

Long and short term evolutionary dynamics of transposable elements in the genome of the endangered Apennine yellow-bellied toad (*Bombina pachypus*)

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INTRODUCTION

Transposable elements (TEs) represent a large fraction of vertebrate genomes, impacting genome architecture and evolution. Interestingly, TE could be the driver of genome expansion in some organisms, playing a major role in genome size variation [1-2].

Bombina pachypus is an endangered anuran species endemic to the Italian peninsula, which has experienced a post-glacial range expansion that left a marked signature of **southern richness - northern purity** on its genetic diversity [3].

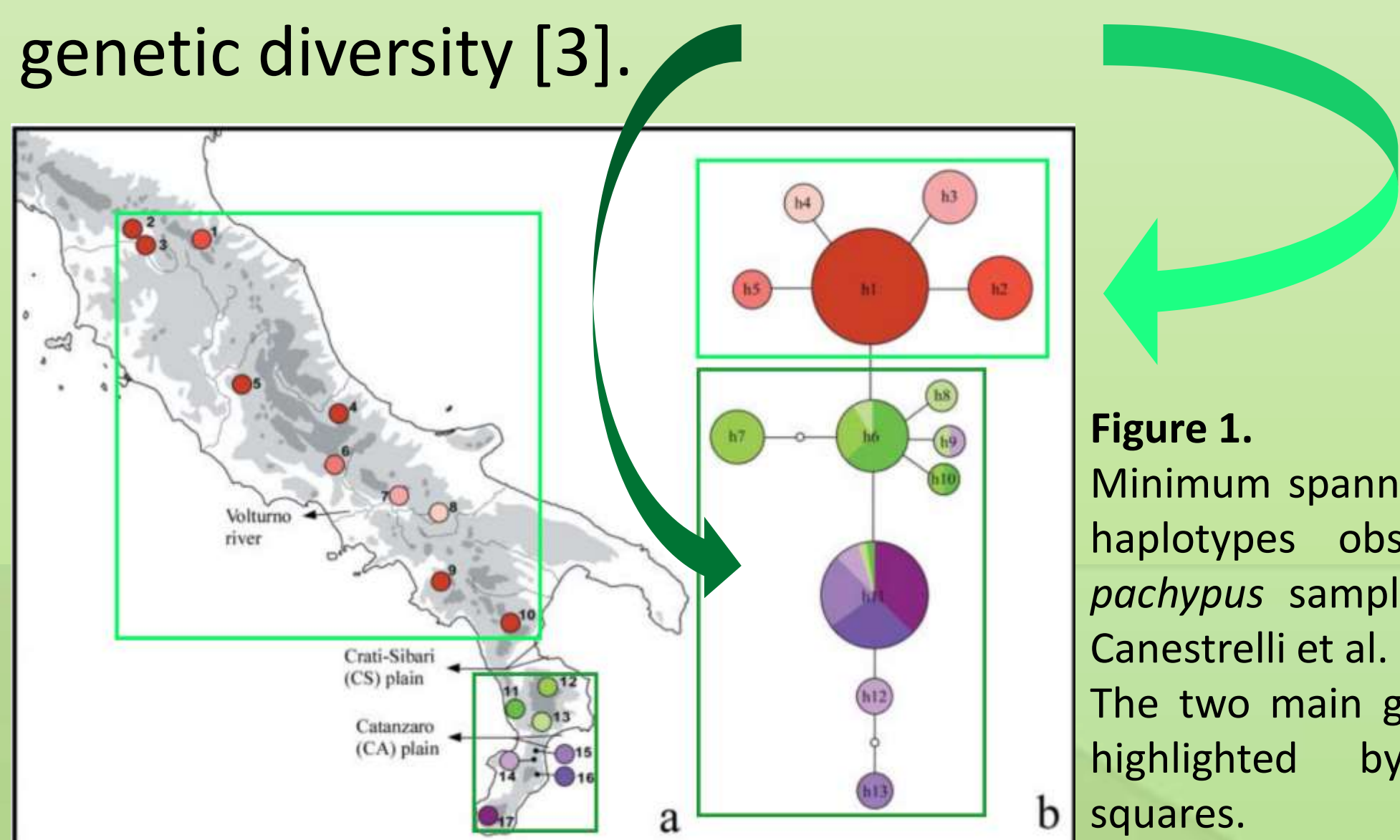


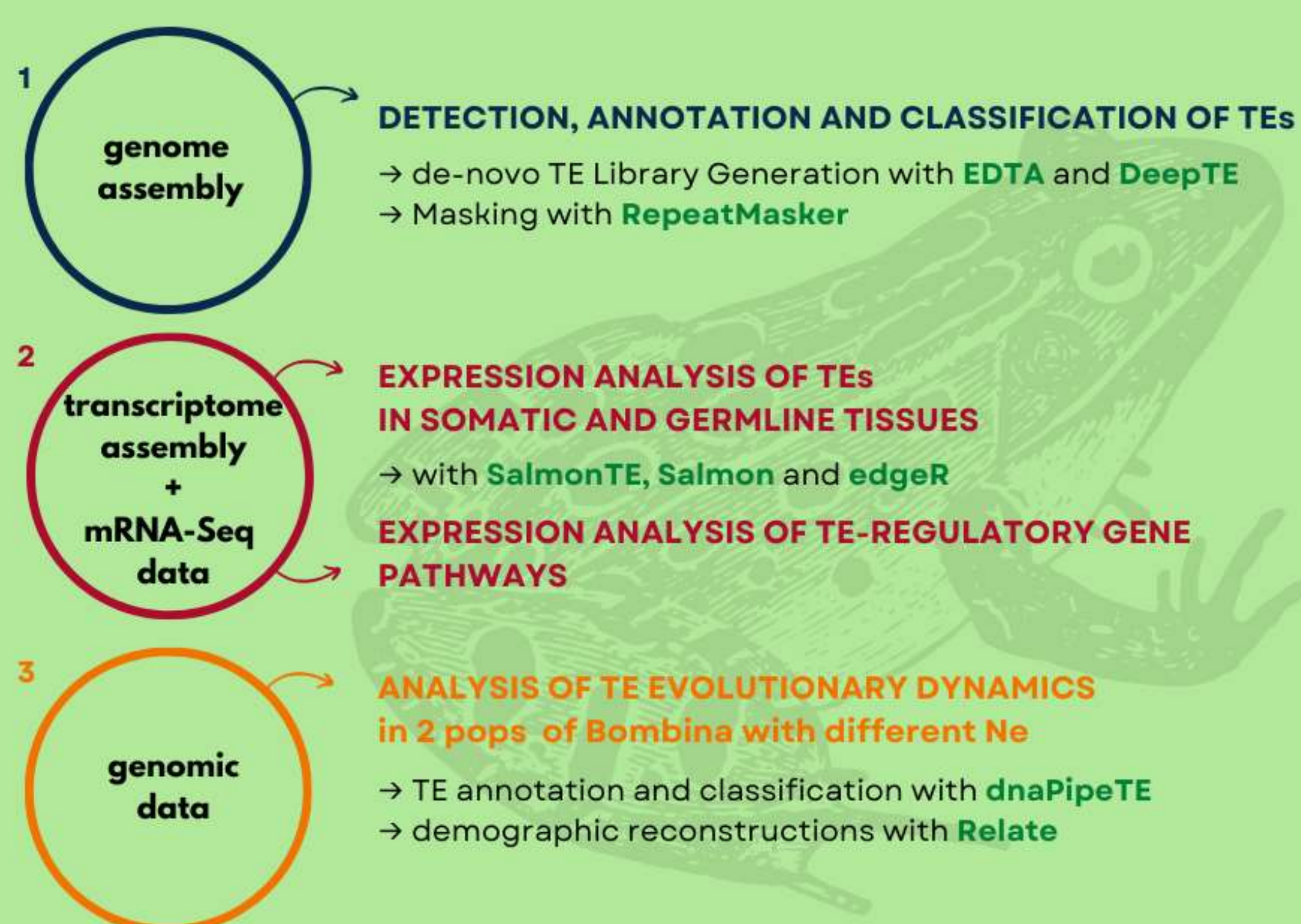
Figure 1. Minimum spanning network for 13 haplotypes observed among *B. pachypus* sampled populations by Canestrelli et al. 2006. The two main genetic clusters are highlighted by two different squares.

The characterization of TEs in the large genome of *Bombina* will allow to explore the genome evolution of this species and to unveil the history of TE-host evolutionary dynamics.

OBJECTIVES

1. Characterize **TE abundance and diversity** in the genome of *Bombina*
2. Investigate the **activity of TE** in somatic and germline tissues and **TE silencing mechanisms**
3. Investigate the **impact of selection on TE dynamics** by comparing two populations of *Bombina* with markedly different effective population sizes
4. Investigate the **correlation between genome size and TE content** with a **comparative analysis** between the 3 orders of amphibians

METHODOLOGY



FIRST YEAR ACTIVITIES

• Expression analyses of TEs

Higher expression of all TE families was found in the **male gonad** compared to the brain, a pattern also confirmed by Pasquesi et al. in a study of recent TE expression in 12 vertebrate lineages [4].

Retrotransposons (Total_ClassI) are the **most active class** in both tissues, with Gypsy family showing the highest activity.

• Differential expression analyses of TEs

Inspecting the differential expressed TEs between the two tissues, the two red clusters in Figure 2b represent the **overexpressed transposons**:

→ in the **brain**, represented mainly by ClassII of **DNA-transposons**, in particular **hAT family**, contrary to the higher proportion of ClassI shown in Figure 2a;

→ in the **gonad**, represented mainly by ClassI of **Retrotransposons**, in particular **Gypsy** and **ERV families**. Looking at the overexpressed DNA-transposons in the gonad, there are few elements of hAT family, probably caused by silencing mechanisms active on this family.

