SNP Web-portal

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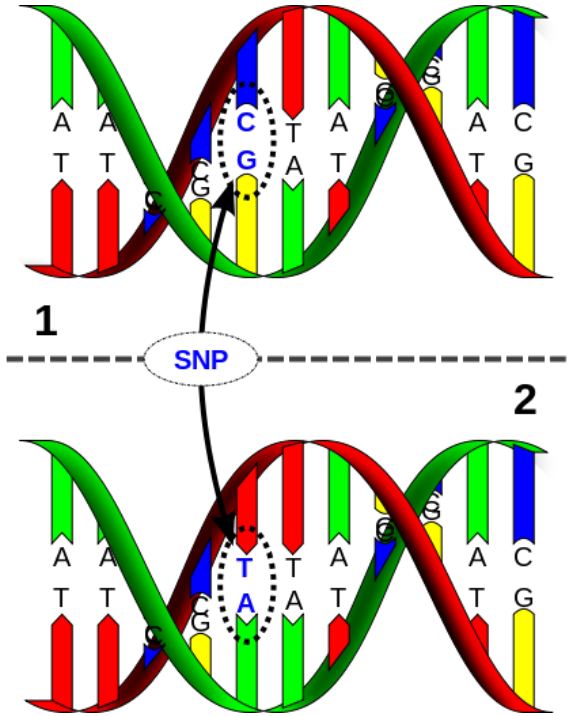
Project description

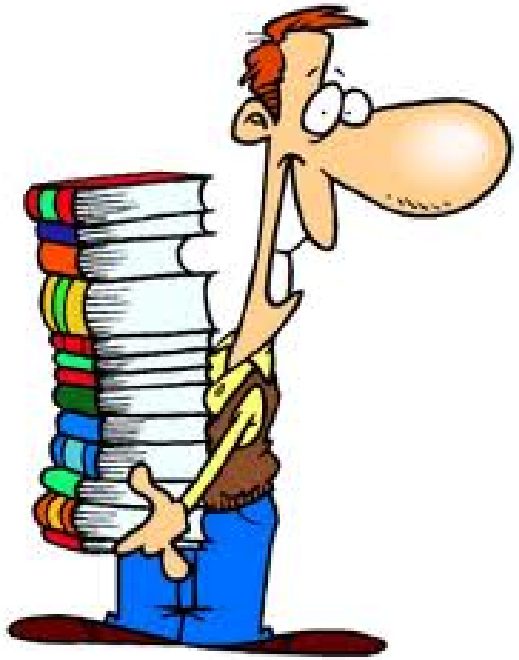
Medical background

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:

