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**Reporting Title:** OI and Bone Fragility Gene Panel**Performing Location:** Rochester**Ordering Guidance:**

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

**Prenatal Specimens**

**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. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.

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. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Molecular Testing, Chorionic Villi/Products of Conception.

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.

**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. [Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information](#)

3. [Osteogenesis Imperfecta and Bone Fragility Gene Panel \(OIBFG\) 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		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
617408	Test Description	Alphanumeric		62364-5
617409	Specimen	Alphanumeric		31208-2

617410	Source	Alphanumeric		31208-2
617411	Result Summary	Alphanumeric		50397-9
617412	Result	Alphanumeric		82939-0
617413	Interpretation	Alphanumeric		69047-9
617414	Additional Results	Alphanumeric		82939-0
617415	Resources	Alphanumeric		99622-3
617416	Additional Information	Alphanumeric		48767-8
617417	Method	Alphanumeric		85069-3
617418	Genes Analyzed	Alphanumeric		48018-6
617419	Disclaimer	Alphanumeric		62364-5
617420	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

- 81406 x 2
- 81408 x 2
- 81479
- 81479 (if appropriate for government payers)
- 88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
- 88240-Cryopreservation (if appropriate)
- 88235-Amniotic fluid culture (if appropriate)
- 81265-Maternal cell contamination (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
_STR1	Comp Analysis using STR (Bill only)	1	81265	No	No, (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)	1	81266	No	No, (Bill only)
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
MATCC	Maternal Cell Contamination, B	1	81265	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	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
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	53291	Released By	Alphanumeric		18771-6
MATCC	55150	Method	Alphanumeric		85069-3

Reference Values:

An interpretive report will be provided.