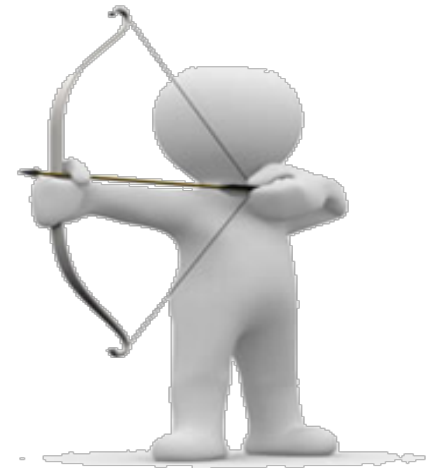


# Haplotypes and Statistical Models: Integrating to the Conceptual Schema of the Human Genome



**José Fabián Reyes Román**  
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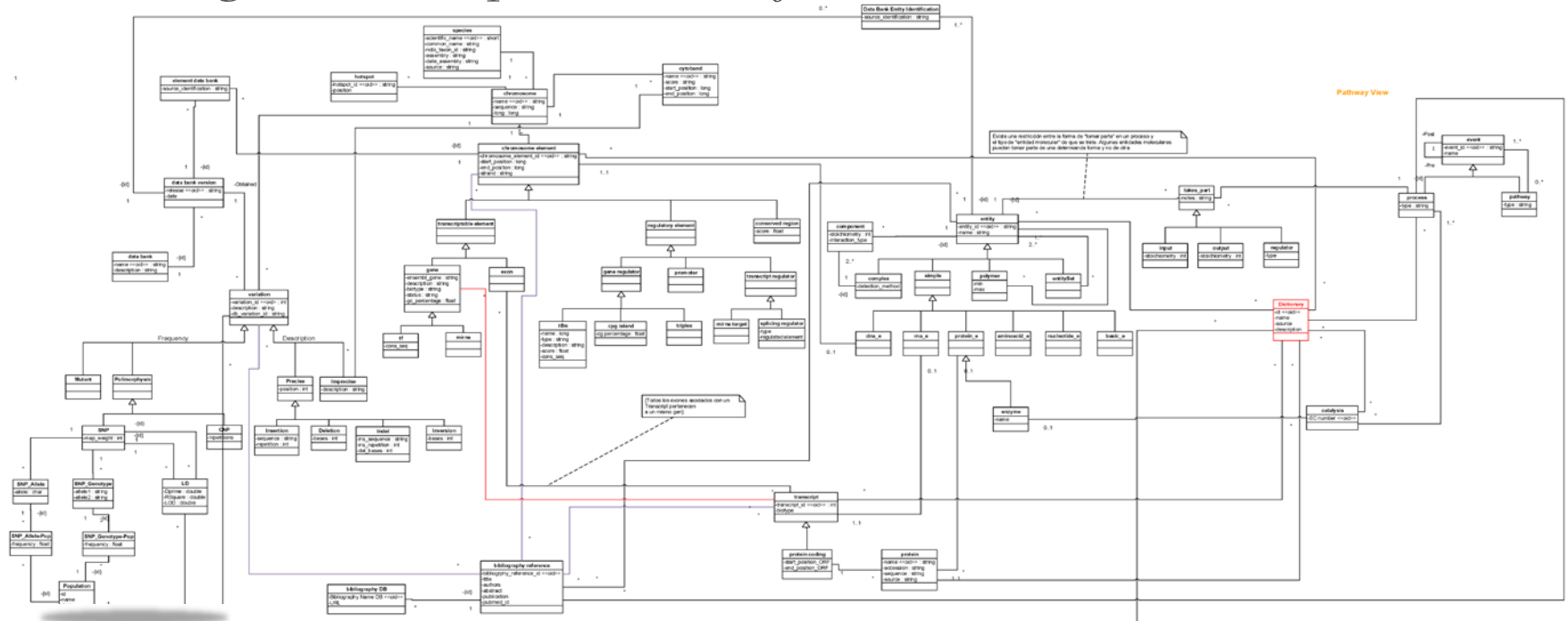
- Motivation
- ¿What does “*haplotypes*” mean?
- Research Methodology
- Conceptual Modeling and Alignment of Haplotypes
- Conclusions & Future Work





Extend our **Conceptual Schema of the Human Genome (CSHG)** to include the specification of **Haplotypes**.

