

Corso di Dottorato di Ricerca in Scienze della Vita e dell'Ambiente - XXXVI

Evolutionary dynamics of transposable elements activity and regulation in the endangered **Apennine yellow-bellied toad (***Bombina pachypus***)** Lorena Ancona; Supervisors: Emiliano Trucchi, Marco Barucca Laboratorio di Genomica, DiSVA

INTRODUCTION

Transposable elements (TEs) are DNA sequences that replicate and mobilise within a host genome, playing a major role in genome size expansion¹.

The evolutionary dynamics interplaying between TE expansion and host silencing machinery are poorly studied in large genomes²⁻³.

Here, we investigate TE and silencing pathways activity in the Apennine yellow-bellied toad (*Bombina pachypus*), an endangered anuran species with a 10 Gb genome, shedding light on the complex dynamics acting between TEs and the host genome.

METHODS

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Transcriptome assembly + mRNA-seq raw reads

TE detection and TE expression

library construction + expression and DE analyses

Characterization and Expression of TE-regulatory gene pathways functional annotation + expression and DE analyses

TE Expression



Fig 1. Expression levels of TEs in *B.pachypus*:

Total_RetroTE: total of all Retrotransposons (from LTR/nc to PLE families); Total_DNATE: total of all DNA transposons; MBR: male brain (blue), FBR: female brain (blue), MG: male gonad (green), FG: female gonad (pink).

Higher expression of **TEs** in the **germline Retrotransposons** → the most active class in MG **DNA transposons** → the most active class in **FG**



Fig 2. Volcano plot showing the up-regulated DE-TEs between female and male gonads: Overexpressed TEs in ovary (pink) and in testis (green). Dotted lines indicate logFC ±2

MALE GONADS:

- → more overexpressed TEs
 - → higher fold changes

TE-Regulatory Gene Pathways Expression



Fig 3. Expression levels of key genes involved in negative regulation of TE activity in *B.pachypus*: MBR: male brain (blue), FBR: female brain (blue), MG: male gonad (green), FG: female gonad (pink).

Tissue-specific strategies in the **gonads**:

Higher expression of **PIWI:piRNA pathway** in male(MG) and female (FG) gonads → related to involvement of these protein in the Ping-Pong cycle that requires ongoing expression of the cluster and target transposons

Higher expression of KRAB-ZFP and NuRD **complex** in female gonad (FG) which correlates with the lower TE expression in FG



Fig 4. Volcano plots showing the up-regulated DEGs (TE-regulatory gene pathways)

between female and male gonads (4a) and between gonads and brain (4b):

Overexpressed DEGs in ovary (pink), testis (green), Female Brain (blue) and Male Brain

(lightblue). Dotted lines indicate $logFC \pm 2$



1. Cibele G. Sotero-Caio, et al. Evolution and Diversity of Transposable Elements in Vertebrate Genomes, Genome Biology and Evolution, Volume 9, Issue 1, 2017, 161–177;

2. Pasquesi GIM, Perry BW, et al. Vertebrate Lineages Exhibit Diverse Patterns of Transposable Element Regulation and Expression across Tissues. Genome Biology and Evolution, 2020, 12(5):506–521;

3. Wang J, Yuan L, et al. Transposable element and host silencing activity in gigantic genomes. Front Cell Dev Biol. 2023 Feb 24;11:1124374.