



Whole Exome Sequencing (WES) Requisition

**ALBERTA PRECISION
LABORATORIES**

For detailed testing information, refer to
APL Test Directory: <http://ahsweb.ca/lab/apl-td-lab-test-directory>
APL Genetics & Genomics Website:
<http://ahsweb.ca/lab/if-lab-genetics-and-genomics>

Scanning Label or Accession # (lab only)

Patient	PHN	Expiry: _____	Date of Birth (dd-Mon-yyyy)		
	Legal Last Name		Legal First Name		Middle Name
	Alternate Identifier	Preferred Name	<input type="checkbox"/> Male <input type="checkbox"/> Non-binary	<input type="checkbox"/> Female <input type="checkbox"/> Prefer not to disclose	Phone
	Address		City/Town	Prov	Postal Code
Provider(s)	Authorizing Provider Name (last, first, middle)		Copy to Name (last, first, middle)		Copy to Name (last, first, middle)
	Address		Phone	Address	Address
	CC Provider ID	CC Submitter ID	Phone	Phone	
	Clinic Name		Clinic Name	Clinic Name	
Collection	Date (dd-Mon-yyyy)	Time (24 hr)	Location	Collector ID	

Genetic Counsellor/Clinic Contact Name				Phone
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Specimen

<input type="checkbox"/> Whole Blood in EDTA Tube	<input type="checkbox"/> Extracted DNA	<input type="checkbox"/> Fluid, amniotic*
<input type="checkbox"/> Tissue, chorionic villi*	<input type="checkbox"/> Cord blood*	<input type="checkbox"/> Other (specify): _____

**If specimen type is prenatal or cord blood, maternal specimen must be collected for maternal cell contamination studies*

Health Care Provider Important Information

- All sections of the requisition must be completed.
- By providing this requisition to the patient/family, the health care provider confirms that they have reviewed the pre-test counselling information (available on the Genetics & Genomics website) with the patient/family, and the patient/family consents to testing.
- Direct patient to take requisition to a local blood collection location to have blood specimen drawn

Billing Information: Must be completed if the patient does not have a valid Alberta Personal Health Number. Genetic testing is not covered by inter-provincial billing agreements. Alberta Precision Laboratories (APL) will bill a provincial medical services plan provided there is a letter of pre-approval received with the requisition or Institutional Billing information provided below. By completing the Institutional Billing section, the health care provider confirms they have obtained any necessary pre-approval. For patient pay, contact the testing laboratory.

Institutional Billing Information (if pre-approval letter not attached)

Address	
Contact Name (last, first)	
Phone	Fax

MGL Use Only

Patient Number	Family Number	Received	Quantity
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Last Name (Legal)	First Name (Legal)
PHN	

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Section I - Test Selection

Please refer to the APL test directory for ordering instructions and WES eligibility guidelines.

Section I. 1 - Test Type (select only one)

- Exome Analysis – Proband (complete Section I and II - Proband Order Questions)
- Exome Analysis – Family Member (complete Section I and III - Family Member Order Questions)
- Exome Re-analysis (complete Section I and IV - Exome Re-analysis Order Questions)
- Familial Variant – WES Follow-up (complete Section I.3 and V - Familial Variant Testing Order Questions)

Section I. 2 - Type of Analysis

- Singleton Duo Trio Quad Other _____

For Duo, Trio and Quad testing, provide demographic information for other family members

Name	PHN	Relationship to Proband

Section I. 3 - Test Details

Is RUSH testing needed? Yes (please select reason below) ▼ No

Results will alter the **immediate** management/treatment of this patient (specify): _____

Results will impact an **ongoing pregnancy** (provide gestational age or EDD, and procedure date if applicable)

Gestational Age/EDD	Procedure Date (dd-Mon-yyyy)
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If expedited testing is required, please provide a target date (dd-Mon-yyyy) _____

Has this individual received a blood product in the previous three months?

Yes ► Type of blood product _____ No

Has this individual received a bone marrow transplant?