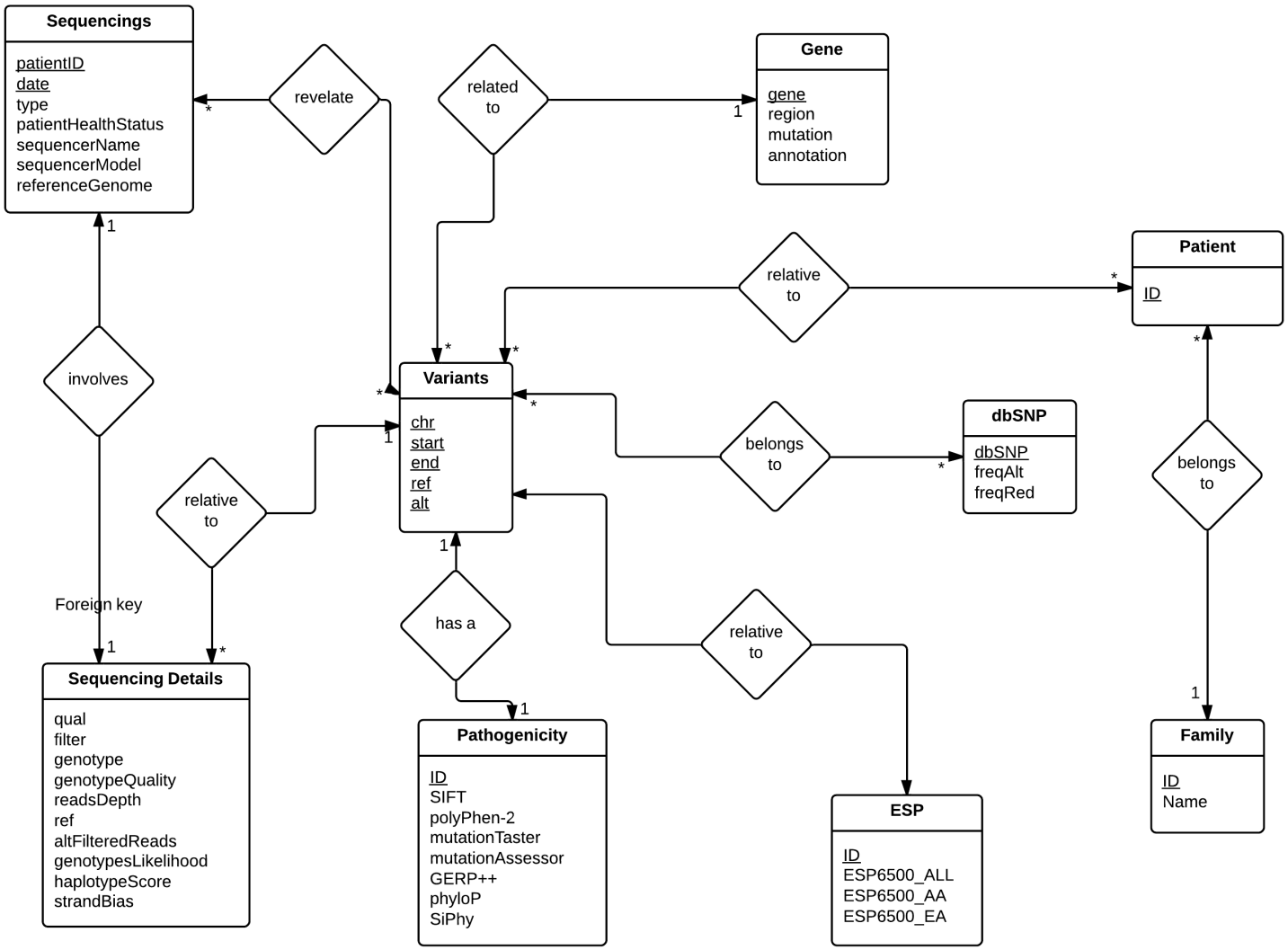
- create a "family" with one or more members
- authorize users
- modify inserted values

Authorized User:

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

Project Structure

Database:


Sequencings
patientID
date
type
patientHealthStatus
sequencerName
sequencerModel
referenceGenome
revelate
related to
Gene
gene
region
mutation
annotation
relative to
Patient
ID
involves
Variants
chr
start
end
ref
alt
belongs to
dbSNP
dbSNP
freqAlt
freqRed
relative to
belongs to
Foreign key
has a
relative to
Sequencing Details
qual
filter
genotype
genotypeQuality
readsDepth
ref
altFilteredReads
genotypesLikelihood
haplotypeScore
strandBias
Pathogenicity
ID
SIFT
polyPhen-2
mutationTaster
mutationAssessor
GERP++
phyloP
SiPhy
ESP
ID
ESP6500_ALL
ESP6500_AA
ESP6500_EA
Family
ID
Name

Database - Website interaction:

Occurs through queries that allow searching for:

