The 1st International Workshop on Conceptual Modeling for Life Sciences (CMLS) in conjunction with the 39th International Conference on Conceptual Modeling (ER 2020)

Towards the Generation of a Species-Independent Conceptual Schema of the Genome

Alberto García S. and Juan Carlos Casamayor

UNIVERSITAT POLITÈCNICA DE VALÈNCIA

PROS
Centro de Investigación en Métodos de Producción de Software

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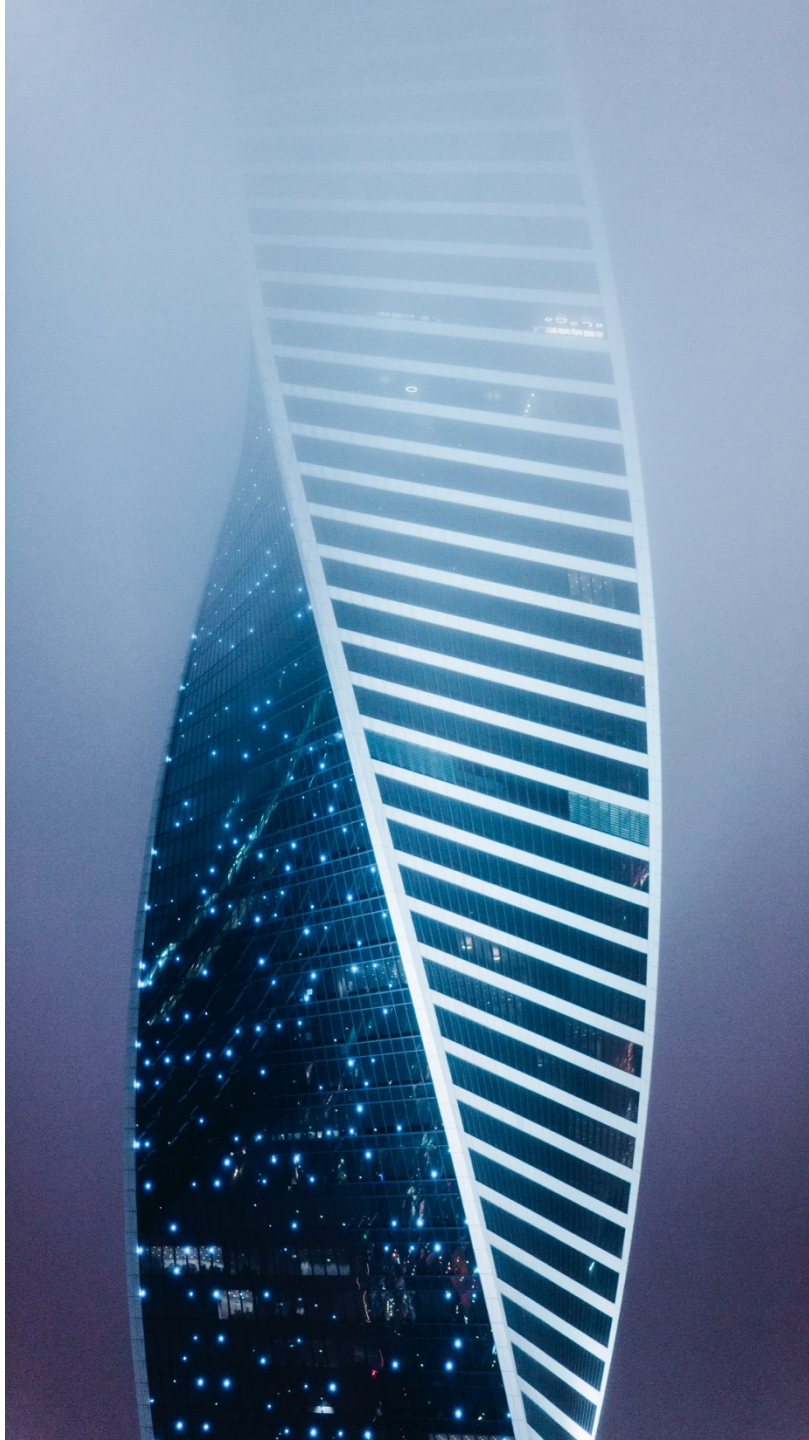
INTRODUCTION

What is the Genomic Domain?

- It is an interdisciplinary field of biology.
- It studies of genomes as a whole and other intragenomic interactions.
- It focuses on understanding the internal processes that drive life.
- It uses multiple approaches: structure, functionality, evolution, edition …



INTRODUCTION

! Why is Conceptual modeling **specially necessary** in the Genomic Domain?

- Relevant concepts are not clearly defined.
- The ever-changing nature of the domain, with new knowledge emerging continuously.
- Data-intensive domain where the information is exceedingly heterogeneous, dispersed, and isolated.

