

DATE:	12 November 2024
TO:	All Zones
FROM:	Genetics and Genomics, Molecular Genetics Laboratory North
RE:	Ordering criteria for the Fanconi Anemia and DNA Repair Disorders Panel

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Key Message

- Effective immediately, the Molecular Genetics Laboratory – North (MGL) has implemented clinical ordering criteria for the Fanconi Anemia and DNA Repair Disorders Panel.

Background

- The Fanconi Anemia and DNA Repair Disorders Panel has replaced chromosome breakage studies as the first-tier test for patients suspected of having a DNA repair disorder.
- Patients who meet the following criteria are eligible:
 - For Fanconi anemia, patients with 2 or more clinical features from at least 2 of the following categories:

Physical Features	Laboratory Findings	Cancer
Short stature	Progressive bone marrow failure not related to cancer treatment	Cancer diagnosed < 5 years of age
Abnormal skin pigmentation	Aplastic anemia	Solid tumour diagnosed before age 50
Skeletal malformations	Myelodysplastic syndrome or AML	
Microcephaly	Macrocytosis	
Ophthalmic anomalies	Cytopenia	
Genitourinary tract anomalies	Increased fetal hemoglobin	
	Inordinate toxicities from chemotherapy or radiation	

- Patients with a personal history suggestive of ataxia telangiectasia, Bloom syndrome, Nijmegen breakage syndrome, or RECQL4-related disorders.

How this will impact you

- The Fanconi Anemia and DNA repair disorder panel is the first-tier testing for patients suspected of having a DNA repair disorder.
- Cytogenetic chromosome breakage analysis may be available in some situations, such as functional testing for variants of uncertain clinical significance identified by the panel. Please contact the Cytogenetics South Laboratory at (403) 955-2886 for additional information.



Action Required

- Refer to the test directory for current information on the DNA Repair Disorder Panel Alberta Precision Laboratories | Lab Services ([Alberta Precision Laboratories | Lab Services \(albertahealthservices.ca\)](https://albertahealthservices.ca))
- To order in Connect Care select **Fanconi Anemia and DNA Repair Disorders Panel LAB2595**
- To order using a requisition complete a Molecular Genetics Cancer and Endocrine Next Generation Sequencing Requisition. The test can be found in Section VI – Cancer and Endocrine NGS Panel Requests, under Hematological Cancers select Fanconi Anemia and DNA Repair Disorders Panel [Molecular Genetics Laboratory Cancer and Endocrine NGS Requisition \(albertahealthservices.ca\)](https://albertahealthservices.ca)
- Clinical indications for testing must be provided. Failure to provide clinical criteria may result in test delays or cancellations.

Effective Immediately

Questions/Concerns

- G&G North Genetic Counsellors at 780-407-1015

Approved by

- Dr. Dennis Bulman, Medical/Scientific Director, Genetics and Genomics, APL
- Dr. Carolyn O'Hara, Chief Medical Laboratory Officer (Interim), APL