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## Genome Bioinformatics at GDEC

Frédéric Choulet

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# Genome Bioinformatics at GDEC

*Frédéric Choulet*



INRAE

UCA  
UNIVERSITÉ  
Clermont  
Auvergne





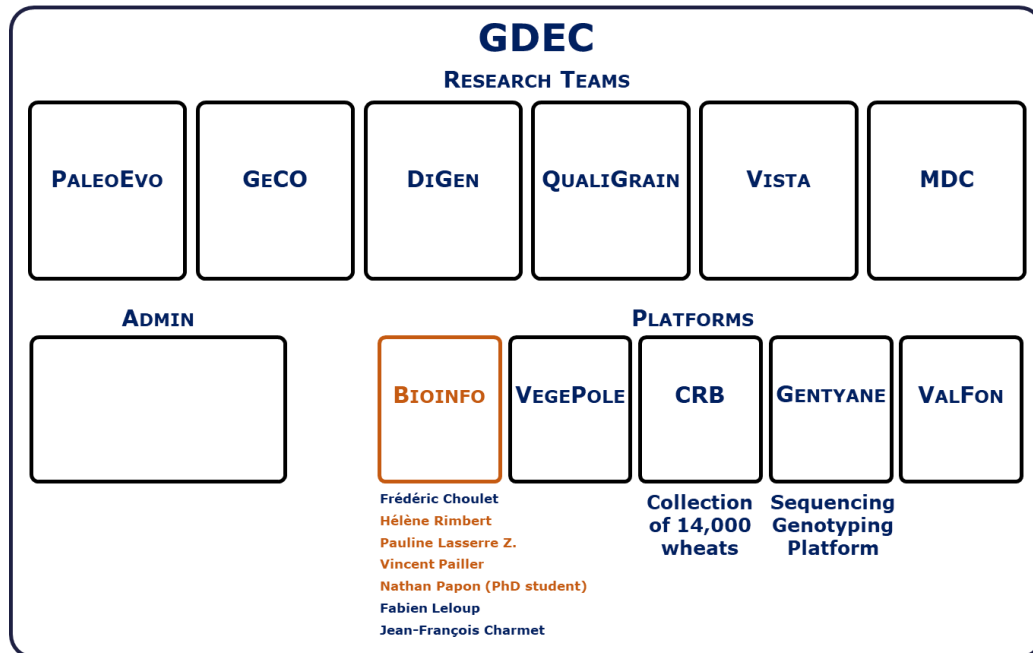
LA GLACIERE  
 CHANTERANNE  
 LA GAUTHIERE  
 METRO  
 Aulnat  
 Lycée Professionnel Roger Claustres  
 Musée d'Art Roger Quilliot  
 Parc Montluzet  
 Usine Michelin de Cataroux  
 Jardiland  
 LE VIEUX MONTFERRAND  
 Maison des Sports de Clermont Ferrand  
 CPAM du Puy-de-Dôme  
 Polydôme Clermont Ferrand  
 L'Avventure Michelin  
 McDonald's  
 McDonald's  
 Hôtel (F) Clermont Ferrand est  
 Intermarché SUPER Clermont Ferrand et  
 LA GARE  
 Casino hyperfrails  
 Leroy Merlin Clermont Ferrand  
 E. Leclerc CLERMONT FERRAND  
 Cathédrale Notre-Dame de l'Assomption  
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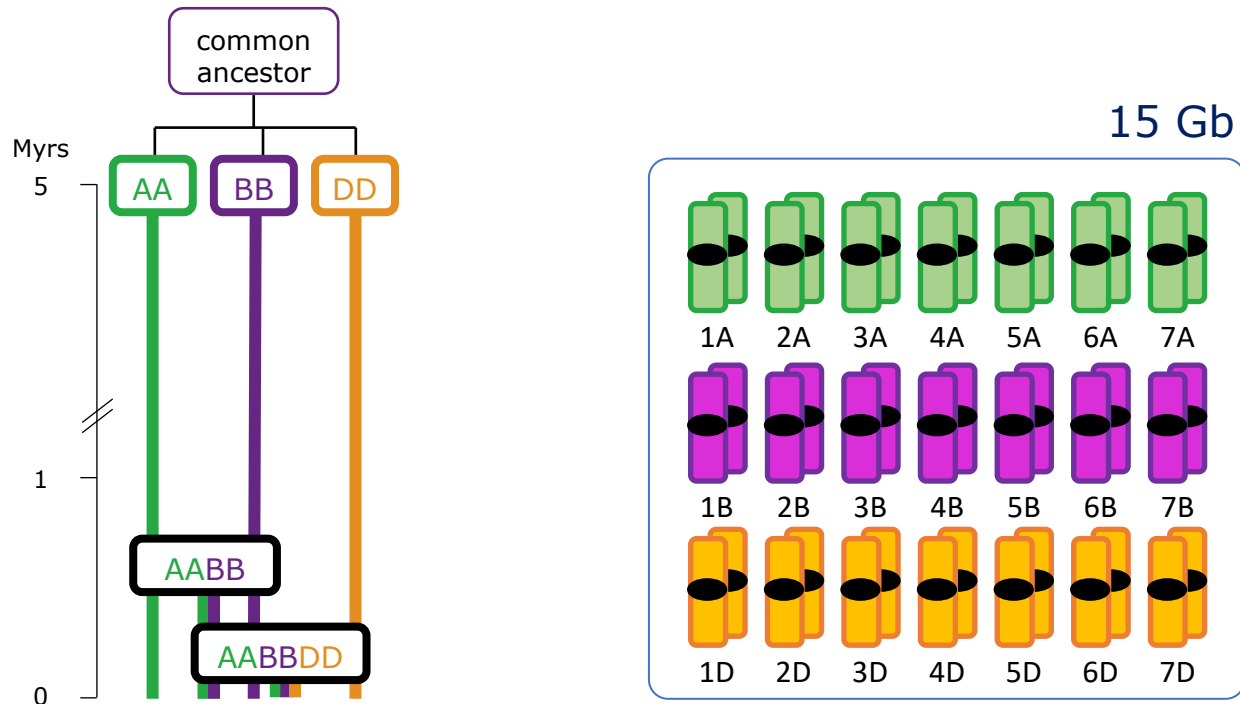
- Bioinfo for bread wheat and *Triticeae* (pan)genomics



