Reporting Title: Whole Genome Sequencing
Performing Location: Rochester

Ordering Guidance:
The American College of Medical Genetics and Genomics recommends that whole genome sequencing be considered as a first-tier or second-tier test for patients with one or more congenital anomalies, or developmental delay or intellectual disability with onset prior to age 18 years.(1)

If a specific diagnosis is suspected, single gene or panel testing may be a more appropriate first-tier testing option.

This test is for affected patients (probands) only. For family member specimens being sent as comparators, order CMPRG / Family Member Comparator Specimen for Genome Sequencing, Varies. If this test is ordered on a family member comparator, this test will be canceled and CMPRG performed as the appropriate test.

This test is not appropriate for identification of somatic variants in solid tumors or other malignancies. Multiple oncology (cancer) gene panels are available. For more information see Oncology Somatic NGS Testing Guide. If testing for other malignancies is needed, contact the laboratory for test selection guidance.

This testing does not provide genotyping of patients for pharmacogenomic purposes. For an assessment for genes with strong drug-gene associations, order PGXQP / Focused Pharmacogenomics Panel, Varies.

Targeted testing for familial variants (also called site-specific or known variant testing) is available for variants identified by this test. See FMTT / Familial Variant, Targeted Testing, Varies.

Prenatal specimens (amniocentesis or chorionic villi) are not currently accepted for this test.

Additional Testing Requirements:
To order whole genome sequencing for the patient and the family member comparator specimens, see the following steps:
1. Order this test (WGSDX) on the patient (proband).
2. Order CMPRG / Family Member Comparator Specimen for Genome Sequencing, Varies on all family members' specimens being submitted as comparators.
 a. When available, the patient's biological mother and biological father are the preferred family member comparators.
 b. If one or both of the patient's biological parents are not available for testing, specimens from other first-degree relatives (siblings or children) can be used as comparators. Testing typically includes up to two family member comparators. Contact the laboratory at 800-533-1710 for approval to send specimens from other relatives or to send the patient and three first-degree relatives (quad).
 c. The cost of analysis for family member comparator specimens is applied to the patient's (proband's) test. Family members will not be charged separately.
3. Collect patient (proband) and family member specimens. Label specimens with full name and birthdate. Do not label family members' specimens with the proband's name.
4. For each family, complete the following portions of the Whole Genome Sequencing: Ordering Checklist. A separate form is not needed for each family member.
 a. Patient Information is required for all clients
 b. Informed Consent is required for New York State clients
 c. If the patient wishes to opt-out of receiving secondary findings or change the DNA storage selection, select the appropriate boxes in the Informed Consent section.

5. Attach clinic notes from specialists relevant to patient's clinical features, if available.
6. Attach pedigree, if available.
7. Send paperwork to the laboratory along with the specimens. If not sent with the specimens, fax a copy of the paperwork to 507-284-1759, Attn: WGS Genetic Counselors.

For more information see Whole Exome and Genome Sequencing Information and Test Ordering Guide.

Shipping Instructions:
Specimen preferred to arrive within 96 hours of collection.

Necessary Information:
Whole Genome Sequencing: Ordering Checklist is required for all patients, and Informed Consent is required for New York clients. Fill out one form for the family and send with the specimens.

Specimen Requirements:
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
Additional Information: If a cord blood specimen is received, MATCC / Maternal Cell Contamination, Molecular Analysis, Varies will be performed at an additional charge; maternal blood sample is required.

Specimen Minimum Volume:
1 mL

Forms:
1. Whole Genome Sequencing: Ordering Checklist is required
2. New York Clients-Informed consent is required and is included in the above form. Document on the request form or electronic order that a copy is on file.
3. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
614364	Interpretation	Alphanumeric		69047-9
614464	Specimen	Alphanumeric		31208-2
614317	Source	Alphanumeric		31208-2
614473	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental

CPT Code Information:

81425-Patient only
81425, 81426-Patient and one family member comparator sample (duo) (as appropriate)
81425, 81426 x 2-Patient and two family member comparator samples (trio or non-traditional trio) (as appropriate)
81425, 81426 x 3-Patient and three family member comparator samples (quad) (as appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
G227	Number of Comparators for WGSDX			No	No (Bill Only)
MATCC	Maternal Cell Contamination, B			No	Yes
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.