

SNP Web-portal

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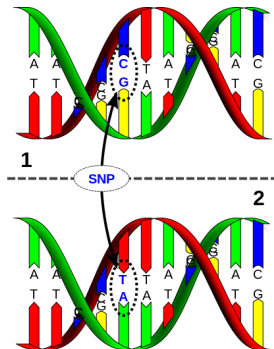
Biomedical Informatics, 2014

Project description

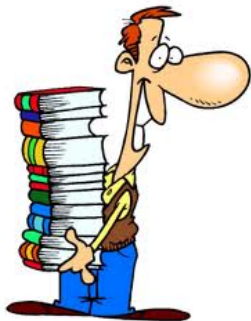
Medical background

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:

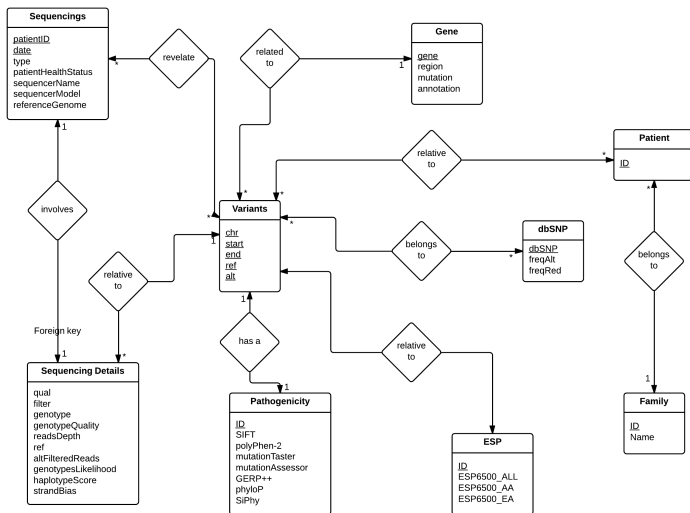
- create a “family” with one or more members
- authorize users
- modify inserted values

Authorized User:

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

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

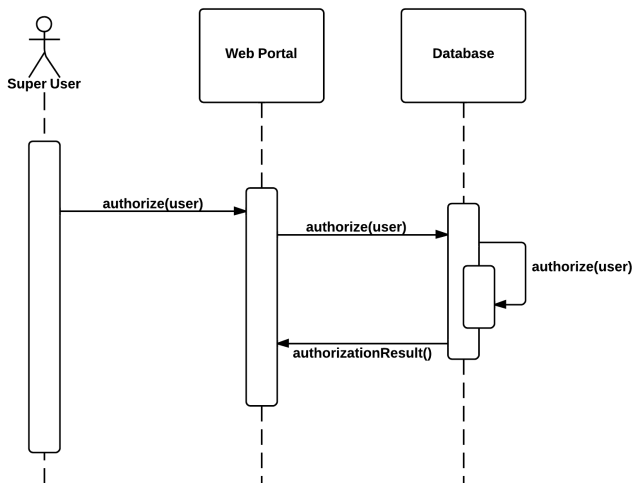
Website (User Interface):

Enables to load and retrieve data via user-friendly UI

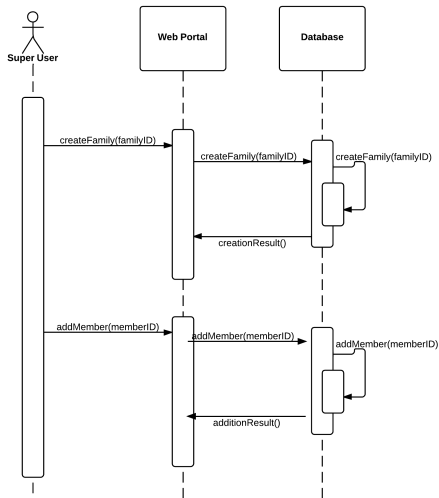


Use cases

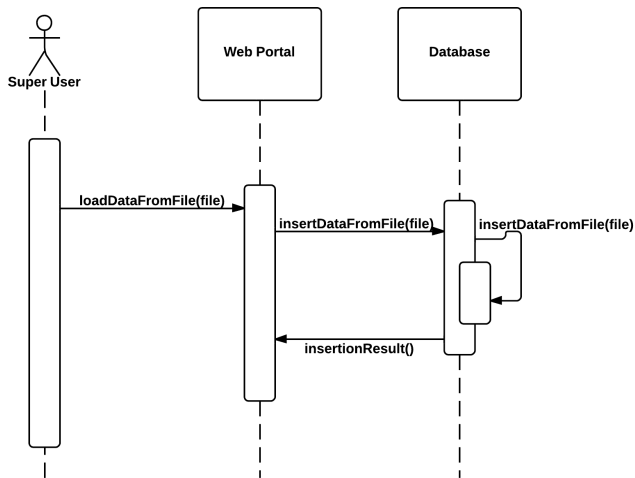
Use case diagram: Super User authorizes an User



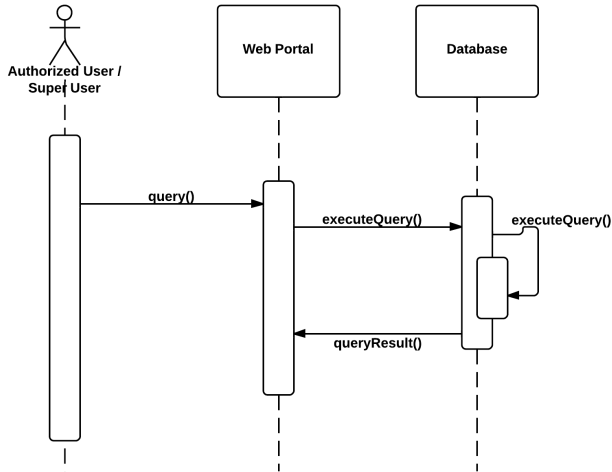
Use case diagram: Super User creates/populates a family



Use case diagram: Super User loads data

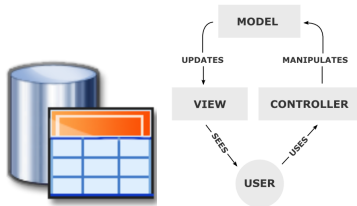


Use case diagram: Authorized User executes a query



Software architecture and technologies

To realize the project we will use:



Database: **MongoDB** Model: **NodeJS**



Framework: **ExpressJS**



Website: **AngularJS**

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Work breakdown

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- **Database:** Damian Tosoni, Ugo Buonadonna
- **Database - Website interaction:** Ugo Buonadonna, Davide Sicignani
- **Website:** Damian Tosoni, Davide Sicignani