# ❖ Research area and questions

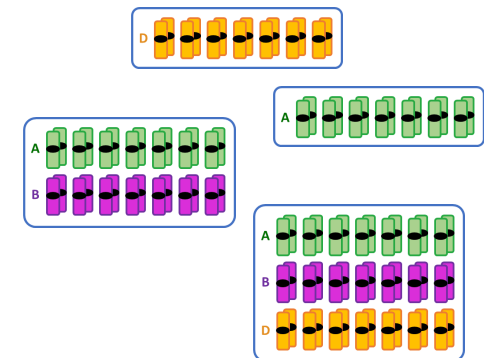
- Methodologies
  - Gene and transposon prediction in genomes
  - Comparative genomics
  - How to compare “complex” genomes?
- Genome Biology
  - What is the extent of diversity? Pangenomics
  - What is the dynamics of transposons?
  - What is the impact of polyploidy on transposon activity?
  - ...

# ❖ Hexaploid genome



## *Triticum/Aegilops*

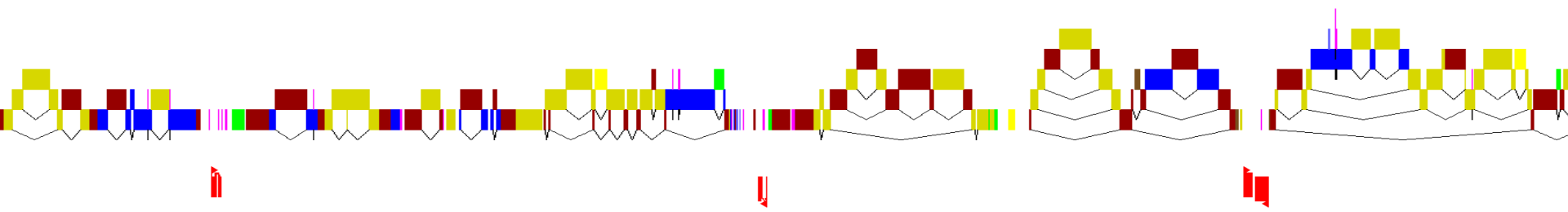
- Wild **diploids** (AA, BB, DD, SS, UU, NN, ...)
- Wild & cultivated **tetraploids** (AABB, DDNN, ...)
- Cultivated **hexaploids** (AABBDD)





