Genetic Resource Centre Established Testing Menu

This is a list of commonly ordered tests and preferred labs (**this is not a pre-approved test list**)

For testing that has more than one lab listed, please choose your preferred test/lab based on gene content

To search for a test, click “Ctrl-F” on the computer keyboard and type in part of the test name. Click “enter” until you find the test of interest.

GRC Contact Information Phone: 403-955-5400 Email: grc@albertaprecisionlabs.ca Fax: 403-592-4238	**Ordering restrictions** - Carrier testing/presymptomatic testing is currently restricted to Clinical Genetics. Testing for symptomatic patients may be restricted based on clinical specialty. Please contact the Genetic Resource Centre if you have questions regarding ordering restrictions.	**How to use an online portal?** For assistance, please contact: Blueprint Genetics: Sarah De Souza (sarah.desouza@blueprintgenetics.com) GeneDx: Cassandra Dawson (cdawson@genedx.com) Invitae: Hana Sroka (hana.sroka@invitae.com) Prevention Genetics: Dominique Robeyns (dominique.robeyns@preventiongenetics.com)

Indication / Suspected Diagnosis	Test Name	Laboratory	Link to Online Portal or Requisition	Notes
Add-on testing				
Any NGS panel	Flex Testing	Blueprint Genetics	Online Portal	Can add up to 200 additional genes to Blueprint Genetics panel, free of charge
Site Specific Testing - PLEASE READ NOTES SECTION BEFORE SELECTING LAB				
Site-specific testing	Familial Variant Testing/Targeted Variant Testing	Blueprint Genetics	Online Portal	Select this lab if they performed testing for the index patient, if the lab who tested the index patient is not listed below, or if you are requesting testing to confirm a research variant. Blueprint Genetics cannot perform targeted testing for copy number variants or prenatal targeted testing if they did not test the index patient. Contact the GRC if you have any questions.
		Exeter Genomics Laboratory	Diabetes Genetic Test Referral Forms	Select this lab if they performed testing for the index patient (MODY testing)
			Click on "MODY (R141 & R142) Genetic Testing Referral Form"	

	Familial Variant Testing/Targeted Variant Testing	Molecular Otolaryngology & Renal Research Laboratories	Requisition form	Select this lab if they performed testing for the index patient
Cancer/Tumour/Overgrowth				
Legius syndrome	*SPRED1* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *NF1* sequencing and del/dup by NGS
Megalencephaly	Megalencephaly Panel	University of Washington	Website	Click on "ordering & collection" to access the link for the requisition form
MMR deficient tumour	ColoSeq™ Tumor Panel	University of Washington	Website	Click on "ordering & collection" to access the link for the requisition form
	ColoSeq™ Tumor Single Gene			
Schwannomatosis	*LZTR1* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Schwannomatosis Panel – *NF2*, *SMARCB1*
Somatic overgrowth	Somatic Overgrowth Gene Set	Washington University (St. Louis, MO)	Requisition form	
Cardiology				
Congenital structural heart disease	Congenital Structural Heart Disease Panel	Blueprint Genetics	Online Portal	
Heterotaxy and situs inversus	Heterotaxy and Situs Inversus Panel	Blueprint Genetics	Online Portal	
Dermatology				
Adams-Oliver syndrome	Adams-Oliver Syndrome Panel	Blueprint Genetics	Online Portal	
Albinism	Albinism Panel	Blueprint Genetics	Online Portal	

Ectodermal dysplasia	Ectodermal Dysplasia Panel	Blueprint Genetics	Online Portal	
Epidermolysis bullosa	Epidermolysis Bullosa Panel	Blueprint Genetics	Online Portal	
Ichthyosis	Ichthyosis Panel	Blueprint Genetics	Online Portal	
Incontinentia pigmenti	*IKBKG* (NEMO) Gene Sequencing & Common Del/Dup	GeneDx	Online Portal	
Palmoplantar Keratoderma	Palmoplantar Keratoderma Panel	Blueprint Genetics	Online Portal	
Pseudoxanthoma elasticum	*ABCC6* single gene test	Blueprint Genetics	Online Portal	
Dysmorphology				
Brachydactyly/syndactyly	Brachydactyly / Syndactyly Panel	Blueprint Genetics	Online Portal	
CHARGE syndrome	*CHD7* single gene test	Blueprint Genetics	Online Portal	
Cleft lip/palate (syndromic)	Cleft Lip/Palate and Associated Syndromes Panel	Blueprint Genetics	Online Portal	
Cleft lip/palate (non-syndromic)	Cleft Lip/Cleft Palate Panel	Prevention	Website Click on "log in/sign up myPrevent" to access online portal	
Cornelia de Lange syndrome	Cornelia de Lange Syndrome Panel	Blueprint Genetics	Online Portal	
Craniosynostosis	Craniosynostosis Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Craniosynostosis panel
Facial dysostosis	Facial Dysostosis and Related Disorders Panel	Blueprint Genetics	Online Portal	
Kabuki Syndrome	Kabuki Syndrome Panel	Blueprint Genetics	Online Portal	
Rasopathy Disorders	Noonan Syndrome Panel	Blueprint Genetics	Online Portal	
van der Woude syndrome	Invitae van der Woude Syndrome Panel	Invitae	Online Portal	

