myVision inSite™

Myriad's *inSite*™ RNA lab: a specialty tool which has supported our variant classification program for unique cases since 2015

Most mutations do not require RNA studies!

Myriad follows an IRB approved research protocol:

Germline testing performed with initial review

Variant of interest identified using factors such as predicted splicing impact

GC contacts ordering healthcare provider and patient to obtain verbal consent to RNA study

Blood collection kit sent to acquire RNA

RNA data analyzed and reviewed by reclassification committee

Amended report issued if variant is reclassified

From December 2015 to September 2019, Myriad sent out over 69,000 amended reports to patients using tools such as RNA as a part of our robust variant classification program

RNA analysis is one of many tools used by Myriad to analyze and classify variants

By identifying variants beyond -/+5 in the intron, Myriad has identified pathogenic variants affecting more than 2900 patients. Here are some examples:

Case Study 1

RNA analysis yields a definitive answer

BRCA1 (c.135-8A>G)

Variant identified

Meets criteria for RNA analysis

Patient consent obtained and sample collected

RNA analysis is performed

RNA results reviewed by reclassification committee

Result:

Sufficient data to confidently reclassify as suspected deleterious based on RNA analysis in 2019

Case Study 2

Other tools yield an answer for a splicing variant

MSH2 (c.2459-12A>G)

Variant identified

Several classification tools applied to variant to gather additional evidence for classification

Pheno® flags variant as meeting threshold for reclassification, prompting review by reclassification committee

Result:

Sufficient data to confidently reclassify as suspected deleterious based on Pheno in 2015 without needing RNA lab