

SNP Web-portal

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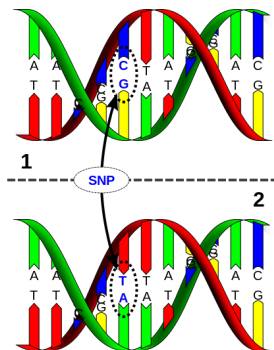
Section 1

Project description

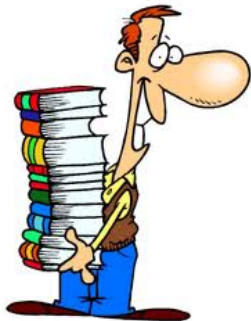
Medical background

Single-Nucleotide Polymorphism

DNA sequence variation occurring when a Single Nucleotide — A, T, C or G — in the genome (or other shared sequence) differs between members of a biological species or paired chromosomes.



Goal



Allow users to store and query Single Nucleotide Polymorphism genomics variants.

How to reach it?

Realization of a **Web Portal**.

Functionalities

Super User:

- create a “family” with one or more members
- authorize users
- modify inserted values

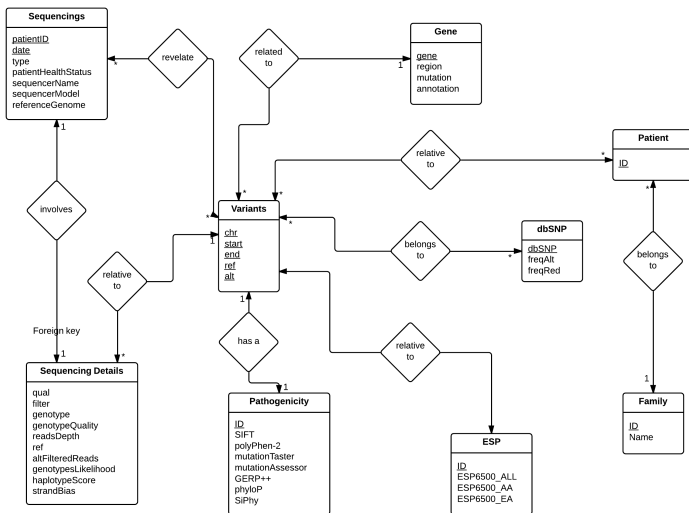
Authorized User:

- search for a patient, gene, mutation, ...

Section 2

Project Structure

Database:



Database - Website interaction:

Occurs through queries that allow searching for:

- patient's SNPs
- gene's SNPs
- region's SNPs
- all SNPs with certain Mutation, Genotype, Freq alt, ...
- patients with same SNP or Genotype
- SNPs within a genomic region
- specific SNP

Website (User Interface):

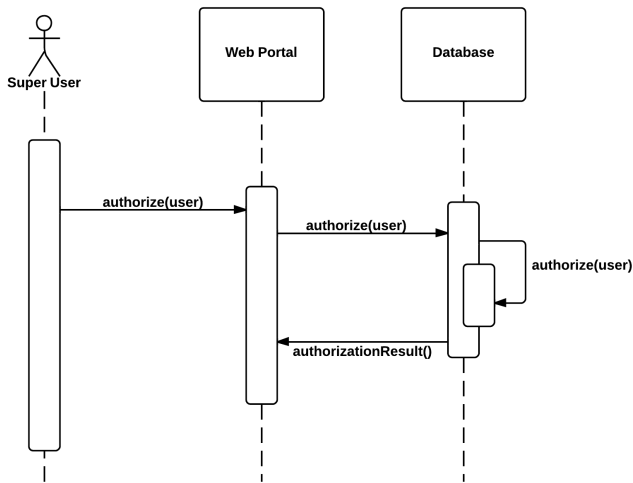
Enables to load and retrieve data via user-friendly UI



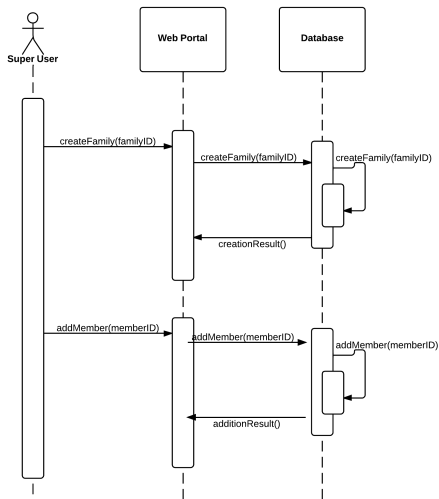
Section 3

Use cases

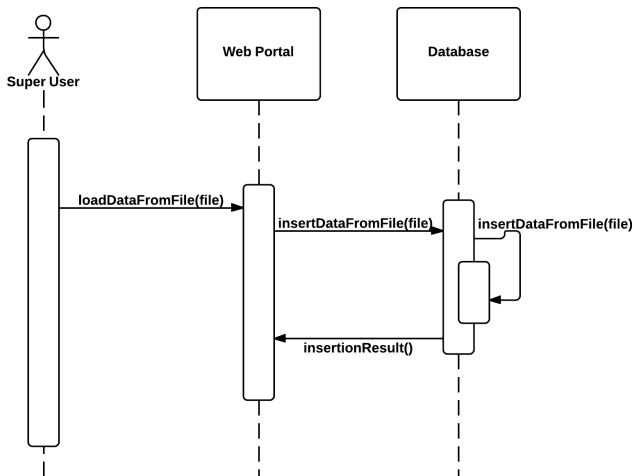
Use case diagram: Super User authorizes an User



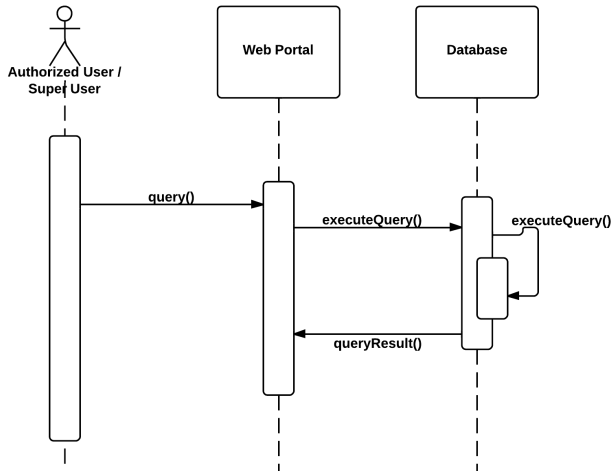
Use case diagram: Super User creates/populates a family



Use case diagram: Super User loads data



Use case diagram: Authorized User executes a query



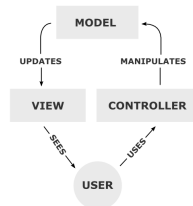
Section 4

Software architecture and technologies

To realize the project we will use:



Database: **MongoDB**



Model: **NodeJS**



Website: **AngularJS**



Framework: **ExpressJS**