Ear, Nose & Throat				
Branchio-Oto-Renal Syndrome	Branchio-Oto-Renal (BOR) Syndrome Panel	Blueprint Genetics	Online Portal	
Non-syndromic hearing loss	Non-Syndromic Hearing Loss Panel	Blueprint Genetics	Online Portal	
	OtoSCOPE® Panel	Molecular Otolaryngology & Renal Research Laboratories	Requisition form	
Syndromic hearing loss	Comprehensive Hearing Loss and Deafness Panel (or sub-panel)	Blueprint Genetics	Online Portal	
Waardenburg syndrome	Waardenburg Syndrome Panel	Blueprint Genetics	Online Portal	
Endocrinology				
Congenital adrenal hyperplasia	Congenital Adrenal Hyperplasia Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *CYP21A2* sequencing and MLPA *CYP11B1* sequencing
Congenital hyperinsulinism	Congenital Hyperinsulinism Panel	Exeter Genomics Laboratory	Hyperinsulinism Web Page Click on "Request Form"	**Coordinate parental blood sample collection** at the time that you submit your funding request. Parental blood samples are required to help with result interpretation. **
Diabetes insipidus	Diabetes Insipidus Panel	Blueprint Genetics	Online Portal	
Disorders of sexual development	Abnormal Genitalia / Disorders of Sexual Development Panel	Blueprint Genetics	Online Portal	

Familial hyperaldosteronism	*CYP11B1/CYP11B2* Fusion Gene	Center for Nephrology and Metabolic Disorders	Requisition form Consent form	For non-urgent cases where both fusion gene testing and panel testing are indicated, testing should be performed stepwise (requires two GRC funding requests)
	Primary Aldosteronism Panel	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	
Familial hyperparathyroidism	Hyperparathyroidism Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Endocrine Disorders Panel - *AIP, CASR, CDC73, CDKN1B, MEN1, PRKAR1A, RET*
Familial hypocalciuric hypercalcemia	Familial Hypocalciuric Hypercalcemia (FHH) Panel	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	In-house testing must be performed first, if relevant: *CASR* sequencing and del/dup by NGS
GNAS-related disorders	*GNAS* – Methylation and Del/Dup analysis	Genome Diagnostics, Amsterdam UMC	Website*	For disorders of *GNAS* inactivation: Pseudohypoparathyroidism, Pseudopseudohypoparathyroidism Progressive osseous heteroplasia Osteoma cutis -Test includes sanger sequencing *Use Google Translate as needed in Chrome browser
	Next-Gen Sequencing of *GNAS* (GNAS-NG)	University of Alabama	Requisition form	For postzygotic *GNAS* somatic variants: McCune-Albright syndrome
Hypogonadotropic hypogonadism/Kallmann syndrome	Kallmann Syndrome Panel	Blueprint Genetics	Online Portal	

Hypophosphatemic rickets	Hypophosphatemic Rickets Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *ALPL* sequencing and del/dup by NGS
	PHEX single gene test	Blueprint Genetics	Online Portal	
Liddle syndrome	Liddle Syndrome Panel	Blueprint Genetics	Online Portal	
Mature onset diabetes of the young (MODY)	*GCK* sequencing or MODY Panel	Exeter Genomics Laboratory	Diabetes Genetic Test Referral Forms Click on "MODY (R141 & R142) Genetic Testing Referral Form"	
	MODY Panel	Blueprint Genetics	Online Portal	
Monogenic obesity	Monogenic Obesity Panel	Blueprint Genetics	Online Portal	
Thyroid hormone resistance	*THRB* single gene test	Blueprint Genetics	Online Portal	
Gastroenterology				
Cholestasis	Cholestasis Panel	Blueprint Genetics	Online Portal	
Congenital diarrhea	Congenital Diarrhea Panel	Blueprint Genetics	Online Portal	
Gilbert syndrome	*UGT1A1* single gene test	Blueprint Genetics	Online Portal	
Hirschsprung disease	Hirschsprung Disease Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *RET* and *PHOX2B* sequencing and del/dup by NGS