# ❖ Genome composition

- Transposons (85%)
- Genes (2%)



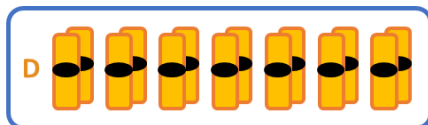
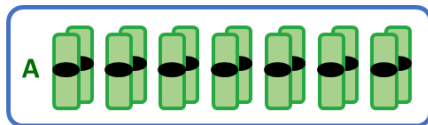
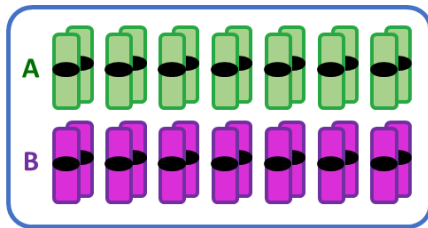
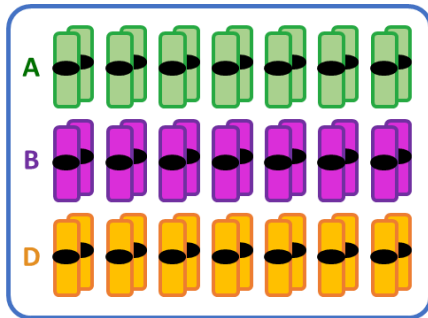
1<sup>st</sup> genome seq assembled:

**Chinese Spring** (*IWGSC Science 2018*)

- 4 million TEs
- ~107,000 genes



# ❖ Pangenomics



- *T. aestivum*

- **Chinese Spring** (Illumina)
- 10 other varieties (Illumina)
- Zang1817 (Illumina)
- Fielder (Pacbio)
- Kariega (Pacbio)
- Renan (ONT)
- Attraktion (Pacbio)