Further statistical tests will be performed to validate the two analyses and the investigation of TE silencing mechanism will allow to clarify the expression dynamics in the two tissues.

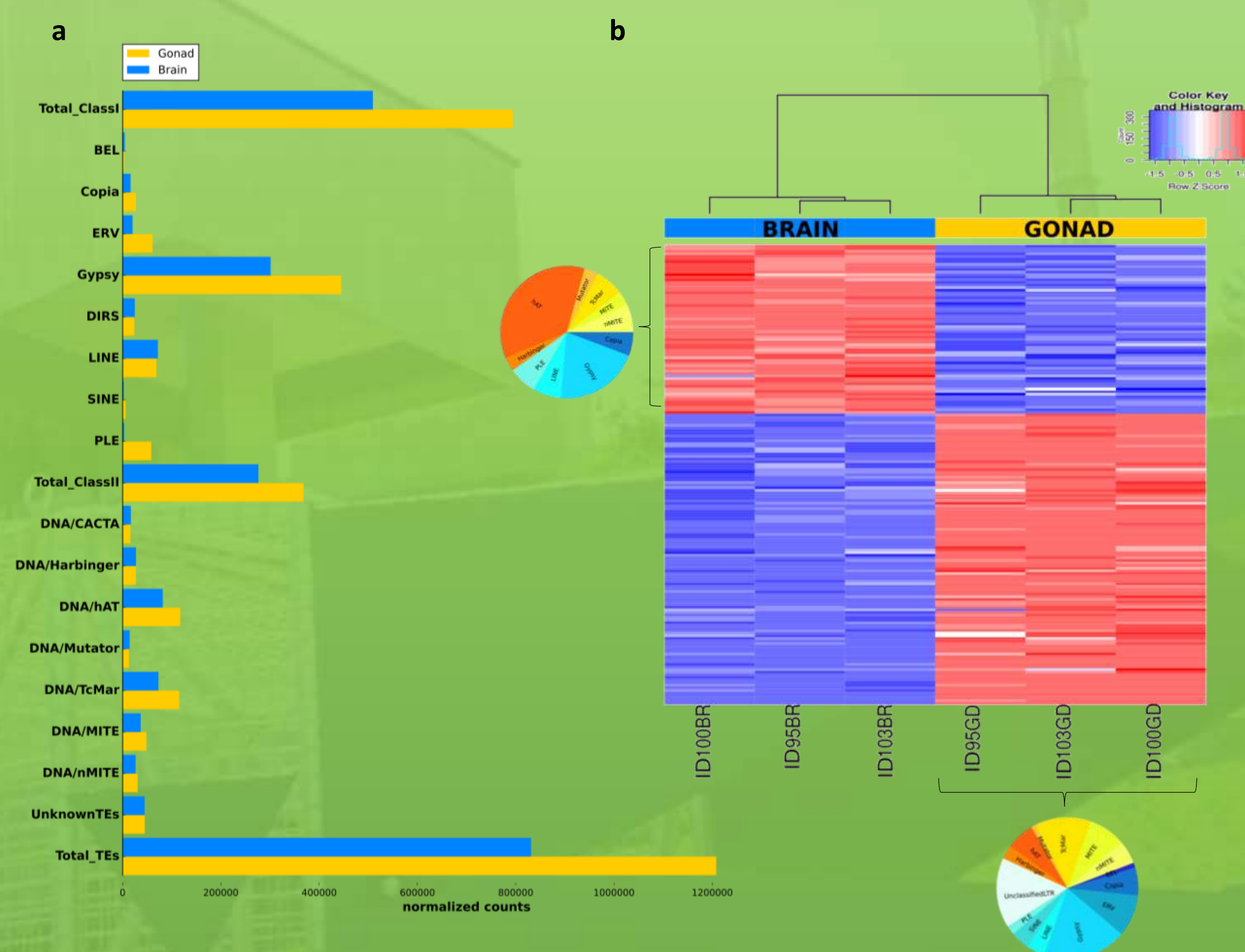


Figure 2.

a) Expression levels of TEs in the transcriptomes obtained from brain and male gonad tissues of *Bombina*: Total_ClassI: total of all Retrotransposons (from BEL to PLE families); Total_ClassII: total of all DNA transposons.

b) Differential expression analysis of TEs between the two tissues: the two red clusters represent overexpressed TEs ($\log_{2}FC > 2$); pie charts show the abundance and diversity of overexpressed TEs.

References

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2. Sun C, Shepard DB, et al. LTR retrotransposons contribute to genomic gigantism in plethodontid salamanders. *Gen Biol Evol* 2012;4:168–83
3. Canestrelli D, et al. Genetic diversity and phylogeography of the Apennine yellow-bellied toad *Bombina pachypus*, with implications for conservation. *Molecular Ecology* 2006; 15:3741–3754
4. Pasquesi G.I.M, et al. Vertebrate Lineages Exhibit Diverse Patterns of Transposable Element Regulation and Expression across Tissues. *Genome Biology and Evolution*, (2020), 12(5):506–521