*Figure 1. Conceptual Schema of the Human Genome, version 3*



**This model should be extended in two ways:**

*Integrating treatment of haplotypes.*

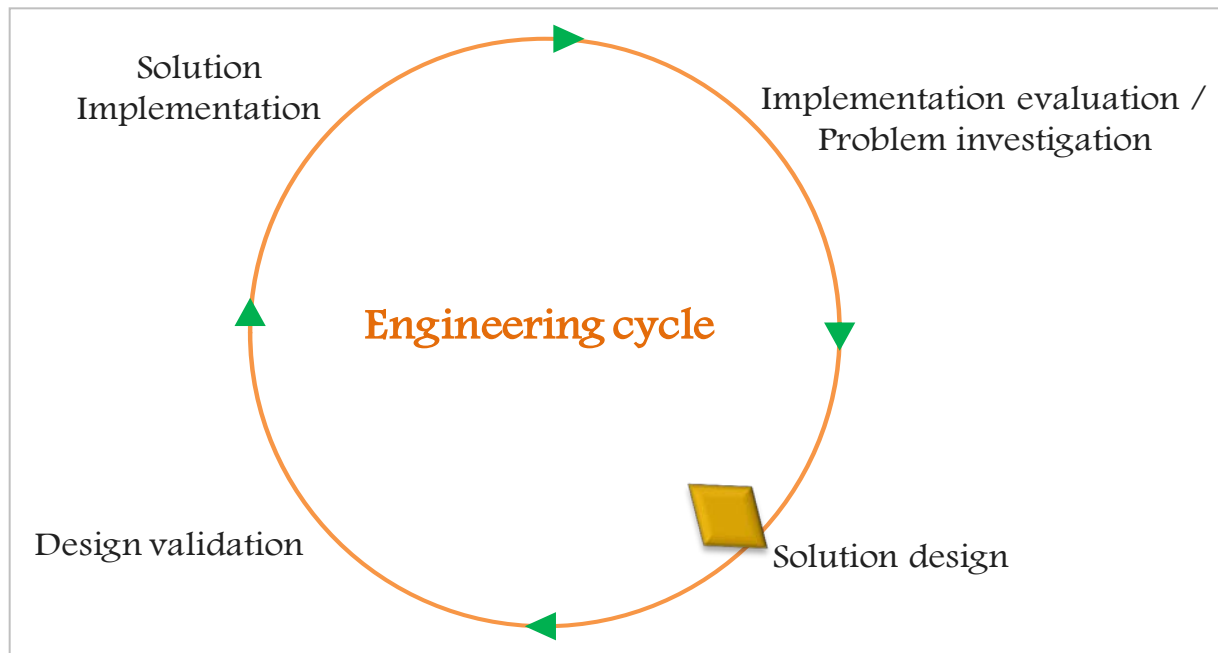
*The application of statistical models.*

# What does “haplotypes” mean?

- From a Bioinformatics point of view, **Haplotypes** are a set of **SNP's** (*single nucleotide polymorphism*) that are **inherited** and found **together** on a **chromosome**.
- Currently, there is a significant set of genetic diseases where the influence of haplotypes has been well-established such as: **breast cancer, alcohol sensitivity**, and others.



- We introduce our research methodology using the –**Engineering Cycle**– proposed by Roel Wieringa, which includes the following steps:

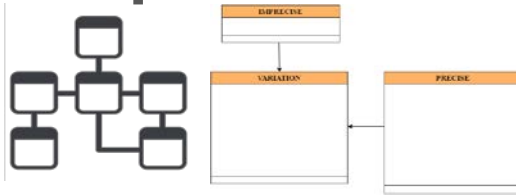


*Figure 3. Engineering Cycle by Roel Wieringa*



# Conceptual Modeling and Alignment of Haplotypes

## Conceptual Schemas



## ANALYSIS



## Databases



To evaluate this work, we have check if the concepts are suitable to support the information provided by popular haplotypes repositories: trying to represent a good conceptual alignment with validated databases from the community.

With this knowledge we can improve the generation of **genetic diagnosis**.



With the aim of providing conceptual models for improving diagnosis in medical contexts where **Genomic Information Systems (GeIS)** play a key role.



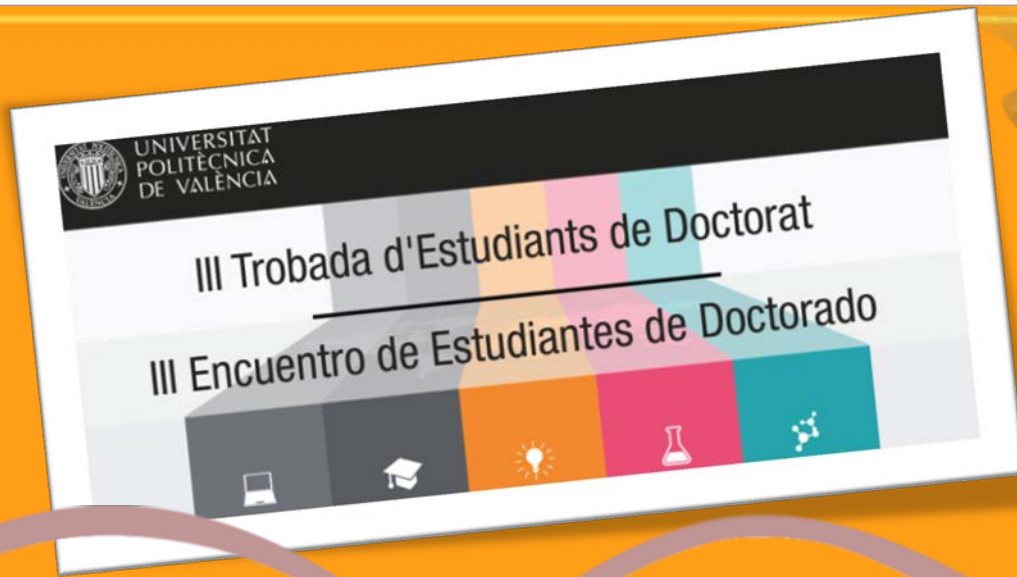


**GRACIAS** **THANK** **YOU**

**ARIGATO** **SHUKURIA** **BOLZIN** **MERCI**

**JUSPAXAR** **EFCHARISTO** **GOZAIMASHITA** **KOMAPSUNIDA** **MAAKE** **GRAZIE** **MEHRBANI** **PALDIES** **SHUKRIA** **BIYAN** **SHUKRIA** **TINGKI** **TASHAKKUR ATU** **YAQHANYELAY** **SUKSAMA** **EKHMET** **DANKSCHEEN** **GRACIAS** **ARIGATO** **SHUKURIA** **BOLZIN** **MERCI** **JUSPAXAR** **EFCHARISTO** **GOZAIMASHITA** **KOMAPSUNIDA** **MAAKE** **GRAZIE** **MEHRBANI** **PALDIES** **SHUKRIA** **BIYAN** **SHUKRIA** **TINGKI** **TASHAKKUR ATU** **YAQHANYELAY** **SUKSAMA** **EKHMET** **DANKSCHEEN**

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