*IWGSC 2018*

*Walkowiak et al. 2020*

*Guo et al. 2020*

*Sato et al. 2021*

*Athiyannan et al. 2022*

*Aury et al. 2022*

*Kale et al. 2022*

- *T. durum* **AABB**

- *T. dicoccoides* **AABB**

- *T. urartu* **AA**

- *Ae. tauschii* **DD**

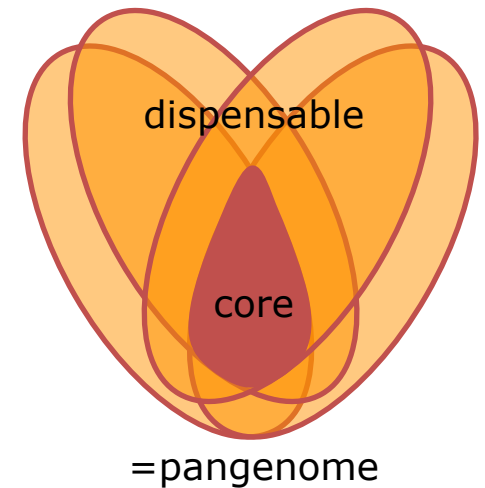
- *Ae. searsii* **DD**

- *Ae. bicornis* **DD**

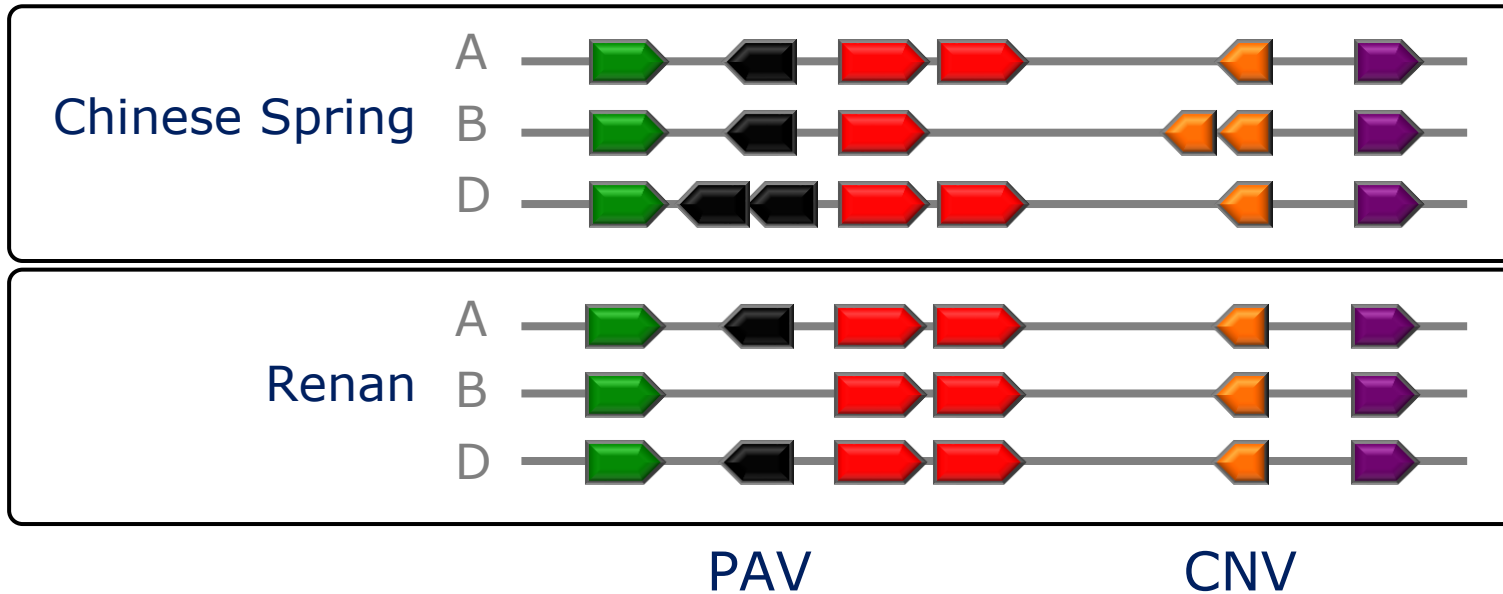
- *Ae. longissima* **DD**