Pancreatitis	Pancreatitis Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *PRSS1* and *CFTR* targeted sequence analysis
Polycystic liver disease	Polycystic Liver Disease Panel	Blueprint Genetics	Online Portal	
Hematology				
Anemia	Anemia Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *HBA1/HBA2* multiplex PCR and sequencing *HBB* sequencing and MLPA
Bone marrow failure	Bone Marrow Failure Syndrome Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Fanconi Anemia and DNA Repair Disorders Panel - *ATM, BLM, BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51C, REQL4, SLX4* *SBDS* sequencing and del/dup by NGS
Coagulopathy	Bleeding Disorder/Coagulopathy Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Hemophilia A Inverse Shifting PCR
Congenital neutropenia	Congenital Neutropenia Panel	Blueprint Genetics	Online Portal	
Diamond-Blackfan anemia	Diamond-Blackfan Anemia Panel	Blueprint Genetics	Online Portal	
Hereditary leukemia	Hereditary Leukemia Panel	Blueprint Genetics	Online Portal	For hematological malignancies**,** the ideal tissue type for germline testing is fibroblast culture. However, buccal samples will be accepted to allow for a rapid turnaround time if urgently required for clinical management.

Hereditary spherocytosis	Red Blood Cell Membrane Disorder Panel	Blueprint Genetics	Online Portal	
Hermansky-Pudlak syndrome	Hermansky-Pudlak Syndrome Panel	Blueprint Genetics	Online Portal	
Platelet Disorder	Platelet Function Disorder Panel	Blueprint Genetics	Online Portal	
Thrombocytopenia	Thrombocytopenia Panel	Blueprint Genetics	Online Portal	
Immunology				
Complement system disorder	Complement System Disorder Panel	Blueprint Genetics	Online Portal	
Familial Mediterranean fever	*MEFV* sanger sequencing	BC Molecular Genetics Laboratory	Requisition form	
Hemophagocytic lymphohistiocytosis	Hemophagocytic Lymphohistiocytosis Panel	Blueprint Genetics	Online Portal	
Inborn errors of immunity	Comprehensive Immune and Cytopenia Panel*	Blueprint Genetics	Online Portal	For patients under investigation for inborn errors of innate and adaptive immunity where the timing of the diagnosis is crucial. Includes the genes from the Primary Immunodeficiency Panel, Severe Combined Immunodeficiency Panel, Bone Marrow Failure Syndrome Panel, HLH Panel, and more. Please see Blueprint Genetics website for more information on gene content. *This panel cannot have flex genes added.
Periodic fever syndrome	Autoinflammatory Syndrome Panel	Blueprint Genetics	Online Portal	

Primary immunodeficiency	Primary Immunodeficiency Panel	Blueprint Genetics	Online Portal	
Primary immunodeficiency and primary ciliary dyskinesia	Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel	Blueprint Genetics	Online Portal	See Pulmonology section for Primary Ciliary Dyskinesia Panel
Severe combined immunodeficiency	Severe Combined Immunodeficiency Panel	Blueprint Genetics	Online Portal	
Metabolic				
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL* single gene test	Blueprint Genetics	Online Portal	
	Invitae Elevated C5-OH Panel	Invitae	Online Portal	Panel includes genes for secondary newborn screening targets (*MCCC1*, *MCCC2*, *AUH*, *TAZ*, *OPA3*, *SERAC1*, *DNAJC19*, *ACAT1*, *HSD17B10*)
Biotinidase deficiency	*BTD* single gene test	Blueprint Genetics	Online Portal	
Carnitine deficiency, systemic primary	*SLC22A5* single gene test	Blueprint Genetics	Online Portal	
Citrullinemia	Invitae Elevated Citrulline Panel	Invitae	Online Portal	
Fatty Acid Oxidation Syndrome	Fatty Acid Oxidation Syndrome Panel	Blueprint Genetics	Online Portal	
	Invitae Fatty Acid Oxidation Defects Panel	Invitae	Online Portal	
Galactosemia	*GALT* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant (**This does not apply to urgent cases**): GALT targeted sequence analysis

