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MAYO CLINIC LABORATORIES

Test Definition: CKDGP

Cystic Kidney Disease Gene Panel, Varies

Overview

Useful For

Providing a genetic evaluation for patients with a personal or family history of cystic kidney disease

Establishing a diagnosis of hereditary cystic kidney disease

Reflex Tests

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

Genetics Test Information

This test utilizes next-generation sequencing to detect single nucleotide, deletion-insertion, and copy number variants in 45 genes associated with cystic kidney disease: *ALG8, ALG9, ANKS6, BICC1, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TRAF3IP1, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, WDR35, XPNPEP3*. See Targeted Genes and Methodology Details for Cystic Kidney Disease Gene Panel in Method Description for additional details.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for cystic kidney disease.

Testing Algorithm

For prenatal specimens only:
If an amniotic fluid specimen or nonconfluent cultures are received, amniotic fluid culture for a genetic test will be performed at an additional charge.

If a chorionic villi specimen is received, fibroblast culture for a genetic test will be performed at an additional charge.

For any prenatal specimen received, maternal cell contamination testing will be performed at an additional charge.

Special Instructions

- Informed Consent for Genetic Testing

- Informed Consent for Genetic Testing (Spanish)
- Hereditary Renal Genetic Testing Patient Information
- Targeted Genes and Methodology Details for Cystic Kidney Disease Gene Panel

Method Name
Sequence Capture and Amplicon-based Next-Generation Sequencing (NGS)

NY State Available
Yes

Specimen

Specimen Type
Varies

Ordering Guidance
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.

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.

Specimen Required
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. 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.

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. Hereditary Renal Genetic Testing Patient Information (T918)
3. If not ordering electronically, complete, print, and send a Renal Diagnostics Test Request (T830) with the specimen.

Specimen Minimum Volume

Blood: 1 mL; Amniotic fluid/CVS: See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information

Hereditary forms of cystic kidney disease have several underlying genetic etiologies and may present in childhood or adulthood, with or without extrarenal features. The two most common categories of hereditary cystic kidney disease are the ciliopathic disorders and the phakomatoses.(1)

Ciliopathic disorders causing cystic kidney disease include polycystic kidney disease (PKD), nephronophthisis (NPHP), and medullary cystic kidney disease (MCKD). The *PKD1* and *PKD2* genes cause the majority of autosomal dominant PKD (ADPKD), while the *GANAB* and *DNAJB11* genes are implicated in a minority of cases.(2,3) The *PKHD1* gene is the major gene associated with autosomal recessive PKD (ARPKD), while *DZIP1L* has been more recently identified as a secondary cause.(4) ARPKD is often diagnosed in utero due to oligohydramnios. NPHP is an autosomal recessive condition characterized by cystic kidney disease, inflammation, fibrosis, and progression to kidney failure. MCKD is an autosomal dominant condition characterized by cysts in the medullary region of the kidney, kidney tubule fibrosis, hyperuricemia, and slowly worsening kidney function. Genes included on this panel for MCKD include *HNF1B*, *UMOD*, and *SEC61A1* (note the *MUC1* gene is not included on this panel).

Phakomatoses, also known as neurocutaneous syndromes, are a broad group of hereditary disorders characterized by involvement of structures that arise from the embryonic ectoderm (central nervous system, skin, and eyes). Cystic renal lesions are common in these disorders. Phakomatoses genes included on this panel are the *TSC1* and *TSC2* tumor suppressor genes associated with tuberous sclerosis complex (TSC) and the *VHL* gene associated with von Hippel-Lindau syndrome.

Reference Values

An interpretive report will be provided.

Interpretation

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(5) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

Clinical Correlations:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of

at-risk individuals.

To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact Mayo Clinic Laboratories genetic counselors at 800-533-1710.

