NGS Bioinformatics

Module topic: Pathogen variant calling
Contact session title: Inferring genetic relatedness
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Participant: <*write your name here*>
Date: <*write today's date here*>

Genetic relatedness

Introduction

In the assignment, you will use the variants identified in the previous session (variant calling) to generate a pseudogenome, this is done by replacing the bases in the reference genome for which we identified variants from mapping. This will create a modified reference genome with all the variants inserted in the genome referred here as pseudogenome. This will be used to infer genetic relatedness (pairwise SNP difference) and construct a phylogenetic tree. The tree created will then be visualized in figtree.

Tools used in this session

Samtools, snp-sites, snp-dists, iqtree, figtree

Please note

- **Hand-in information** please upload your completed assignment to the Vula 'Assignments' tab. Take note of the final hand-in date for each assignment, which will be indicated on Vula.

Session 3: Genetic relatedness

Q 3.1: What is the length of the alignment in "snpsitesOut.fa" file?

>1000 (it can vary from 1169 to 1800)

Q 3.2: What is the pairwise SNP difference between the following pairs: MD001-- repMD001 and NC_00962.3 – repMD001?

a) MD001—repMD001: 846 (can be variable)
b) NC_00962.3 – repMD001: 984

Q3.3: What is the pairwise SNP difference for following pairs:

a) MD001-MD003: *386*
b) MD012-MD024: *1890*
c) MD003-MD012: *217*
d) MD001-MD024: *1827*

Q3.4: Report the resistance conferring variant identified for the following isolate drug combination: (record the position/coordinate and the mutation identified in the cases where isolates are resistant)

Isolate	Drug	Gene	Position	Genotype (R/S)
MD003	Isoniazid	*katG*	*2155168*	*R (S315T)*
MD003	Streptomycin	*rpsL*	*781687*	*R (K43R)*
MD012	Rifampicin	*rpoB*	*761155*	*R (S450L)*
MD012	Fluoroquinolone	*gyrA*	*7582*	*R (D94G)*
MD024	Isoniazid	*katG*	-	S
MD024	Streptomycin	*rpsL*	-	S
MD024	Rifampicin	*rpoB*	-	S
MD024	Fluoroquinolone	*gyrA*	-	S