Glutaric acidemia type 1	*GCDH* single gene test	Blueprint Genetics	Online Portal	Can add on genes for secondary targets (*ETFA, ETFB, ETFDH*) or reflex these genes free of charge within 90 days *SUGCT* is not currently available at Invitae, but can be ordered as part of the Blueprint Genetics "Organic Acidemia/Aciduria & Cobalamin Deficiency Panel" if patient has a broad differential diagnosis
	Invitae Glutaric Acidemia Type I Test	Invitae	Online Portal	
Glycogen storage disorder	Glycogen storage disorder panel	Blueprint Genetics	Online Portal	
Hunter syndrome	*IDS* single gene test	Blueprint Genetics	Online Portal	
Hurler syndrome	*IDUA* single gene test	Blueprint Genetics	Online Portal	
Hyperammonemia	Hyperammonemia and Urea Cycle Disorder Panel	Blueprint Genetics	Online Portal	
Hypoglycemia	Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	Blueprint Genetics	Online Portal	
Isovaleric acidemia	*IVD* single gene testing	Blueprint Genetics	Online Portal	
Leukodystrophy	Leukodystrophy and Leucoencephalopathy panel	Blueprint Genetics	Online Portal	
Lipodystrophy	Congenital and Familial Lipodystrophy Panel	Blueprint Genetics	Online Portal	
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Invitae Elevated C16-OH, C16:1-OH, C18-OH and C18:1-OH Panel	Invitae	Online Portal	

Lysosomal storage disorders	Lysosomal Disorders and Mucopolysaccharidosis Panel	Blueprint Genetics	Online Portal	
Maple syrup urine disease	Invitae Elevated Leucine (MSUD) Panel	Invitae	Online Portal	
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant (**This does not apply to urgent cases**): *ACADM* targeted sequence analysis
	Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test	Invitae	Online Portal	Can add on genes for secondary newborn screening targets (*ETFA, ETFB, ETFDH*) or reflex these genes free of charge within 90 days
Metabolic Myopathy and Rhabdomyolysis	Metabolic Myopathy and Rhabdomyolysis Panel	Blueprint Genetics	Online Portal	
Methylmalonic acidemia	Invitae Methylmalonic Acidemia Panel	Invitae	Online Portal	Can add on combined methylmalonic acidemia and homocystinuria genes
Mitochondrial DNA Depletion Syndrome	Mitochondrial DNA Depletion Syndrome Panel	Blueprint Genetics	Online Portal	
Niemann-Pick disease type C	Invitae Niemann-Pick Disease Type C Panel	Invitae	Online Portal	
Organic Acidemia	Organic Acidemia/Aciduria & Cobalamin Deficiency Panel	Blueprint Genetics	Online Portal	
	Invitae Organic Acidemias Panel	Invitae	Online Portal	
Phenylketonuria	Hyperphenylalaninemia panel	Blueprint Genetics	Online Portal	
	PAH single gene test	Blueprint Genetics	Online Portal	

Pompe disease	*GAA* single gene test	Blueprint Genetics	Online Portal	
Porphyria	Porphyria Panel	Blueprint Genetics	Online Portal	
Propionic acidemia	Invitae Propionic Acidemia Panel	Invitae	Online Portal	
Tri-functional protein deficiency	Invitae Elevated C16-OH, C16:1-OH, C18-OH and C18:1-OH Panel	Invitae	Online Portal	
Tyrosinemia, type 1	*FAH* single gene test	Blueprint Genetics	Online Portal	
	Invitae Elevated Tyrosine (Tyrosinemia) Panel	Invitae	Online Portal	Includes genes for secondary newborn screening targets (*TAT, HPD*)
Very long chain acyl-CoA dehydrogenase deficiency	*ACADVL* single gene test	Blueprint Genetics	Online Portal	
	Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test	Invitae	Online Portal	Can add on genes for secondary newborn screening targets (*ETFA, ETFB, ETFDH, CPT2, SLC25A20*) or reflex these genes free of charge within 90 days
Wilson Disease	*ATP7B* single gene test	Blueprint Genetics	Online Portal	
Nephrology				
Adult tubulointerstitial kidney disease	Renal Malformation Panel + *UMOD* (flex)	Blueprint Genetics	Online Portal	
Alport syndrome	Alport Syndrome Panel	Blueprint Genetics	Online Portal	
Atypical hemolytic uremic syndrome	Hemolytic Uremic Syndrome Panel	Blueprint Genetics	Online Portal	
Bartter syndrome	Bartter Syndrome Panel	Blueprint Genetics	Online Portal	

