

Test Definition: RSCGP

Nephrocalcinosis, Nephrolithiasis, and Renal Electrolyte Imbalance Gene Panel, Varies

Reporting Title: Renal Stone/Electrolyte Gene Panel **Performing Location:** Rochester

Ordering Guidance:

A next-generation sequencing (NGS) panel of the 6 genes associated with Bartter syndrome, a rare renal salt-wasting disorder, is available. See RBART / Bartter Syndrome Gene Panel, Varies. It is inappropriate to order both RBART and this test on the same patient because the genes on the RBART panel are included on this panel.

Testing for CASR is available individually. See CASRG / CASR Full Gene Sequencing with Deletion/Duplication, Varies.

With a few exceptions, this panel is focused on conditions where the primary phenotype is impaired osmoregulation that may result in secondary extrarenal symptoms. If interested in testing for syndromic disorders that are associated with kidney disease but feature broader clinical phenotypes and multisystem involvement, see NEPHP / Comprehensive Nephrology Gene Panel, Varies.

Targeted testing for familial variants (also called site-specific or known mutations/variants 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.

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.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

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.
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
Invert several times to mix blood.
Send whole specimen in original tube. Do not aliquot.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Minimum Volume:

1 mL

Forms:

1.New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file.



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The following documents are available: -Informed Consent for Genetic Testing (T576) -Informed Consent for Genetic Testing-Spanish (T826) 2. Hereditary Renal Genetic Testing Patient Information (T918)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
618129	Test Description	Alphanumeric		62364-5
618130	Specimen	Alphanumeric		31208-2
618131	Source	Alphanumeric		31208-2
618132	Result Summary	Alphanumeric		50397-9
618133	Result	Alphanumeric		82939-0
618134	Interpretation	Alphanumeric		69047-9
618135	Additional Results	Alphanumeric		82939-0
618136	Resources	Alphanumeric		99622-3
618137	Additional Information	Alphanumeric		48767-8
618138	Method	Alphanumeric		85069-3
618139	Genes Analyzed	Alphanumeric		48018-6
618140	Disclaimer	Alphanumeric		62364-5
618141	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81404 x 4 81405 x 2 81406 x 8 81407 x 2 81479 81479 (if appropriate for government payers)



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Reference Values:

An interpretive report will be provided.