

A Cross-Sectional Study for Variant Interpretation and Reporting of NGS Data Adopting Tertiary Analysis Software: Navify® Mutation Profiler

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Navify mutation Profiler successfully interpretes

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Allow correct RNA analysis.

In order to generate a report on NMP software by using “OncoPrint_Fusions_only” assay, input VCF files was modified following listed workflow parameters:

1. IonReporterSoftwareVersion=5.14.0.0
2. IonReporterWorkflowVersion=ir514
3. OncoPrintVariantAnnotationToolVersion=3.0.8

Briefly, these modifications allow VCF files analysis of RNA-derived samples on NMP system. It has been suggested to assess these technical specifications opening the VCF file as “text editor”. After technical modification, file can be saved and loaded on NMP.

```
Not Modified - Blocco note di Windows
File Modifica Formato Visualizza ?
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##fileDate=2022/10/21
##reference=hg19
##IonReporterAnalysisName=RNA_LC_X4_LibPrep201
##annotationSources=[dra_20170914, drugbank_20200619, g
##sampleDiseaseType=Non-Small Cell Lung Cancer
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##IonReporterSoftwareVersion=5.16.0.0
##FusionSampleOverallCall=POSITIVE,[DriverGene=ROS1,Evg
##sampleGender=Unknown
##FusionSampleQC=PASS,[AllRNAProcessControlsDetected>6]
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##IonReporterWorkflowVersion=ir516
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##ALT=<ID=CNV,Description="Copy number variable region"
##ALT=<ID=DEL,Description="Deletion"
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##ALT=<ID=DUP,Description="Duplication"
##ALT=<ID=DUP:TANDEM,Description="Tandem Duplication"
##ALT=<ID=ExprControl,Description="Expression of the Co
##ALT=<ID=Fusion,Description="Fusion between two differ
##ALT=<ID=GeneExpression,Description="Expression of the
##ALT=<ID=INS,Description="Insertion of novel sequence"
##ALT=<ID=INS:ME:ALU,Description="Insertion of ALU eleme
##ALT=<ID=INS:ME:L1,Description="Insertion of L1 eleme
##ALT=<ID=INV,Description="Inversion"
##ALT=<ID=ProcControl,Description="Expression Control g
##ALT=<ID=RNAExonTiles,Description="Gene Level result s
##ALT=<ID=RNAExonVariant,Description="Expression of the
##ALT=<ID=RNA_Hotspot,Description="Expression of a Targ
##MeanReadLength=101
##TotalMappedFusionPanelReads=80467
##TotalUnmappedReads=1305291
##OncoPrintVariantAnnotationToolVersion=3.1.8
##OncoPrintAnnotationKeys=oncoPrintGeneClass,oncoPrintVar

Modified - Blocco note di Windows
File Modifica Formato Visualizza ?
##fileformat=VCFv4.1
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##reference=hg19
##IonReporterAnalysisName=RNA_LC_X4_LibPrep201
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##sampleDiseaseType=Non-Small Cell Lung Cancer
##IonReporterExportVersion=0.1.5
##IonReporterSoftwareVersion=5.14.0.0
##FusionSampleOverallCall=POSITIVE,[DriverGene=ROS1,EvidenceLevel=Targete
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##FusionSampleQC=PASS,[AllRNAProcessControlsDetected>6]
##IonReporterWorkflowName=RDx Ampliseq Fusions Template
##IonReporterWorkflowVersion=ir514
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##ALT=<ID=CNV,Description="Copy number variable region"
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##ALT=<ID=DUP,Description="Duplication"
##ALT=<ID=DUP:TANDEM,Description="Tandem Duplication"
##ALT=<ID=ExprControl,Description="Expression of the Control House keepin
##ALT=<ID=Fusion,Description="Fusion between two different Gene Transcrip
##ALT=<ID=GeneExpression,Description="Expression of the Target Gene Trans
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##ALT=<ID=RNAExonTiles,Description="Gene Level result summary from the RN
##ALT=<ID=RNAExonVariant,Description="Expression of the Target Gene Trans
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##MeanReadLength=101
##TotalMappedFusionPanelReads=80467
##TotalUnmappedReads=1305291
##OncoPrintVariantAnnotationToolVersion=3.0.8
##OncoPrintAnnotationKeys=oncoPrintGeneClass,oncoPrintVariantClass
```

Figure: Screenshots of RNA VCF files before (left) and after (right) required modifications by using Windows “text editor”.