Updating MiSeq Reporter with dbSNP version 137

This document provides instructions for updating the dbSNP information used by the MiSeq Reporter software. Use the file provided to update MiSeq Reporter with dbSNP version 137.

MiSeq Reporter uses dbSNP in the variant calling step of the Resequencing, PCR Amplicon, and TruSeq Amplicon workflows. SNPs that are identified by MiSeq Reporter variant calling are cross-referenced against the dbSNP information. If a match is found in dbSNP, the dbSNP identifier (rs number) is written to the VCF file. For more information, see the associated workflow guide available MiSeq Report support page on the Illumina website.

dbSNP is the NCBI database used to annotate variants found during analysis by MiSeq Reporter. It is downloaded from UCSC using the link below. The file is not edited or altered by Illumina.

http://hgdownload.cse.ucsc.edu/goldenPath/hg19/database/snp137.txt.gz

Downloading the Update

1   From the Accellion site, click the checkbox next to the file name **snp137.zip**.
2   Click **Download via Applet**.
    Note that downloading the file can take up to several hours due to the file size.
3   Extract the zipped file to obtain the file **snp137.txt**.

Installing the Update

Important: Make sure that the MiSeq Reporter service is idle before beginning the update. If any analyses are in progress, wait until MiSeq Reporter is finished.

1   Place the MiSeq in Windows mode. A mouse and keyboard are recommended.
2   Navigate to the Annotation folder on the MiSeq instrument computer:
    C:\Illumina\MiSeq Reporter\
    Genomes\Homo_sapiens\UCSC\hg19\Sequence\WholeGenomeFASTA\Annotation
3   In the Annotation folder, rename or delete the file dbsnp.txt.
4   Save the file **snp137.txt** to the Annotation folder.
5   Rename snp137.txt to **dbsnp.txt**.
6   In the Annotation folder, rename or delete the file dbsnp.txt.idx, if present.
    The dbsnp.txt.idx file is an index file that MiSeq Reporter builds using the dbSNP.txt file.
    Deleting the old file triggers MiSeq Reporter to automatically recreate dbsnp.txt.idx.

You do not need to restart the MiSeq Reporter Service.