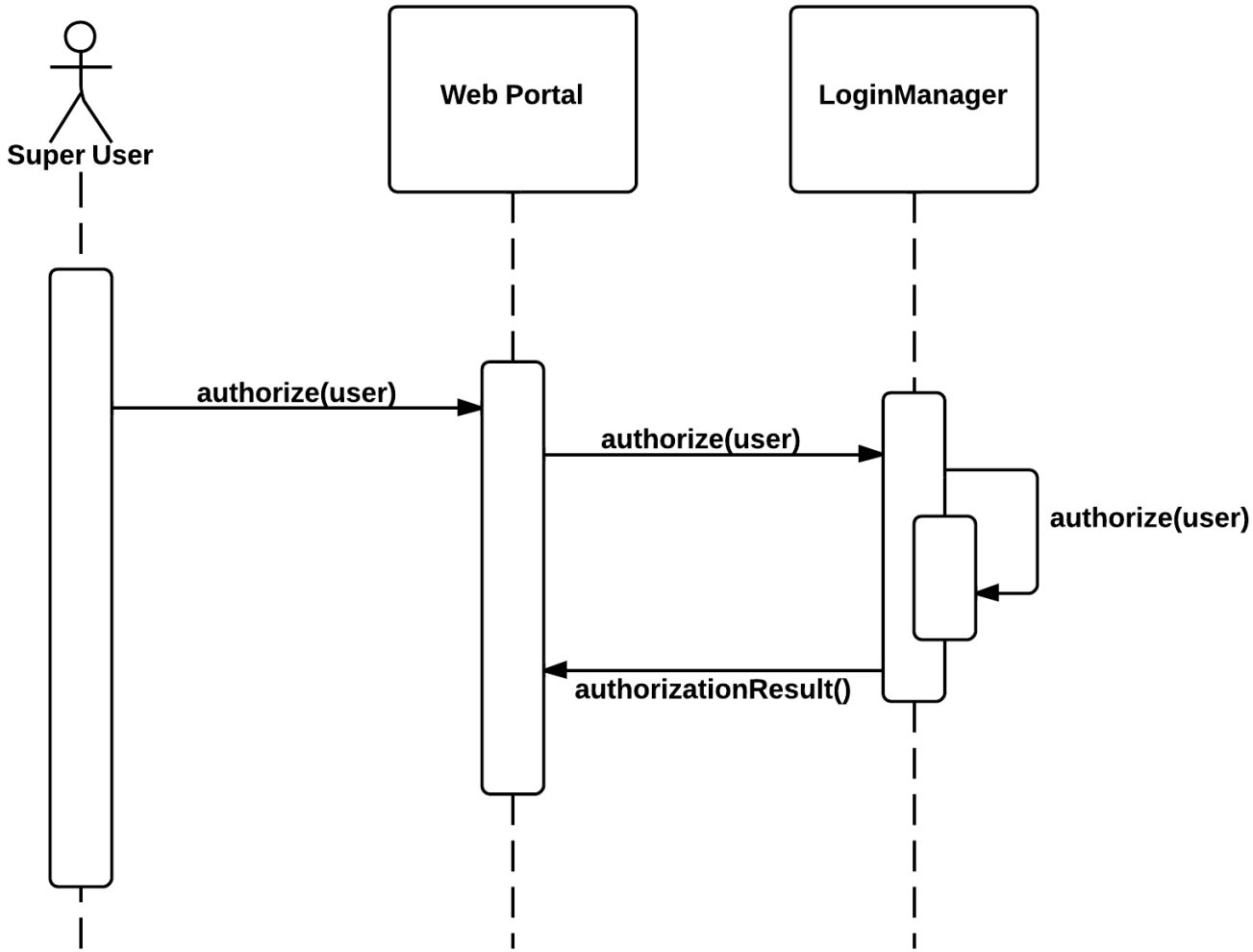
- patient's SNPs
- gene's SNPs
- region's SNPs
- all SNPs with certain Mutation, Genotype, Freq alt, ...
- patients with same SNP or Genotype
- SNPs within a genomic region
- specific SNP

Website (User Interface):

