ALBERTA PRECISION LABORATORIES
Leaders in Laboratory Medicine

Pre-test Counselling Information: Genetics and Genomics Testing

The following points are recommended to be discussed with the patient and/or guardian(s) prior to ordering testing through Genetics and Genomics (North and South).

1. Sample(s) (for example, blood, amniotic fluid, chorionic villi, cheek swabs) will be collected and nucleic acids (typically DNA) will be extracted. Testing will be performed in one of the provincial Genetic and Genomics Laboratories or in another accredited laboratory approved by Genetics and Genomics. After testing has been completed, any remaining nucleic acid may be retained in the laboratory and used for quality assurance or test development purposes.

2. The accuracy, implications and limitations of testing should be reviewed prior to testing.

3. Genetic testing is limited to the requested test and cannot rule out all genetic conditions or pathogenic (disease causing) variants.

4. Current testing may not include all genes associated with a given condition and/or may not be able to detect all pathogenic variants associated with the suspected condition. Patients should be managed based on their diagnosis and clinical presentation.

5. The correct clinical diagnosis in this patient, or affected family members, is important for accurate, clinically relevant genetic test results. The laboratory assumes and tests based upon the diagnosis as stated on the laboratory requisition. Testing may be refused if insufficient clinical information is provided.

6. Testing may identify a genetic variant for which there is currently insufficient evidence to conclude that it is either disease-causing or benign (called a variant of uncertain significance). Such variants cannot be used to alter the clinically established risk of disease.

7. Genetic testing is interpreted most appropriately in the context of the family. It is therefore essential that we receive a comprehensive and accurate pedigree (family tree) that indicates all other known affected individuals or carriers of the condition and includes the names of all close relatives. When the genotypes (specific pathogenic variants) of individuals are known, these should be indicated on the pedigree. Requests for testing may be refused if a suitable pedigree is not provided.

8. Genetic testing may reveal non-paternity or an undisclosed adoption. Accurate test and clinically relevant results depend on knowing the correct relationships between family members. The laboratory assumes and tests based upon the information on family relationships provided with the test requisition.

9. Testing and interpretation of results is based on current knowledge. It is the patient and/or physician's responsibility to periodically seek up-dated information especially before any reproductive decisions are made. Patients are responsible for keeping their physicians informed of address changes and new medical and family history information.

10. Improved or additional testing may become available either because of changes in laboratory techniques or because of new information regarding the genetic cause of the condition(s). It is the responsibility of the patient's physician(s) to initiate up-dated testing. The laboratory will not undertake new testing unless initiated by the patient's physician.

11. Genetic testing may reveal incidental findings which are not related to the referral indication (example: carrier status for recessive conditions). Disease causing variants will be reported if detected regardless of the reason for referral.

12. Confidentiality will be maintained as required by the applicable provincial health privacy laws, Alberta Health Services policies and accreditation requirements of the College of Physicians and Surgeons of Alberta. Results will most commonly be reported via a laboratory information system and form part of the patient's health care record, including their electronic health record. For a limited number of adult onset conditions, results will not be visible electronically. Results may be used anonymously by the laboratory to help interpret test results for other family members. Information that a nucleic acid sample has been banked may be shared to prevent repeat blood drawing.

13. Anonymous samples of DNA may be given to research centers, with appropriate research ethics board approval, to help further research.

14. Participation in genetic testing is completely voluntary. Patients may withdraw consent or request that their nucleic acid sample be destroyed at any time.

15. Genetic test results may result in some forms of discrimination (insurance, employment or other) as they form part of the patient's medical record and may be assessed when informed consent by the patient is given to a third party.

16. For some conditions, condition specific information is available at Genetics and Genomics and the laboratory recommends that ordering providers review this information prior to initiating testing.