Figure S5

RNA Alignment with Tophat

RDD Calling with rddChecker

Removal of SNPs, indels, CNVs based on Individual’s Genome Sequence

Filter Mismappings with BLAT & Blastn

Identify RDDs which can be explained as an uncalled SNP

Validate Candidate RDDs

**Mismapping Example**

Gene X Genomic DNA

ACTGAA CAGTCAGTTCAGTA...

RNA Alignments

ACTGAA CAGTCAGTTCAGTA...

GAA TAGTCAATTCAGTA...

...CTGAA TAGTCAATTCAGTA...

...GCC TAGGCATTATCCAT...

...GCC AAGGCATTATCC....

...GCC AAGGCATTATCC....

...GCC TAGGCATTATCCATAC.

...GCC AAGGCATTATCC....

...GCC AAGGCATTATCC....

...GCC AAGGCATTATCC....

**Uncalled SNP Example**

Reference DNA

GCC 

AGCC 

AGCC 

AGCC 

AGCC 

AGCC

DNA Alignments

TAGCATATCCAT...

...AGCC ...AGCCATTATCC...

...AGCC ...AGCCATTATCCATAC.

...AGCC ...AGCCATTATCCAT...

...AGCC ...AGCCATTATCC...

RNA Alignments

...GAA 

...GAA 

...GAA 

...GAA 

...GAA

Called RDD

T 

T 

T 

T 

T

The putative RDD is a SNP

8,252 SNPs 646 Low Genome Coverage 115 Substitutions 5 Deletions 57 CNVs

548 Mismappings Identified

8,200 0 200 400 600 800 8,252

SNPs Low Genome Coverage Substitutions Deletions CNVs

8,000 Count

548 Mismappings Identified

Deletions Substitutions