

UEG27 Research Project

Red face, flushing and ALDH2 mutation

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Introduction

A patient (female, 19-year-old) came to the ER with a very red face and complaining about an accelerated heart rate. You immediatly suspect a mutation in the ALDH2 gene that is very frequent in people from East Asia and that causes this type of reaction under particular conditions.

Data at your disposal

To investigate this question, you have access to a reference genome of the human ALDH2 mRNA sequence and the reads from the patient (that are already filtered to match the right region).

Files

The given files are the following:

- The reference sequence of the the ALDH2 mRNA:
 - File name `NM_000690.3.fa`
- The set of reads generated on an illumina sequencer:
 - File name `NM_000690.3.fasta.fq`