INTRODUCTION

What is our **previous work** in the Genomic Domain?

- Improving precision medicine. The Conceptual Schema of the Human Genome (CSHG) is used in this context.

- Collaborating with agro-food researchers. The Conceptual Schema of the Citrus Genome (CSCG) is used in this context.

INTRODUCTION

? What **lessons** have we **learned**?

- Having different, independent conceptual schemes depending on the species increases complexity, and it is time-consuming.
- Can a holistic conceptual schema, valid to work with different species, be obtained?
- Can species-specific conceptual views be instantiated from such global schema?

A HOLISTIC CONCEPTUAL SCHEMA

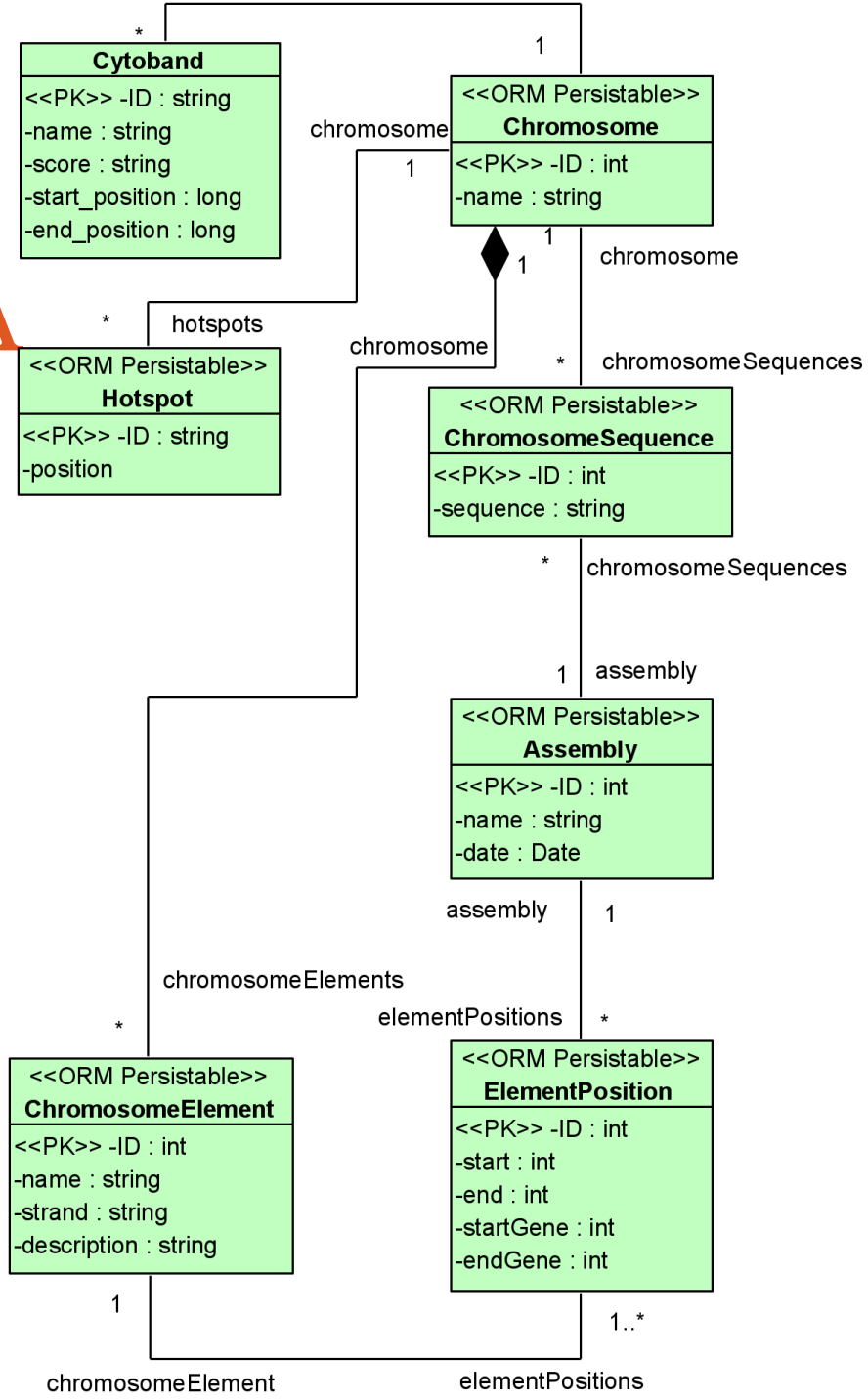
How to **achieve** our **goal**?