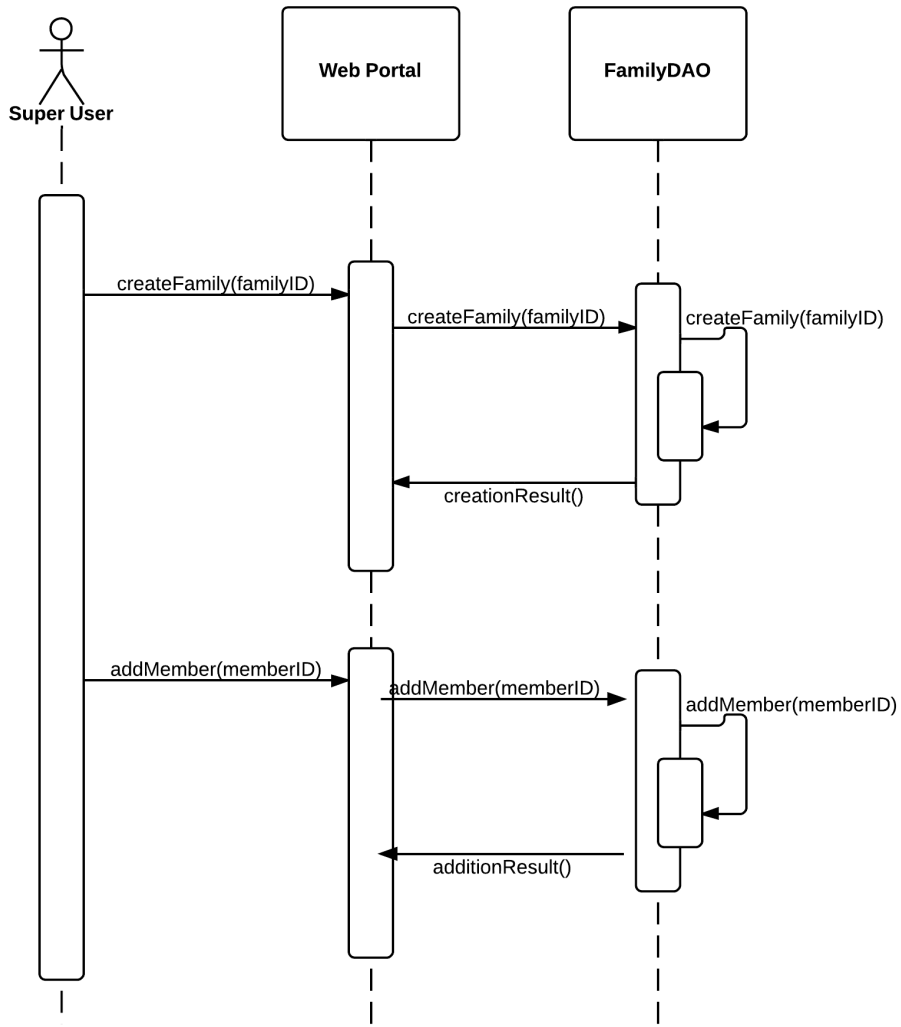
Enables to load and retrieve data via user-friendly UI

