Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Optic Atrophy	Optic Atrophy	Not currently available at the IWK CGL. Please contact the lab for options	Diagnosis: GENETICS, Neuro-Ophthalmology
Retinal Dystrophy	Retinal Dystrophy Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Retinitis Pigmentosa (RP)	Retinitis Pigmentosa Panel	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Septo-Optic Dysplasia	Septo-Optic Dysplasia Panel	IWK Clinical Genomics Laboratory: HESX1-GLI2-HK1-OTX2-PAX6-SOX2	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	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only

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SKELETAL			
Achondroplasia (common mutations only)	FGFR3 c.1138G>A and c.1138G>G via NGS	IWK Clinical Genomics Laboratory: FGFR3	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	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only
Cleidocranial Dysplasia (CCD) Spectrum Disorder	RUNX2 Single Gene Testing	IWK Clinical Genomics Laboratory: RUNX2	GENETICS Only
Craniosynostosis	Craniosynostosis Panel	IWK Clinical Genomics Laboratory: ALPL-ALX4-ASXL1-CD96-CDC45-COLEC11-CYP26B1-ESCO2-EFNB1-ERF-FGF9-FGFR1-FGFR2-FGFR3-FREM1-GLI3-IFT122-IFT140-IL11RA-KAT6A-MASP1-MEGF8-MSX2-P4HB-POR-PPP3CA-RECQL4-SEC24D-SKI-SLC25A24-SMAD6- SOX6-SPECC1L-TCF12-TGFBR1-TGFBR2-TMCO1-TWIST1-WDR19-WDR35-ZIC1	GENETICS Only
Limb Malformations	Limb Malformations Panel	IWK Clinical Genomics Laboratory: ARHGAP31-ARID1A-ARID1B-ATR-BHLHA9-BRCA2-BRIP1-CDH3-DHODH-DLL4-DLX5-DOCK6-EOGT-ERCC4-ESCO2-FANCA-FANCB-FANCC-FANCD2-FANCE-FANCF-FANCG-FANCI-FANCL-FANCM-FGF10-FGFR2-GDF5-GLI3-HDAC8-KYNU-LMBR1-LRP4-NIPBL-NOTCH1-NSDHL-PALB2-PITX1-PUF60-RAD21-RAD51C-RBM8A-RBPJ-RECQL4-RSPO2-SALL1-SALL4-SF3B4-SLX4-SMC1A-SMC3-TBX3-TBX5-THPO-TP63-WNT7A-WNT10B-XRCC2	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-	Diagnosis: GENETICS, IWK Suspected Trauma and Abuse Response Team (START)

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		TNFRSF11B-WNT1-XYLT2	
Skeletal Dysplasia	Comprehensive Growth Disorders/Skeletal Dysplasias and Disorders Panel (or sub-panel)	Not currently available at the IWK CGL. Please contact the lab for options	GENETICS Only