- *Ae. sharonensis* **DD**

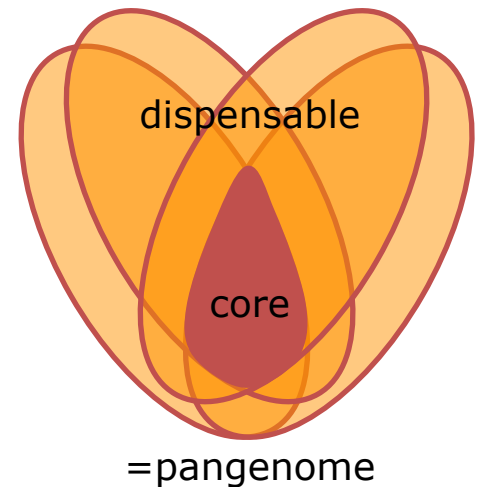
- *Ae. speltoides* **SS**

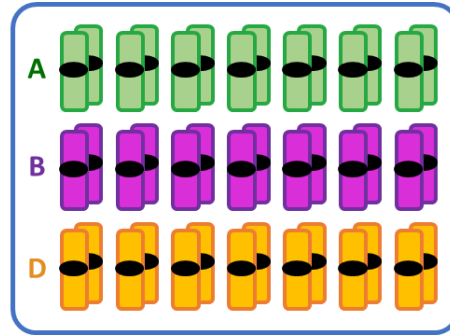


# ❖ Pangenomics

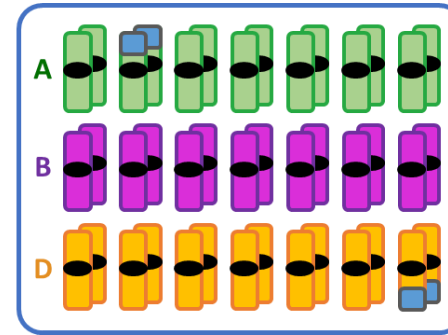


- Chromosome-scale assemblies required
- Quality of annotation matters



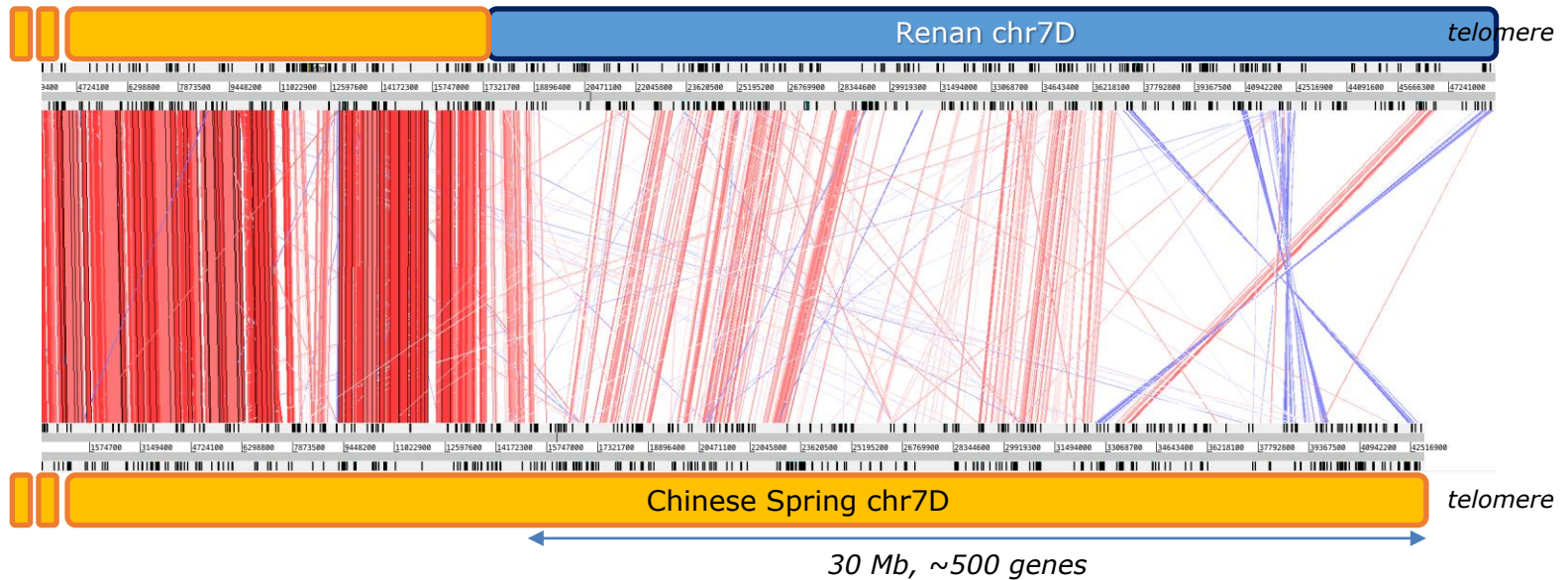


Chinese Spring



Renan

- ✓ Nanopore
- ✓ Illumina
- ✓ Bionano
- ✓ HiC





❖ **Genes**

