THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Custom mutation analysis

What is custom mutation analysis?

Custom mutation analysis refers to testing of any gene for families with previously identified mutations associated with common or rare genetic conditions. We are CLIA certified and CAP accredited, thereby ensuring compliance with quality assurance and quality control guidelines that are not required of research laboratories.

Who can benefit from custom mutation analysis?

- Patients that have had a mutation identified in a research laboratory and would like clinical confirmation for management or diagnostic purposes
- Families in which a mutation was identified previously in an affected family member in a research laboratory, and the family is seeking prenatal or carrier testing
- Families with an identified mutation, who are interested in prenatal or carrier testing which is not currently available in a clinical laboratory

Note: Research findings must be confirmed prior to testing of additional family members.

Steps

1. Contact Laboratory Genetic Counselor (773-834-0555; ucgslabs@genetics.uchicago.edu) to discuss the individual case.
2. Provide written documentation of the identified mutation. This can be either a formal report from a clinical or research laboratory or just an email containing the gene of interest, mutation, PCR primer sequence and PCR amplification conditions used to identify the mutation (not absolutely necessary). We can also contact the research or clinical laboratory to obtain this information.
3. If little is known regarding the mutation or gene of interest, we may request evidence from the researcher to suggest that the change is causative of the condition in question.
4. Upon reviewing the above information, Eden will contact you to notify you that we are ready to receive a sample for mutation confirmation.
5. Send us a new blood sample (or a sample stored in a clinical laboratory) from an affected individual or obligate carrier for clinical confirmation of the mutation. Mutation analysis is performed by automated DNA sequencing and gene deletion analysis is performed by real-time quantitative PCR (RT-QPCR).

Clinical confirmation of a known mutation:

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Turn-around time:	4 - 6 weeks

DNA sequencing

Cost:	$540
CPT codes:	81403

Deletion/duplication analysis by RT-QPCR

Cost:	$1000
CPT codes:	81402

6. We will issue a clinical report upon confirmation.

7. Prenatal, carrier or diagnostic testing can then be offered to any other individuals in the family.

Testing of known mutation in additional family members:

Sample specifications: 3 to10 cc of blood in a purple top (EDTA) tube
Turn-around time: 3 - 4 weeks

DNA sequencing
Cost: $500
CPT codes: 81403

Deletion/duplication analysis by RT-QPCR
Cost: $1000
CPT codes: 81402

Prenatal testing of known mutation in additional family members:

Sample specifications: Amniotic fluid or chorionic villi cultures: 2 confluent T-25 flasks
(Direct) amniotic fluid: 10-15 cc
Turn-around time: 1 - 2 weeks

DNA sequencing
Cost: $1800
CPT codes: 81403

Deletion/duplication analysis by RT-QPCR
Cost: $1800
CPT codes: 81402

Results:

Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal and prenatal results will be reported by telephone.

For more information about our testing options, please visit our website at dnatesting.uchicago.edu or contact us at 773-834-0555.

Committed to CUSTOMIZED DIAGNOSTICS, TRANSLATIONAL RESEARCH & YOUR PATIENTS' NEEDS