- We have compared both conceptual schemes to identify the needed changes to provide an expanded CS.
- Conceptual views from this CS are instantiated for both use cases.
- The CSHG has been used as a basis.
- The CSCG has been used as a complement to expand the CSHG.

A HOLISTIC CONCEPTUAL SCHEMA

What are our **findings**?

1. **Structural view:**
   - both schemes follow a similar approach.
   - The CSCG is more technologically oriented.
   - The CSCG offers a mechanism to investigate evolutionary relationships among citrus.

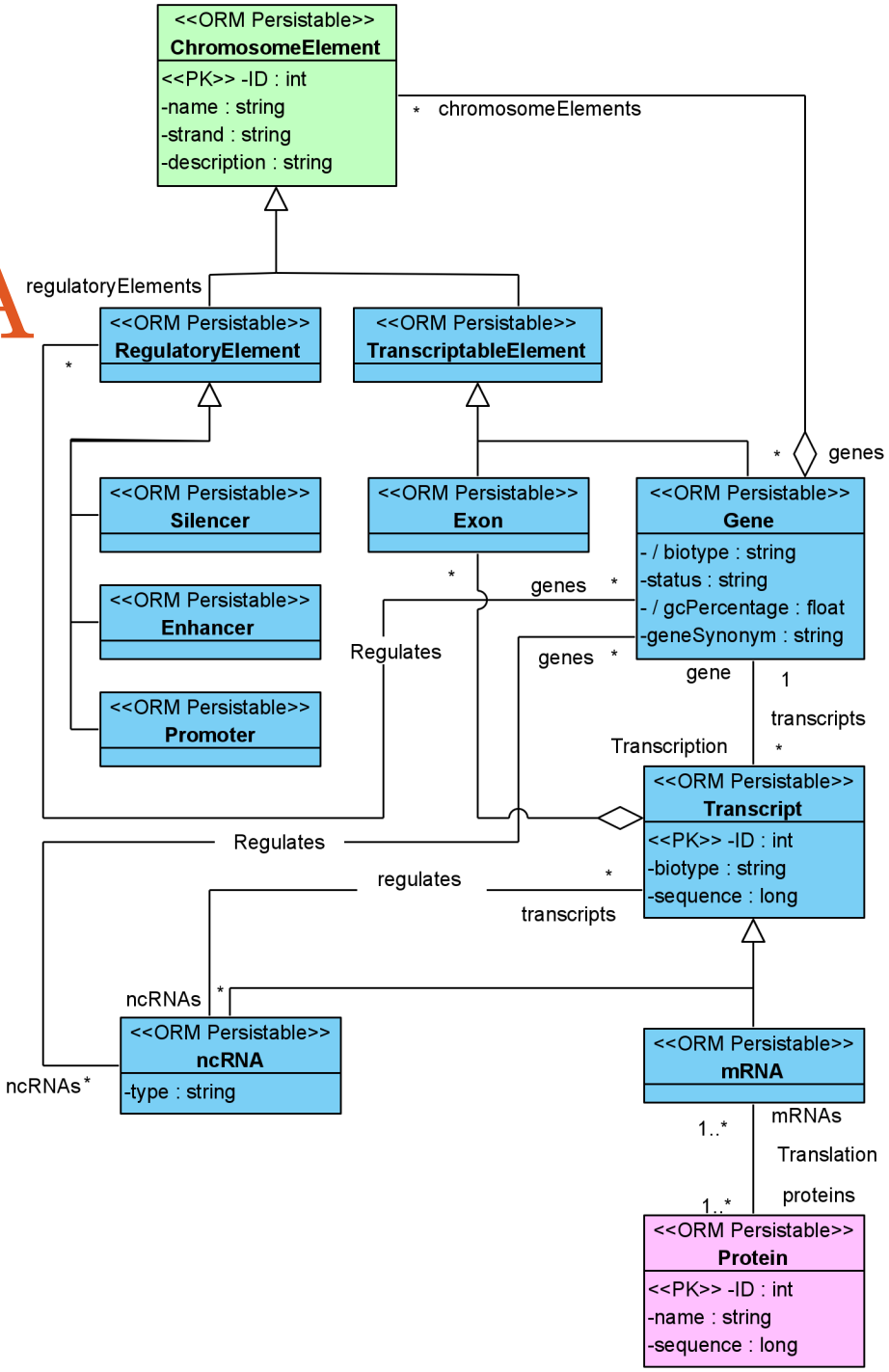


A HOLISTIC CONCEPTUAL SCHEMA

What are our **findings**?