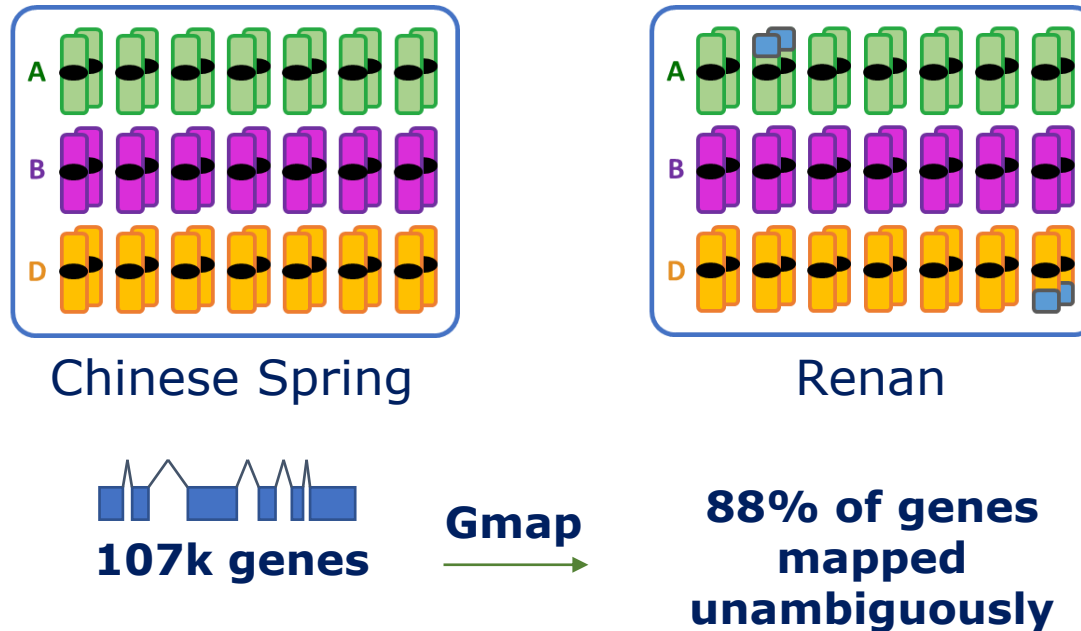
# ❖ Gene annotation

- Predicting genes is not trivial
  - *de novo* gene prediction not adapted to pangenomics



# ❖ Gene annotation

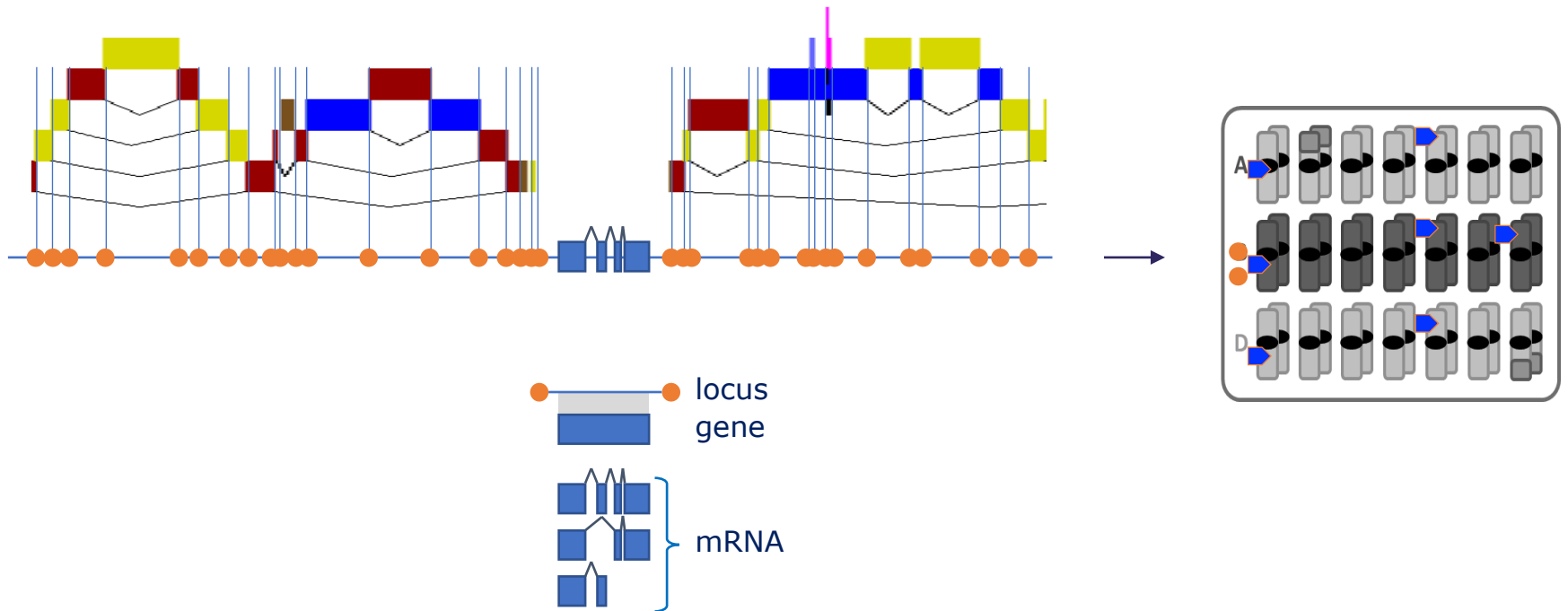
- Predicting genes is not trivial
  - *de novo* gene prediction not adapted to pangenomics
- Mapping a reference gene set sounds better
  - Develop **MAGATT** (*H. Rimbart*)