Use cases

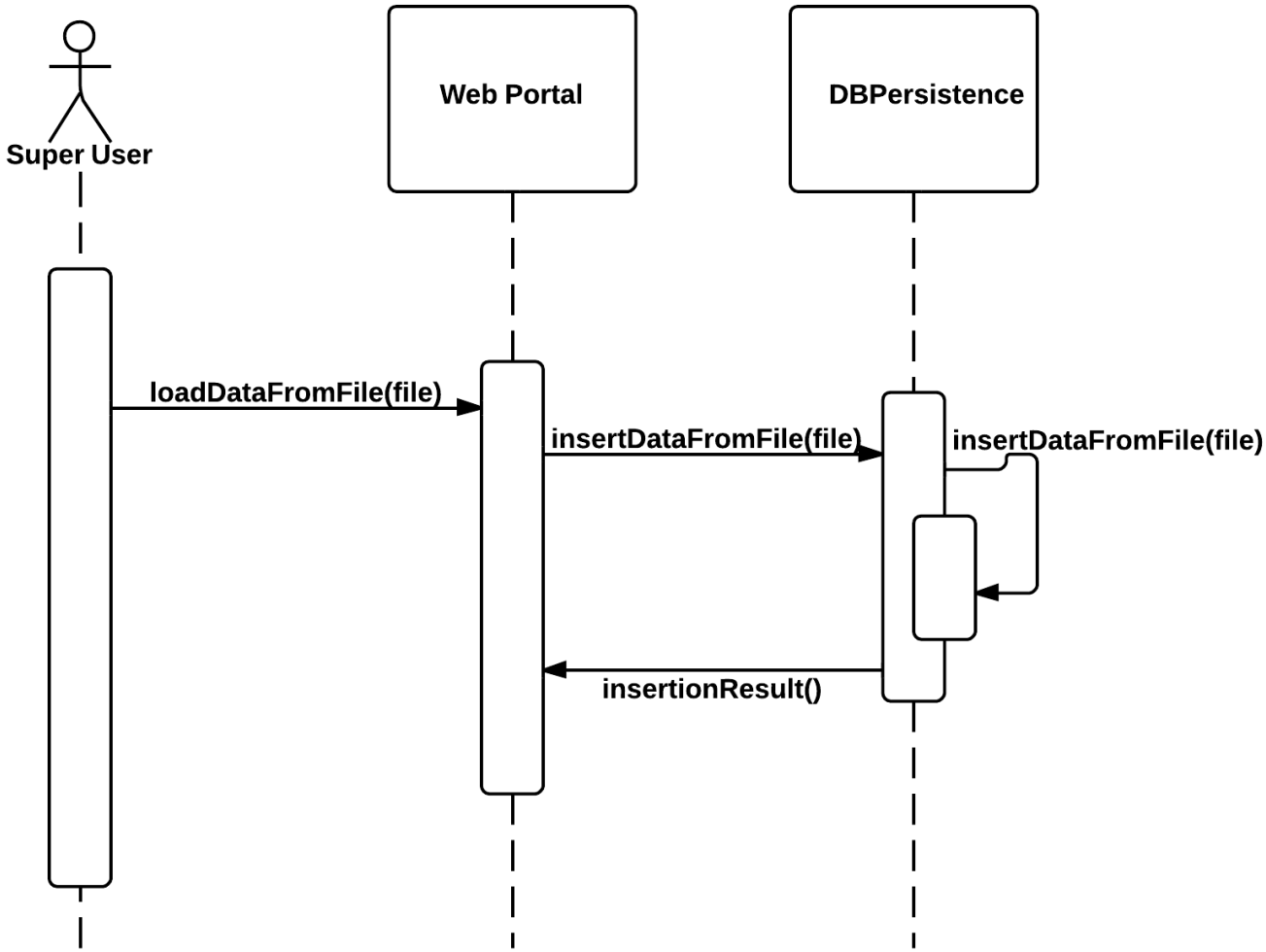
Use case diagram: Super User authorizes an User