2. **Transcription view:**

- The CSCG models the transcription process in a more simple way.
- Non-coding variants are more relevant in the CSCG.
- Protein functionality is studied in more detail in the CSCG.
- The structure of mRNAs is modeled in the CSCG

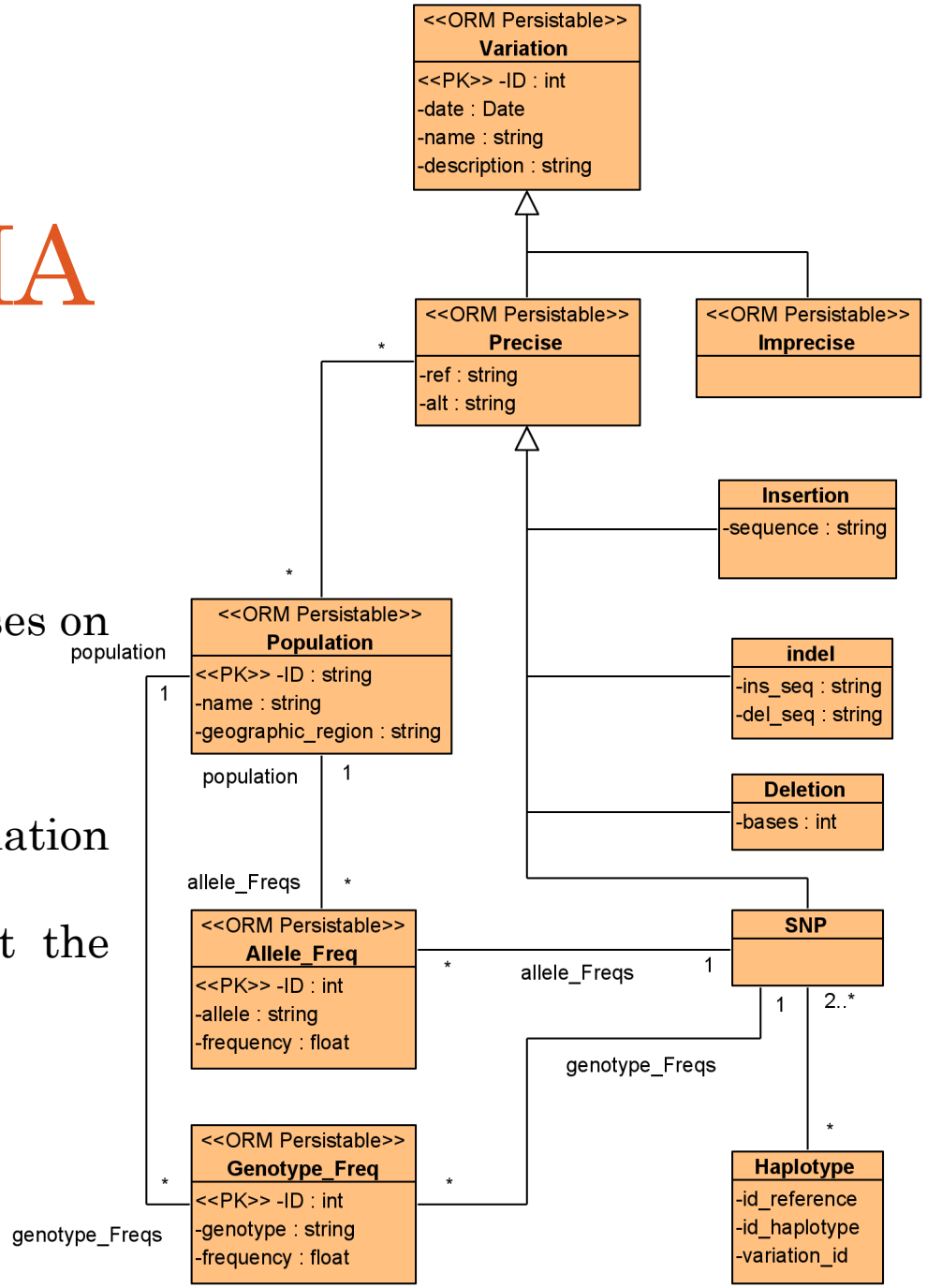


A HOLISTIC CONCEPTUAL SCHEMA

What are our **findings**?

3. **Variation view:**

- the CSHG focuses on SNPs while the CSCG focuses on SNPs and INDELs.
- The CSHG focuses on variations at the population level while the CSCG focuses on variations at the individual level.



CONCLUSIONS

- ✓ The specific particularities of genomic use cases result in too **diverse data analytics**.
- ✓ One of these particularities is the working species, which results in the existence of **multiple conceptual schemes**.
- ✓ It is feasible to have a **holistic CS** that is valid for different species.
- ✓ Specific **views** can be generated from such a holistic schema, **increasing modeling efficiency**, and **knowledge generation**.

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Alberto García S.

Phd Student

Email: **algarsi3@pros.upv.es**

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