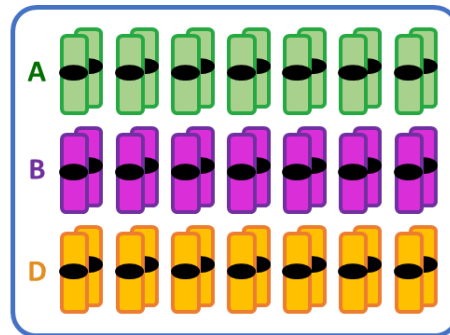
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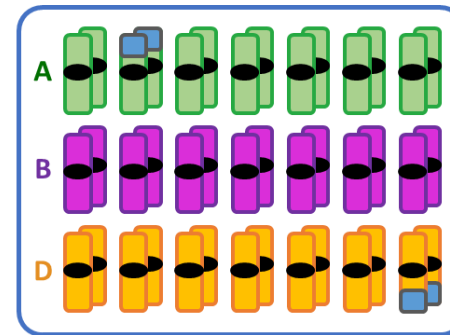


# ❖ Gene annotation

- Predicting genes is not trivial
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Chinese Spring



Renan

  
107k genes

Gmap  
→

88% of genes  
mapped  
unambiguously

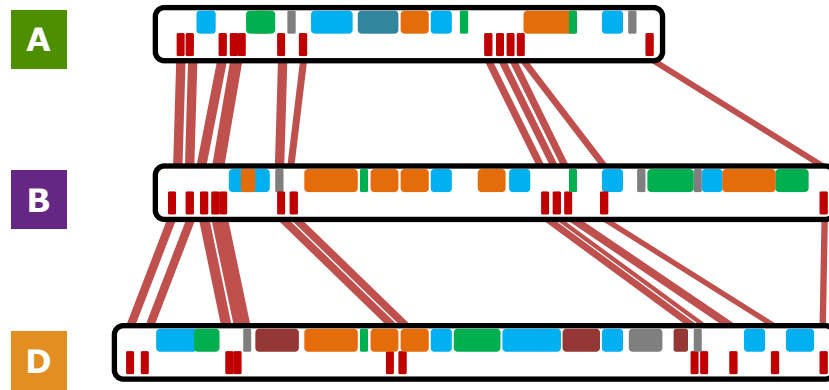
**MAGATT**  
→

**98% of genes  
mapped  
unambiguously**

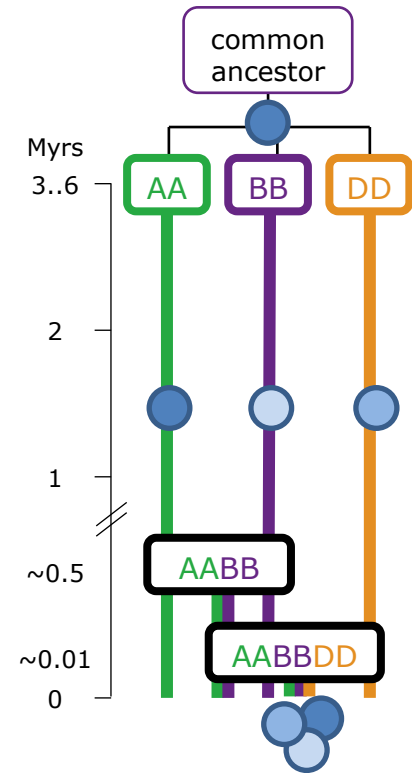
## ❖ **Transposons**



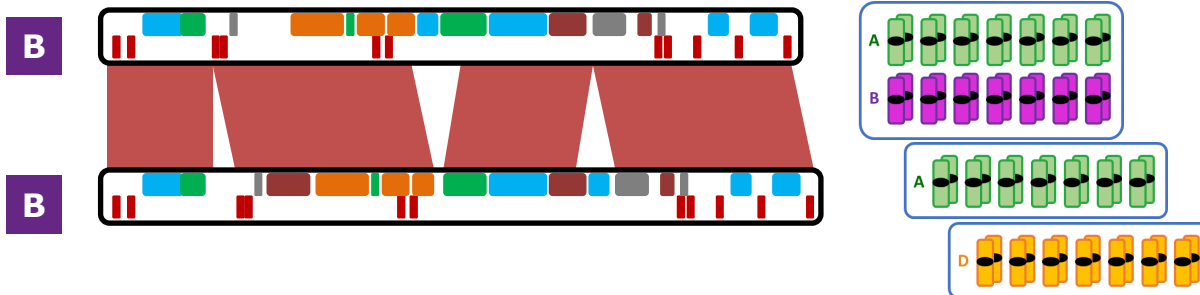
# ❖ Transposon dynamics



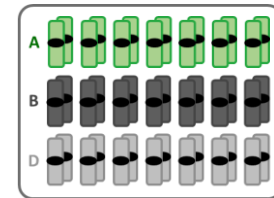
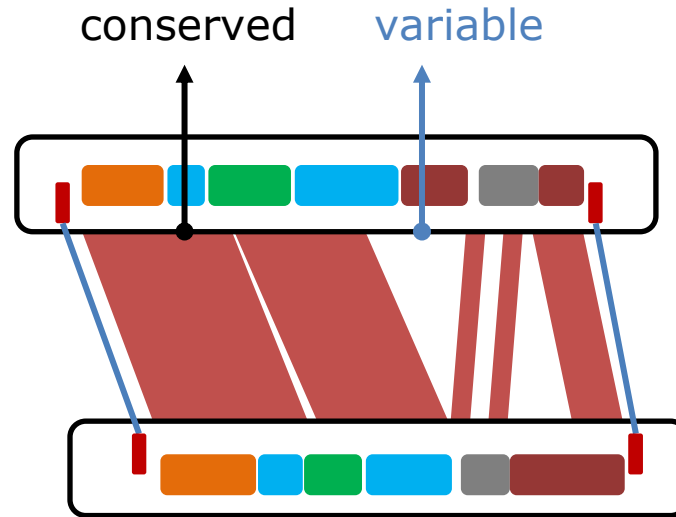
➤ **Equilibrium** model of evolution



PhD Nathan Papon (2019-2023)



# ❖ Transposon dynamics



- **Method: bp-resolved gene anchored whole-genome**

- How variable is the TE space? ➤ 5 .. 15%
- Which TE families are active? ➤ ~ all
- Impact of polyploidization? ➤ none

➤ **Transposition highly regulated; transposons may be a major role in shaping a functional genome architecture**

CSH Cold Spring Harbor Laboratory

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**All families of transposable elements were active in the recent wheat genome evolution and polyploidy had no impact on their activity**

Nathan Papon, Pauline Lasserre-Zuber, H  l  ne Rimbart, Romain De Oliveira, Etienne Paux, Fr  d  ric Choulet  
doi: <https://doi.org/10.1101/2022.11.25.517938>  
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**ABSTRACT**