Branchio-Oto-Renal Syndrome	Branchio-Oto-Renal (BOR) Syndrome Panel	Blueprint Genetics	Online Portal	
Ciliopathies	Ciliopathy Panel	Blueprint Genetics	Online Portal	
Congenital Abnormalities of the Kidney and Urinary Tract	Congenital Abnormalities of the Kidney and Urinary Tract (CAKUT) Panel	Prevention Genetics	Website Click on “log in/sign up myPrevent” to access online portal	
Cystinosis	*CTNS* single gene test	Blueprint Genetics	Online Portal	
Focal segmental glomerulosclerosis, Thin basement membrane disease	Any nephrology panel	Blueprint Genetics	Online Portal	
Gitelman syndrome	Bartter Syndrome Panel + *HNF1B* (flex)	Blueprint Genetics	Online Portal	Per KDIGO (2017), molecular testing for Gitelman syndrome should at minimum include *SLC12A3, CLCNKB*, and *HNF1B*. To order these genes, order the Bartter Syndrome Panel through Blueprint Genetics. Add on *HNF1B* and remove non-relevant genes.
Hypomagnesemia	Hypomagnesemia Panel	Blueprint Genetics	Online Portal	
Nephrolithiasis	Nephrolithiasis Panel	Blueprint Genetics	Online Portal	
Nephrotic Syndrome	Nephrotic Syndrome Panel	Blueprint Genetics	Online Portal	
Polycystic kidney disease, adult onset	Polycystic Kidney Disease Panel	Blueprint Genetics	Online Portal	
Polycystic kidneys; young adult/child	Cystic Kidney Disease Panel	Blueprint genetics	Online Portal	
Neurology				
Adrenoleukodystrophy	*ABCD1* single gene test	Blueprint Genetics	Online Portal	

Amyotrophic lateral sclerosis	Amyotrophic Lateral Sclerosis Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *SOD1* and *C9orf72* targeted sequence analysis *ATXN2* repeat expansion testing
Amyotrophy, hereditary neuralgic	*SEPT9* single gene test	Blueprint Genetics	Online Portal	
Ataxia	Ataxia Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Spinocerebellar ataxia types 1, 2, 3, 6, 7, 8, and 17, DRPLA, Friedreich ataxia, Fragile X-associated tremor/ataxia syndrome
Autism/Intellectual Disability	Autism/ID Xpanded Panel	GeneDx	Online Portal	**Parental blood samples are required for this test, and should be collected at the time you submit your funding request.**
Basal ganglia calcification, idiopathic	Idiopathic Basal Ganglia Calcification Panel	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	
Brain malformations	Comprehensive Brain Malformations Panel	GeneDx	Online Portal	
Charcot-Marie-Tooth	Charcot-Marie-Tooth Neuropathy Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Charcot-Marie-Tooth disease types 1A, 1B, and X
Congenital myasthenic syndrome	Congenital Myasthenic Syndromes Panel	Blueprint Genetics	Online Portal	
Dementia	Dementia Panel	Blueprint Genetics	Online Portal	
Dystonia	Dystonia Panel	Blueprint Genetics	Online Portal	

Dystonia, myoclonic	*SGCE* single gene test	Invitae	Online Portal	
Epilepsy	Comprehensive Epilepsy Panel (or sub-panel)	Blueprint Genetics	Online Portal	
Facioscapulohumeral muscular dystrophy, type 2	*SMCHD1* single gene test	Blueprint Genetics	Online Portal	Testing for Facioscapulohumeral muscular dystrophy type 1 must be performed first, if relevant. This testing is performed at the Children's Hospital of Eastern Ontario, and funded by the Calgary Molecular Genetics Lab. Please contact the lab genetic counsellors at 403-955-3097 with any questions.
Familial hemiplegic migraine	Migraine Panel	Blueprint Genetics	Online Portal	
Hereditary spastic paraplegia	Spastic Paraplegia Panel	Blueprint Genetics	Online Portal	
Holoprosencephaly	Holoprosencephaly Panel	Blueprint Genetics	Online Portal	
Intellectual disability, X-linked	X-linked Intellectual Disability Panel	Blueprint Genetics	Online Portal	
Joubert syndrome	Joubert Syndrome Panel	Blueprint Genetics	Online Portal	
Leukodystrophy	Leukodystrophy and Leucoencephalopathy panel	Blueprint Genetics	Online Portal	
Limb girdle muscular dystrophy	*DYSF* single gene test	Blueprint Genetics	Online Portal	
Malignant hyperthermia	Invitae Malignant Hyperthermia Susceptibility Panel	Invitae	Online Portal	
Microcephaly and/or pontocerebellar hypoplasia	Microcephaly and Pontocerebellar Hypoplasia Panel	Blueprint Genetics	Online Portal	