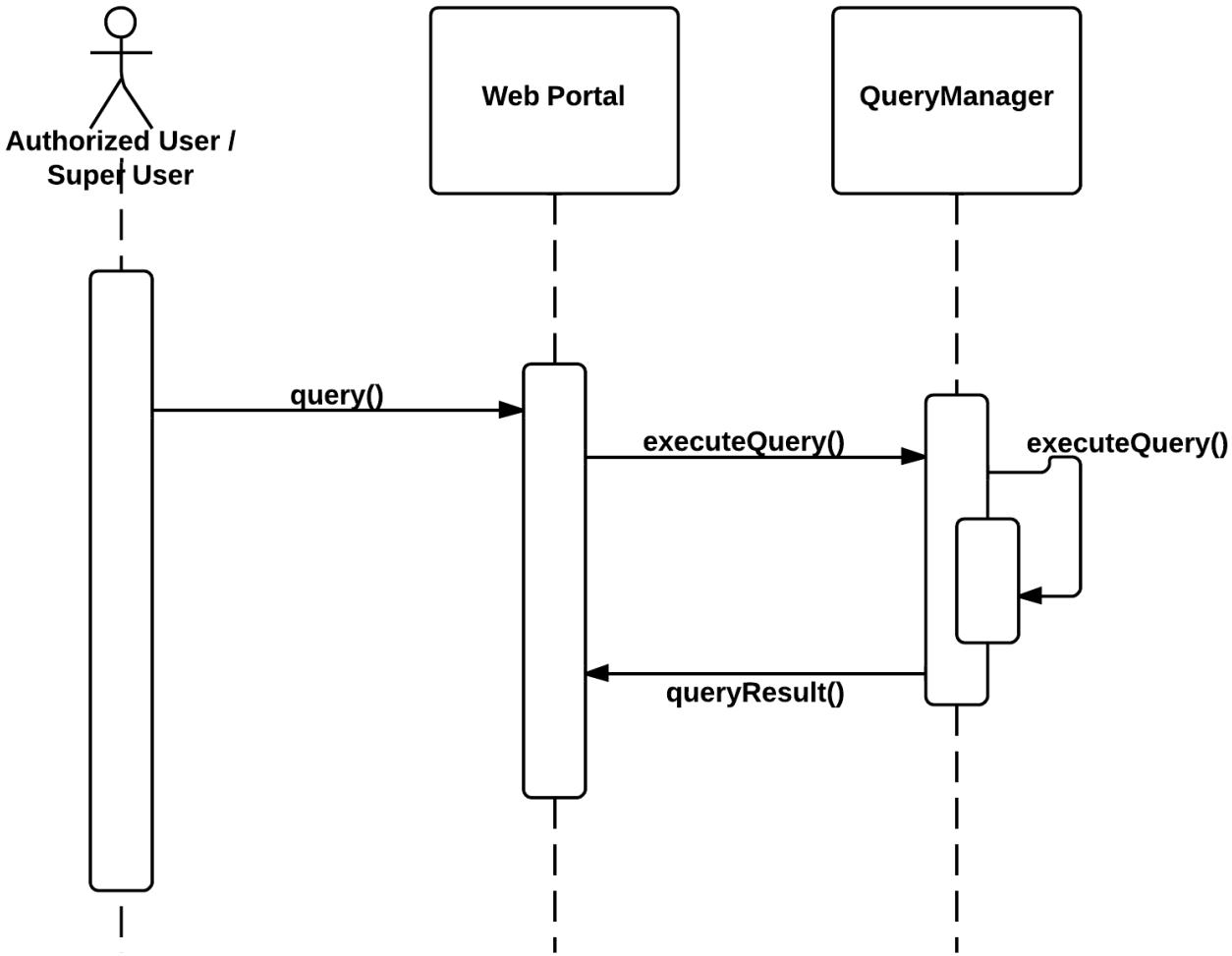
Use case diagram: Super User creates/populates a family



Use case diagram: Super User loads data



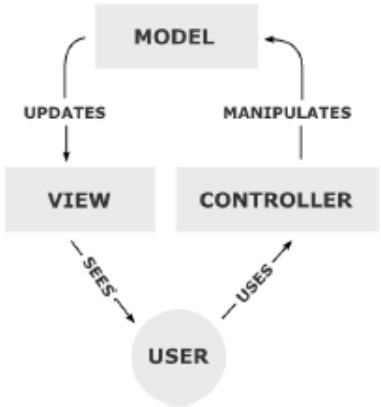
Use case diagram: Authorized User executes a query



Software architecture and tecnologies

To realize the project we will use:

Database: **MongoDB**



Model: **NodeJS**

Website: **AngularJS**

Framework: **ExpressJS**

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Work breakdown

Work breakdown:

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