DISEASE-INVOLVED AND GERMLINE SAMPLES

This test requires submission of both a disease-involved sample (somatic sample) and non-disease-involved sample (germline comparator sample) from the patient. Each submitted sample must be labeled with the full patient's name and at least one other unique patient identifier (i.e. DOB, MRN).

Disease-Involved Sample(s)	Germline Sample
Tumor percentage for malignant conditions: The disease-involved sample must contain a minimum of **20% tumor or blast content for single-nucleotide and small insertion-deletion variant resolution** OR a minimum of **60% tumor or blast content for sensitive resolution of copy number variation (CNV) and loss of heterozygosity (LOH)** to enable interpretation and reporting. Sensitivity in calling CNV and LOH will be limited, and at times, assay resolution of these events will preclude interpretation and reporting of CNV and LOH if the submitted specimen contains less than 60% disease-content. *Specimen types:* One of the following types of specimens is required: • Frozen tissue (30-50 mg). (This type is preferred for oncology cases.) • Fresh tissue (30-50 mg). • FFPE tissue block. (FFPE is preferred for vascular lesions.) • 10-15 FFPE scrolls (5-10 microns thick) and adjacent H&E slide. • Involved bone marrow (4 mL EDTA). • Involved whole blood (4 mL EDTA). Acid decalcified samples are not acceptable for this testing. Samples processed with EDTA decalcification can be attempted. *Disease timepoints:* Multiple disease timepoints may be submitted for testing. Additional charges will be applied.	*Specimen type:* One of the following types of specimens is required: • Whole blood (4 mL EDTA). (This type is preferred.) • Saliva (two collection tubes). • Buccal swabs (four swabs). Please contact the laboratory if the patient has a history of allogenic bone marrow transplantation to discuss options for the comparator normal sample.

Consent

Genetic counseling and submission of an informed consent form is required. If the informed consent form is incomplete, this will delay the initiation of testing. When a parent or legal guardian completes the consent form on behalf of their child, the parent's or guardian's own initials should be used (not the initials of their child). Each item on the consent form requires a response (i.e. initials or signatures) as indicated; missing initials/signatures constitute an incomplete consent form.

Submission Checklist

Please ensure all the items below are completed and ready before submission.

✓ Completed	REQUIRED MATERIAL
	Disease-Involved Sample
	Germline Sample
	Informed Consent
	Requisition or Epic order
	Pathology Report (for EVERY disease timepoint)