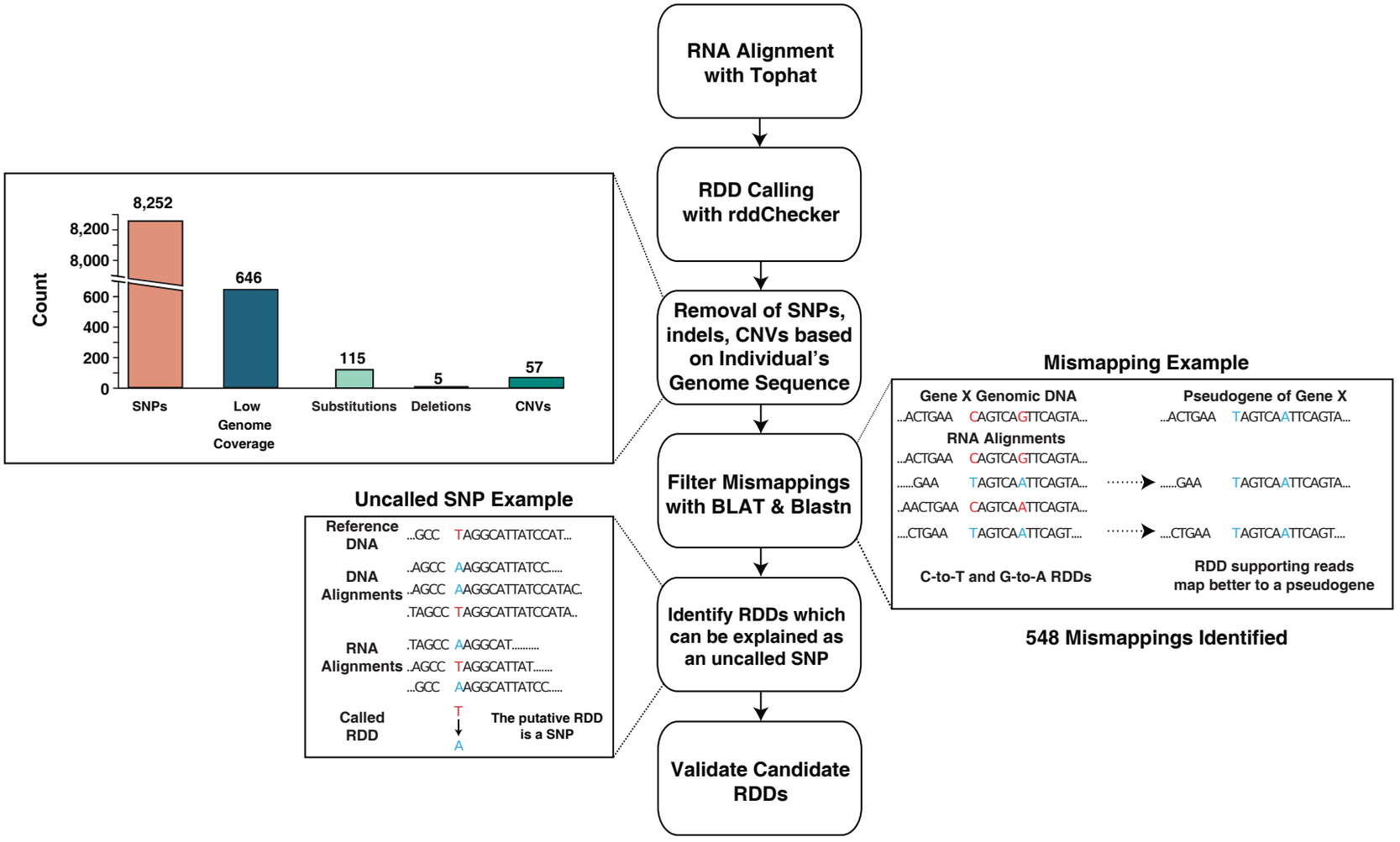
Figure S5


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
Count
8,252
8,200
8,000
600
400
200
0
646
115
5
57
SNPs
Low Genome Coverage
Substitutions
Deletions
CNVs
Uncalled SNP Example
Reference DNA
...GCC TAGGCATTATCCAT...
DNA Alignments
..AGCC AAGGCATTATCC.....
..AGCC AAGGCATTATCCATAC.
.TAGCC TAGGCATTATCCATA..
RNA Alignments
.TAGCC AAGGCAT.....
..AGCC TAGGCATTAT.....
...GCC AAGGCATTATCC.....
Called RDD
T
A
The putative RDD is a SNP
Mismapping Example
Gene X Genomic DNA
...ACTGAA CAGTCAGTTCAGTA...
Pseudogene of Gene X
...ACTGAA TAGTCAATTCAGTA...
RNA Alignments
...ACTGAA CAGTCAGTTCAGTA...
.....GAA TAGTCAATTCAGTA...
.....GAA TAGTCAATTCAGTA...
..AACTGAA CAGTCAATTCAGTA...
....CTGAA TAGTCAATTCAGT....
....CTGAA TAGTCAATTCAGT....
C-to-T and G-to-A RDDs
RDD supporting reads map better to a pseudogene
548 Mismappings Identified