Muscular dystrophy/myopathy	Comprehensive Muscular Dystrophy / Myopathy Panel	Blueprint Genetics	Online Portal	
Myotonia and paramyotonia congenita	Invitae Myotonia and Paramyotonia Congenita Panel	Invitae	Online Portal	
Myotonia congenita	*CLCN1* single gene test	Blueprint Genetics	Online Portal	
Neurodegeneration with brain iron accumulation	Invitae Neurodegeneration with Brain Iron Accumulation Panel	Invitae	Online Portal	
Neuronal migration disorders	Neuronal Migration Disorder Panel	Blueprint Genetics	Online Portal	
Parkinson disease/parkinsonism	Parkinson Disease Panel	Blueprint Genetics	Online Portal	
Periodic paralysis	Periodic Paralysis Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *CACNA1S* and *SCN4A* targeted sequence analysis
SCN9A-related disorders	*SCN9A* single gene test	Blueprint Genetics	Online Portal	
Spinal muscular atrophy	Spinal Muscular Atrophy Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *SMN1* MLPA
Spinocerebellar ataxia	Repeat expansion - *ATXN10* (SCA10), *PPP2R2B* (SCA12), *BEAN1* (SCA31), *NOP56* (SCA36)	Centogene	Online Portal	In-house testing must be performed first, if relevant: Spinocerebellar ataxia types 1, 2, 3, 6, 7, 8, and 17
	Spinocerebellar ataxia (types 10, 12)	Center for Human Genetics	Requisition form	

Ophthalmology				
Achromatopsia	Achromatopsia Panel	Blueprint Genetics	Online Portal	
Cone rod dystrophy	Cone Rod Dystrophy Panel	Blueprint Genetics	Online Portal	
Congenital cataracts	Cataract Panel	Blueprint Genetics	Online Portal	
Congenital stationary night blindness	Congenital Stationary Night Blindness Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: *CACNA1F* targeted sequence analysis
Corneal dystrophy	Corneal Dystrophy Panel	Blueprint Genetics	Online Portal	
Ectopic Lentis	Ectopia Lentis Panel	Blueprint Genetics	Online Portal	
Glaucoma	Glaucoma Panel	Blueprint Genetics	Online Portal	
Leber congenital amaurosis	Leber Congenital Amaurosis Panel	Blueprint Genetics	Online Portal	
Macular dystrophy	Macular Dystrophy Panel	Blueprint Genetics	Online Portal	
Microphthalmia/anopthalmia	Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel	Blueprint Genetics	Online Portal	
Neuro-Ophthalmology Panel	Neuro-Ophthalmology Panel	Blueprint Genetics	Online Portal	
Optic atrophy	Optic Atrophy Panel	Blueprint Genetics	Online Portal	Can be ordered concurrently with mtDNA testing for Leber hereditary optic neuropathy
Retinal dystrophy	Retinal Dystrophy Panel	Blueprint Genetics	Online Portal	
Retinitis pigmentosa	Retinitis Pigmentosa Panel	Blueprint Genetics	Online Portal	
Septo-Optic Dysplasia	Septo-Optic Dysplasia Panel	Blueprint Genetics	Online Portal	
Usher syndrome	Usher Syndrome Panel	Blueprint Genetics	Online Portal	
Vitreoretinopathy	Vitreoretinopathy Panel	Blueprint Genetics	Online Portal	

GRC Established Testing Menu

<table>
<tr><th>Prenatal</th><th></th><th></th><th></th><th></th></tr>
<tr><td>Arthrogryposis</td><td>Arthrogryposes Panel</td><td>Blueprint Genetics</td><td>Online Portal</td><td>A maternal blood sample is required for maternal cell contamination studies to be performed at either the Calgary or Edmonton Molecular Genetics Lab.</td></tr>
<tr><td>Ashkenazi Jewish carrier testing
(for couples where both individuals are of Ashkenazi Jewish descent)</td><td>Targeted Variant Testing</td><td>Blueprint Genetics</td><td>Online Portal</td><td>
<table>
<tr><th>Gene</th><th>HGVSc</th><th>HGVSp</th><th>Transcript</th></tr>
<tr><td>HEXA</td><td>c.1421+1G>C</td><td>N/A</td><td>NM_000520.5</td></tr>
<tr><td>HEXA</td><td>c.1274_1277 dupTATC</td><td>p.(Tyr427Ilefs*5)</td><td>NM_000520.5</td></tr>
<tr><td>HEXA</td><td>c.805G>A</td><td>p.(Gly269Ser)</td><td>NM_000520.5</td></tr>
<tr><td>HEXA</td><td>c.1073+1G>A</td><td></td><td>NM_000520.5</td></tr>
<tr><td></td><td></td><td></td><td></td></tr>
<tr><td>ASPA</td><td>c.693C>A</td><td>p.(Tyr231*)</td><td>NM_000049.2</td></tr>
<tr><td>ASPA</td><td>c.854A>C</td><td>p.(Glu285Ala)</td><td>NM_000049.2</td></tr>
<tr><td></td><td></td><td></td><td></td></tr>
<tr><td>IKBKAP</td><td>c.2204+6T>C</td><td>N/A</td><td>NM_003640.3</td></tr>
<tr><td>IKBKAP</td><td>c.2087G>C</td><td>p.(Arg696Pro)</td><td>NM_003640.3</td></tr>
<tr><td></td><td></td><td></td><td></td></tr>
<tr><td>FANCC</td><td>c.456+4A>T</td><td></td><td>NM_000136.2</td></tr>
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</td></tr>
<tr><td rowspan="3">Familial variant(s)</td><td rowspan="3">Prenatal targeted testing*</td><td>Blueprint Genetics</td><td>Online Portal</td><td>Select this lab if they performed testing for the index patient