Bread wheat (*Triticum aestivum* L.) is a major crop and its genome is one of the largest ever assembled at reference-quality level. It is 15 Gb, hexaploid, with 85% of transposable elements (TEs). Wheat genetic diversity was mainly focused on genes and little is known about the extent of genomic variability affecting TEs, transposition rate, and the impact of polyploidy. Multiple chromosome-scale assemblies are now available for bread wheat and for its tetraploid and diploid wild relatives. In this study, we computed base pair-resolved, gene-anchored, whole genome alignments of A, B, and D lineages at different ploidy levels in order to estimate the variability that affects the TE space. We used assembled genomes of 13 *T. aestivum* cultivars (6x=AABBDD), *T. durum* (4x=AABB), *T. dicoccoides* (4x=AABB), *T. urartu* (2x=AA), and *Aegilops tauschii* (2x=DD). We show that 5 to 34% of the TE fraction is variable, depending on the species divergence. Between 400 and 13,000 novel TE insertions per subgenome were detected. We found lineage-specific insertions for nearly all TE families in di-tetra- and hexaploids. No burst of transposition was observed and polyploidization did not trigger any boost of transposition. This study challenges the prevailing idea of wheat TE dynamics and is more in agreement with an equilibrium model of evolution.

**Competing Interest Statement**

The authors have declared no competing interest.

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Michael Alaux (INRAE URGI)  
Thomas Wicker (U Zurich)  
Heidrun Gundlach (PGSB Munich)  
Manuel Spannagl (PGSB Munich)

## ○ *IWGSC*

Kellye Eversole, et al.



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