Technical Limitations:
Next-generation sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions; assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

Deletion/Duplication Analysis:
This analysis targets single and multi-exon deletions/duplications; however, in some instances single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

Deletion/duplication events that extend past the genes included on the panel may occur. In these instances, genes included in the ordered test are provided on the report and interpreted, and genomic breakpoints are reported if they are confirmed. However, copy number variants for genes not listed in the Method Description are typically not reported or interpreted for haploinsufficiency/triplosensitivity. CMACB / Chromosomal Microarray, Congenital, Blood; WESPR / Panel to Whole Exome Sequencing Reflex Test, Varies; or WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies is recommended for a full interpretation of deletions/duplications predicted to extend past the genes included on the panel.

This test is not designed to detect low levels of mosaicism or to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Genes may be added or removed based on updated clinical relevance. Refer to the Targeted Genes and Methodology Details for Cystic Kidney Disease Gene Panel for the most up to date list of genes included in this test. r detailed information regarding gene specific performance and technical limitations, see Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants:
At this time, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare providers to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time.

Variant Evaluation:
Evaluation and categorization of variants are performed using published American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations as a guideline.(5) Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgement.

Rarely, incidental findings or secondary findings may implicate another predisposition or presence of active disease. Incidental findings may include but are not limited to, results related to the sex chromosomes. These findings will be carefully reviewed to determine whether they will be reported.

Clinical Reference

1. Dillman JR, Trout AT, Smith EA, Towbin AJ: Hereditary renal cystic disorders: Imaging of the kidneys and beyond. Radiographics. May-Jun 2017;37(3):924-946. doi: 10.1148/rg.2017160148
2. Porath B, Gainullin VG, Cornec-Le Gall E, et al: Mutations in GANAB, encoding the glucosidase IIa subunit, cause autosomal-dominant polycystic kidney and liver disease. Am J Hum Genet. 2016 Jun 2;98(6):1193-1207. doi: 10.1016/j.ajhg.2016.05.004
3. Cornec-Le Gall E, Olson RJ, Besse W, et al: Monoallelic mutations to DNAJB11 cause atypical autosomal-dominant polycystic kidney disease. Am J Hum Genet. 2018 May 3;102(5):832-844. doi: 10.1016/j.ajhg.2018.03.013
4. Lu H, Rondon Galeano M, Ott E, et al: Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. Nat Genet. 2017 Jul;49(7):1025-1034. doi: 10.1038/ng.3871
5. Richards S, Aziz N, Bale S, et al: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med 2015 May;17(5):405-424. doi: 10.1038/gim.2015.30

Performance

Method Description

Capture-based and amplicon-based next-generation sequencing (NGS is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known disease-causing variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide

variants, above 94% for deletions-insertions (delins) less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. NGS and/or a polymerase chain reaction based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. See Targeted Genes and Methodology Details for Cystic Kidney Disease Gene Panel for details regarding the targeted genes analyzed for each test and specific gene regions not routinely covered.(Unpublished Mayo method)

Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genes analyzed: *ALG8, ALG9, ANKS6, BICC1, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TRAF3IP1, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, WDR35, XPNPEP3*

PDF Report
No

Day(s) Performed
Varies

Report Available
28 to 42 days

Specimen Retention Time
Whole blood: 2 weeks (if available); Extracted DNA: 3 months; Cultured amniocytes, cultured chorionic villi: 1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81404
81405
81406 x 6
81407 x 4
81408 x 3
81479
81265-Maternal cell contamination (if appropriate)
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
88235-Amniotic Fluid culture (if appropriate)
81479 (if appropriate for government payers)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CKDGP	Cystic Kidney Disease Gene Panel	51966-0

Result ID	Test Result Name	Result LOINC® Value
618073	Test Description	62364-5
618074	Specimen	31208-2
618075	Source	31208-2
618076	Result Summary	50397-9
618077	Result	82939-0
618078	Interpretation	69047-9
618079	Additional Results	82939-0
618080	Resources	99622-3
618081	Additional Information	48767-8
618082	Method	85069-3
618083	Genes Analyzed	48018-6
618084	Disclaimer	62364-5
618085	Released By	18771-6