SNP Web-portal

D. Tosoni, D. Sicignani, U. Buonadonna
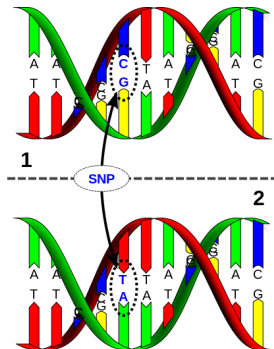
Biomedical Informatics, 2014

Project description
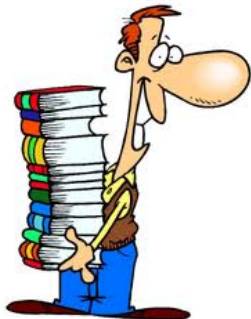
Medical background

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

Main target: **BIOLOGISTS**

Help them in their work of storing and retrieving Single Nucleotide Polymorphism genomics variants.

How to reach it?

Realization of a **Web Portal**.
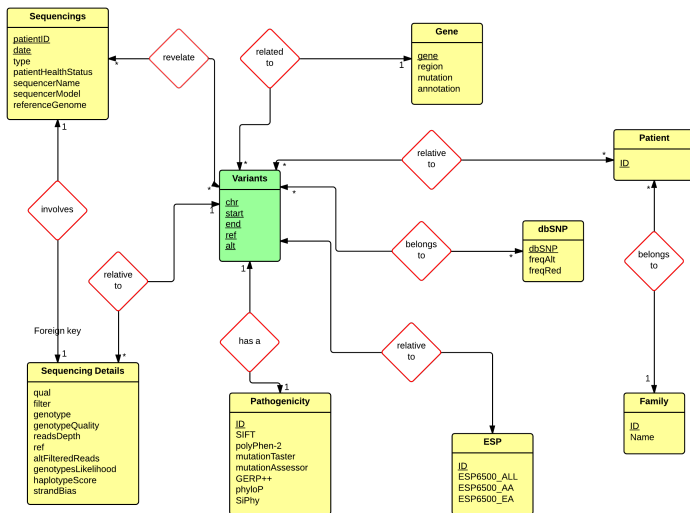
Functionalities

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
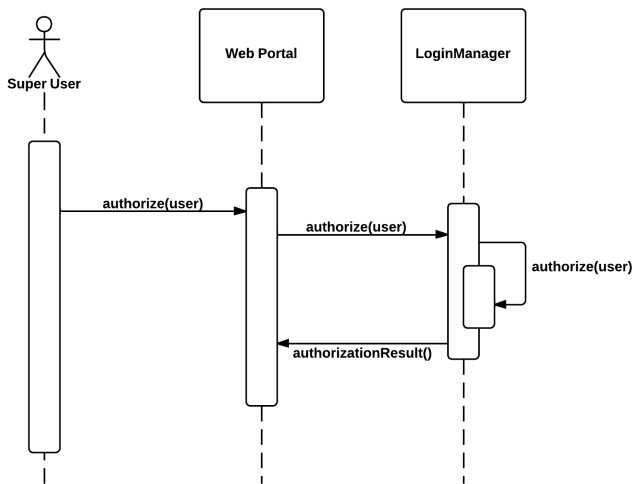- specific SNP

Website (User Interface):

Enables to load and retrieve data via a user interface that:

- allows biologists to use service as easily as possible, and. . .
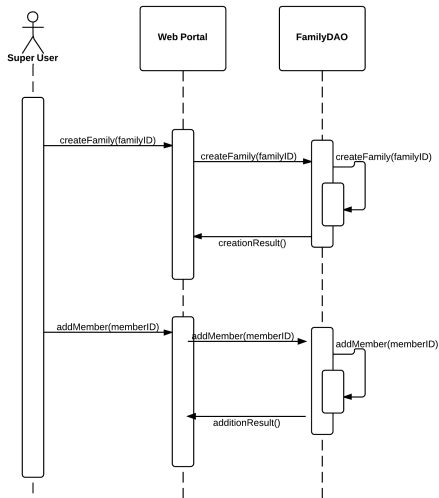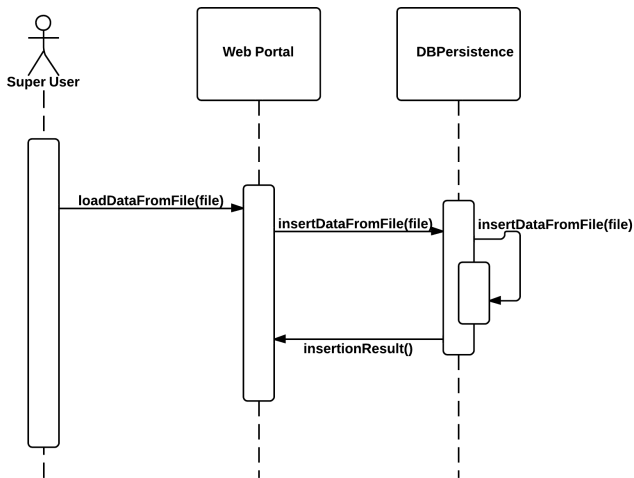- . . . speed up their work.

Use cases

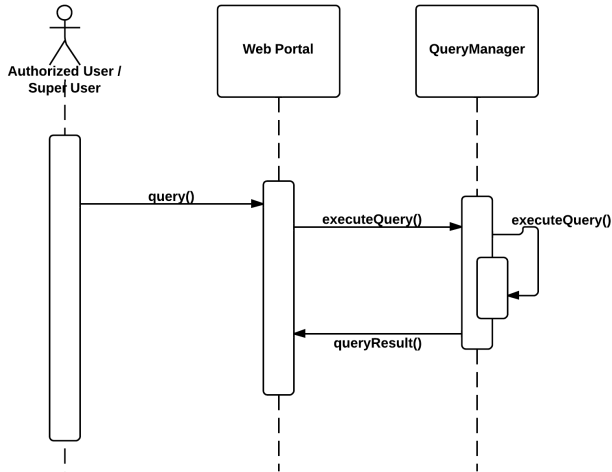# Use case diagram: Super User authorizes an User

Use case diagram: Super User creates/populates a family

Use case diagram: Super User loads data

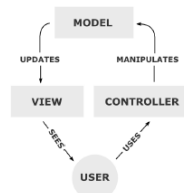# Use case diagram: Authorized User executes a query

Software architecture and tecnologies

To realize the project we will use:

Database: **MongoDB**

Model: **NodeJS**

Website: **AngularJS**

Framework: **ExpressJS**

To realize the project we will use:

Task breakdown

Task breakdown:

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