Reporting Title: Congenital Heart Disease Gene Panel
Performing Location: Rochester

Ordering Guidance:
Chromosomal microarray is often used as a first-tier test in the setting of congenital heart disease. If chromosomal microarray testing is desired, order either CMACB / Chromosomal Microarray, Congenital, Blood or CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling.

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Additional Testing Requirements:
All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen as this must be a different order number than the prenatal specimen.

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

Necessary Information:
Prior Authorization is available, but not required, for this test. If proceeding with the prior authorization process, submit the required form with the specimen.

Specimen Requirements:
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

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.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient
Additional information:
1. If amniotic fluid or nonconfluent cultures are received, CULAF / Culture for Genetic Testing, Amniotic Fluid will be added at an additional charge.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated
Additional Information:
1. If nonconfluent cultures are received, CULFB / Fibroblast Culture for Biochemical or Molecular Testing will be added at an additional charge.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Additional Information: All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Minimum Volume:

Blood: 1 mL; Other specimen types: See Specimen Required

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing (Spanish) (T826)
2. Congenital Heart Disease Genetic Testing Patient Information
3. Congenital Heart Disease Gene Panel (CHDGG) Prior Authorization Ordering Instructions
4. If not ordering electronically, complete, print, and send a Cardiovascular Test Request (T724) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No
CULAF	CG767	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
617198	Test Description	Alphanumeric		62364-5
617199	Specimen	Alphanumeric		31208-2
617200	Source	Alphanumeric		31208-2
617201	Result Summary	Alphanumeric		50397-9
617202	Result	Alphanumeric		82939-0
617203	Interpretation	Alphanumeric		69047-9
617204	Additional Results	Alphanumeric		82939-0
617205	Resources	Alphanumeric		99622-3
617206	Additional Information	Alphanumeric		48767-8
617207	Method	Alphanumeric		85069-3
617208	Genes Analyzed	Alphanumeric		48018-6
617209	Disclaimer	Alphanumeric		62364-5
617210	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental

CPT Code Information:

81404
81405 x 3

81406 x 6
81407 x 3
81408
81479
81479 (if appropriate for government payers)
81265-Maternal cell contamination (if appropriate)
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
88235-Amniotic Fluid culture (if appropriate)
88240-Cryopreservation (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
_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®
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

Test Definition: CHDGG

Congenital Heart Disease Gene Panel, Varies

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

Reference Values:

An interpretive report will be provided