A maternal blood sample is required for maternal cell contamination studies to be performed at either the Calgary or Edmonton Molecular Genetics Lab.</td></tr>
<tr><td rowspan="2">Prevention Genetics</td><td>Website

Click on "log in/sign up myPrevent" to access online portal</td><td>Select this lab if they performed testing for the index patient. Consider using this lab if the index patient was tested at any other lab (coordinate a positive control sample to be sent to Prevention Genetics).

A maternal blood sample is required for maternal cell contamination studies to be performed at Prevention Genetics</td></tr>
<tr><td colspan="3">*Contact the GRC if you have questions about coordinating prenatal targeted testing through a different lab.</td></tr>
</table>

Fetal aneuploidy	Harmony Prenatal Test	Dynacare	Requisition form	For fetal aneuploidy testing, only funded in cases where an invasive procedure is contraindicated and pregnancy is at increased risk of aneuploidy
	Non-invasive prenatal screening (NIPS)	Invitae	Online Portal	
Fetal sex determination	Harmony Prenatal Test	Dynacare	Requisition form	For cases where there is a family history of an x-linked condition and fetal sexing results may prevent the need for an invasive procedure
	Non-invasive prenatal screening (NIPS)	Invitae	Online Portal	
Increased nuchal translucency or nuchal fold	Noonan Syndrome Panel	Blueprint Genetics	Online Portal	**A maternal blood sample is required for maternal cell contamination studies** to be performed at either the Calgary or Edmonton Molecular Genetics Lab
Non-immune hydrops	Non-Immune Hydrops Fetalis Panel	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	**A maternal blood sample is required for maternal cell contamination studies** to be performed at Prevention Genetics
Skeletal dysplasia	Comprehensive Growth Disorders / Skeletal Dysplasias and Disorders Panel (or sub-panel)	Blueprint Genetics	Online Portal	**A maternal blood sample is required for maternal cell contamination studies** to be performed at either the Calgary or Edmonton Molecular Genetics Lab
Pulmonology				
Inherited pulmonary disease	Comprehensive Pulmonology Panel	Blueprint Genetics	Online Portal	

Interstitial pulmonary disease	Interstitial Lung Disease Panel	Blueprint Genetics	Online Portal	
Neonatal respiratory distress	Neonatal Respiratory Distress – Surfactant Dysfunction Panel	Blueprint Genetics	Online Portal	
Primary ciliary dyskinesia	Primary Ciliary Dyskinesia Panel	Blueprint Genetics	Online Portal	
Single Genes				
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL* single gene test	Blueprint Genetics	Online Portal	
Adrenoleukodystrophy	*ABCD1* single gene test	Blueprint Genetics	Online Portal	
Amyotrophy, hereditary neuralgic	*SEPT9* single gene test	Blueprint Genetics	Online Portal	
Biotinidase deficiency	*BTD* single gene test	Blueprint Genetics	Online Portal	
Carnitine deficiency, systemic primary	*SLC22A5* single gene test	Blueprint Genetics	Online Portal	
CHARGE syndrome	*CHD7* single gene test	Blueprint Genetics	Online Portal	
Congenital hypothyroidism	*TRH* Targeted Del/Dup	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	This test is designed to detect a *TRH* deletion in the Hutterite population
Cystinosis	*CTNS* single gene test	Blueprint Genetics	Online Portal	
Dystonia, myoclonic	*SGCE* single gene test	Invitae	Online Portal	

