THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Custom RNA Splicing Analysis

What is custom RNA splicing analysis?

Our custom RNA splicing analysis is designed to test the effect of a previously identified DNA sequence variant on RNA splicing. 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 RNA splicing analysis?

Our custom RNA splicing analysis assay can be beneficial for patients who have previously had a variant of unknown clinical significance identified in a clinical or research laboratory. In some of these cases, RNA splicing analysis help clarify the pathogenicity of the variant by clinically evaluating the effect of the variant on native RNA splicing. Typically variants that fit the following criteria are good candidates for custom RNA splicing analysis:

- *In-silico* predictions on the variant suggest a deleterious effect on splicing;
- The variant is in a gene associated with a disorder that fits the patient's phenotype;
- The mRNA of tested gene/transcript is expressed at a detectable level in blood and/or skin

What are the requirements for this test?

- Prior to sending a sample to our laboratory, please contact one of our laboratory genetic counselors (773-834-0555; ucgslabs@genetics.uchicago.edu) to discuss the individual case.
- If the variant you wish to perform RNA splicing analysis on was not originally identified in our laboratory, you will need to provide written documentation of the identified variant (such as DNA-level and protein-level HGVS nomenclature) and the transcript utilized (e.g. RefSeq). The formal report from a clinical or research laboratory is also accepted. If little is known regarding the variant or gene of interest, we may request evidence from the laboratory that identified the variant to suggest that the variant is causative of the condition in question.
- We will review the available evidence for your variant, and if the case is a good candidate for custom RNA splicing analysis we will arrange to ship a PreAnalytix PAXgene Blood RNA tube to you for blood collection from the affected individual. Further instructions on how to collect the blood will be sent out together with the PreAnalytix PAXgene Blood RNA tubes. Extracted DNA or blood in a lavender top (EDTA) tube cannot be used for custom RNA splicing analysis. If a patient with hematological malignancies is tested we will require 2 T-25 flasks of cultured skin fibroblasts from the affected individual.

How is custom RNA splicing analysis performed?

Splicing analysis is performed by OneStep-RT-PCR and subsequent Sanger sequencing, using customized primers designed to detect defective splicing related to the variant of interest. Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested.

Custom RNA splicing analysis:

Sample specifications:	2.5 cc of blood in a PreAnalytix PAXgene Blood RNA tubes
Turn-around time:	12-16 weeks
Cost:	$1000*
CPT codes:	81479

**If testing fails due to failed amplification of RNA or other technical issues, a processing fee will still be charged. Please contact us for more details.*

For more information about all 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