Yes ► (blood is an incompatible specimen type) _____ No

Sex Assigned at Birth Female Male Unknown

Section I. 4 - Secondary Findings, Data Sharing & Research Consent

Has this individual consented to the reporting of secondary findings?

Yes No

Has this individual consented to share their coded data with Clinical Knowledge Networks approved in Canada?

Yes No

Has this individual consented to be contacted about future research opportunities?

Yes No



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Section II – Proband Order Questions

Section II. 1 - WES Criteria *(select all that apply)*

A genetic etiology is the most likely explanation for the patient’s phenotype, supported by a clinical presentation which includes:

- Severe to profound intellectual disability in the absence of known risk factors

OR at least **two** of the following:

- Moderate to severe developmental or functional impairment
- Multisystem involvement
- Progressive clinical course
- Differential diagnosis which includes two or more well-defined conditions requiring evaluation by multiple targeted gene panels
- Suspected severe undiagnosed genetic syndrome for which multiple family members are also affected, or where parents are consanguineous
- Patient does not meet the above criteria. Please explain:

Clinical WES is predicted to be less cost-effective or to have a low diagnostic yield in the following scenarios:

- The patient’s phenotype is highly specific to a known condition or appears to fit into a single clinical category for which a more cost-effective phenotype-driven panel is available
- The patient had an uninformative comprehensive gene panel reported within the past 2 years which included virtually all known genes related to their clinical indication
- A likely non-genetic etiology has been identified to explain the patient’s symptoms

If any of the above scenarios apply, please provide additional justification for requesting WES:

Section II. 2 - Proband Clinical Features

Detailed clinical information will aid in identification of relevant variants and diagnosis.

Relevant clinical information, previous testing, and Human Phenotype Ontology (HPO) terms must be provided. Clinical consult letters that are not available in Connect Care should be provided to Genetics & Genomics.



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Section II. 3 – Family History

Other family members of your patient previously testing in the Molecular Genetics Lab?

Yes ▼ No

Family member name(s)	MGL reference number(s)
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Relevant family history:

Is there known consanguinity?

Yes ▼ No

Please specify relationships: _____

Ethnicity/Ancestry

Section III – Family Member Order Questions

Section III. 1 – Relationship Information

Relationship to proband Mother Father Child Sibling Other _____

Proband name	PHN
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Section III. 2 – Family Member Clinical Information

Is this individual affected?

Yes No Partially Unknown

List relevant clinical features and any previous testing:

Ethnicity/Ancestry

Section IV– Exome Re-analysis Order Questions

Original Exome Case ID _____

Has the patient had a previous exome reanalysis? Yes No

Has it been at least 2 years since the original exome analysis was reported? Yes No

Has there been a significant change in the patient's clinical presentation? Yes ▼ No

Please provide updated phenotypic and family history information:

Human Phenotype Ontology (HPO) terms:



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Section IV– Exome Re-analysis Order Questions *(continued)*

If the patient is not currently meeting reanalysis criteria, please provide additional information regarding the request for reanalysis:

Section V - Familial Variant – WES Follow-up *(for variants identified on exome analysis in a relative)*

Reason for Testing *(select only one)*:

- Confirmation of diagnosis *(patient has signs or symptoms of the disease/disorder)*
- Presymptomatic or Predictive testing *(patient does not presently have symptoms; positive family history of condition)*
- Carrier testing *(no symptoms of classical disease. At risk of being a carrier of a recessive disorder)*
- Variant segregation
- Other - specify: _____

Was the proband tested in an Alberta Molecular Genetics Lab?

Yes ▼

Proband name / PHN	Exome Case ID	Relationship to Proband

No ► For index patients tested at an external lab, familial variant testing at the same external lab is typically preferred. Please contact the Genetic Resource Centre with any questions.

Relevant clinical and family history information:

Variants to be tested:

Gene	Transcript, cDNA change, protein change
1.	
2.	
3.	
4.	
5.	