Facioscapulohumeral muscular dystrophy, type 2	*SMCHD1* single gene test	Blueprint Genetics	Online Portal	Testing for Facioscapulohumeral muscular dystrophy type 1 must be performed first, if relevant. This testing is performed at the Children's Hospital of Eastern Ontario, and funded by the Calgary Molecular Genetics Lab. Please contact the lab genetic counsellors at 403-955-3097 with any questions.
Familial Mediterranean fever	*MEFV* sanger sequencing	BC Molecular Genetics Laboratory	Requisition form	
Galactosemia	*GALT* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant (**This does not apply to urgent cases**): *GALT* targeted sequence analysis
Gilbert syndrome	*UGT1A1* single gene test	Blueprint Genetics	Online Portal	
Glutaric acidemia type 1	*GCDH* single gene test	Blueprint Genetics	Online Portal	
Hereditary angioedema	*SERPING1* single gene test	Blueprint Genetics	Online Portal	
Hunter syndrome	*IDS* single gene test	Blueprint Genetics	Online Portal	
Hurler syndrome	*IDUA* single gene test	Blueprint Genetics	Online Portal	
Hypophosphatemic rickets	*PHEX* single gene test	Blueprint Genetics	Online Portal	
Incontinentia pigmenti	*IKBKG* (NEMO) Gene Sequencing & Common Del/Dup	GeneDx	Online Portal	

Isovaleric acidemia	*IVD* single gene testing	Blueprint Genetics	Online Portal	
Legius syndrome	*SPRED1* single gene testing	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: NF1 sequencing and del/dup by NGS
Limb girdle muscular dystrophy	*DYSF* single gene test	Blueprint Genetics	Online Portal	
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant (**This does not apply to urgent cases**): ACADM targeted sequence analysis
Myotonia congenita	*CLCN1* single gene test	Blueprint Genetics	Online Portal	
Phenylketonuria	*PAH* single gene test	Blueprint Genetics	Online Portal	
Pompe disease	*GAA* single gene test	Blueprint Genetics	Online Portal	
Pseudoxanthoma elasticum	*ABCC6* single gene test	Blueprint Genetics	Online Portal	
Schwannomatosis	*LZTR1* single gene test	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Schwannomatosis Panel – *NF2, SMARCB1*
SCN9A-related disorders	*SCN9A* single gene test	Blueprint Genetics	Online Portal	
Thyroid hormone resistance	*THRB* single gene test	Blueprint Genetics	Online Portal	
Tyrosinemia, type 1	FAH single gene test	Blueprint Genetics	Online Portal	
Very long chain acyl-CoA dehydrogenase deficiency	*ACADVL* single gene test	Blueprint Genetics	Online Portal	
Wilson disease	*ATP7B* single gene test	Blueprint Genetics	Online Portal	

Skeletal				
Amelogenesis Imperfecta and Dentinogenesis Imperfecta	Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	Blueprint Genetics	Online Portal	
Arthrogryposis	Arthrogryposes Panel	Blueprint Genetics	Online Portal	
Craniosynostosis	Craniosynostosis Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Craniosynostosis panel
Facial dysostosis	Facial Dysostosis and Related Disorders Panel	Blueprint Genetics	Online Portal	
Klippel-Feil syndrome	Klippel-Feil Syndrome Panel	Prevention Genetics	Website Click on "log in/sign up myPrevent" to access online portal	
Limb malformation	Limb Malformations Panel	Blueprint Genetics	Online Portal	
Osteopetrosis	Osteopetrosis and Dense Bone Dysplasia Panel	Blueprint Genetics	Online Portal	
Short stature, growth failure	Comprehensive Short Stature Syndrome Panel	Blueprint Genetics	Online Portal	
Skeletal dysplasia	Comprehensive Growth Disorders / Skeletal Dysplasias and Disorders Panel (or sub-panel)	Blueprint Genetics	Online Portal	

GRC Established Testing Menu

Vascular				
Hereditary angioedema	Hereditary Angioedema Panel	Invitae	Online Portal	
	SERPING1 single gene test	Blueprint Genetics	Online Portal	
Lymphatic malformations	Lymphatic Malformations and Related Disorders Panel	Blueprint Genetics	Online Portal	
Vascular malformations	Vascular Malformation Panel	Blueprint Genetics	Online Portal	In-house testing must be performed first, if relevant: Hereditary Hemorrhagic Telangiectasia/Arteriovenous Malformation Panel
Whole Exome Sequencing				
Various (Please refer to guidelines*) *Ordering restrictions apply	Whole Exome Sequencing (Trio, Duo, Patient only)	Blueprint Genetics	Online Portal	Trio testing is preferred. **Coordinate parental blood sample collection** at the time that you submit your funding request. **If your patient had a previous NGS panel test where sequencing was performed using an exome backbone, **expand to whole exome sequencing** may be funded through the lab that performed the NGS panel unless this lab is no longer a preferred lab. Please contact the GRC if you have any questions**
		GeneDx	Online Portal	
	XomeDxXpress (Trio preferred)	GeneDx	Online Portal	Trio testing is preferred. **Coordinate parental blood sample collection** at the time that you submit your funding request. **Only in cases where results are needed to inform treatment/management decisions within two weeks.**