

Duck pan-genome reveals two transposon insertions caused bodyweight enlarging and white plumage phenotype formation during evolution



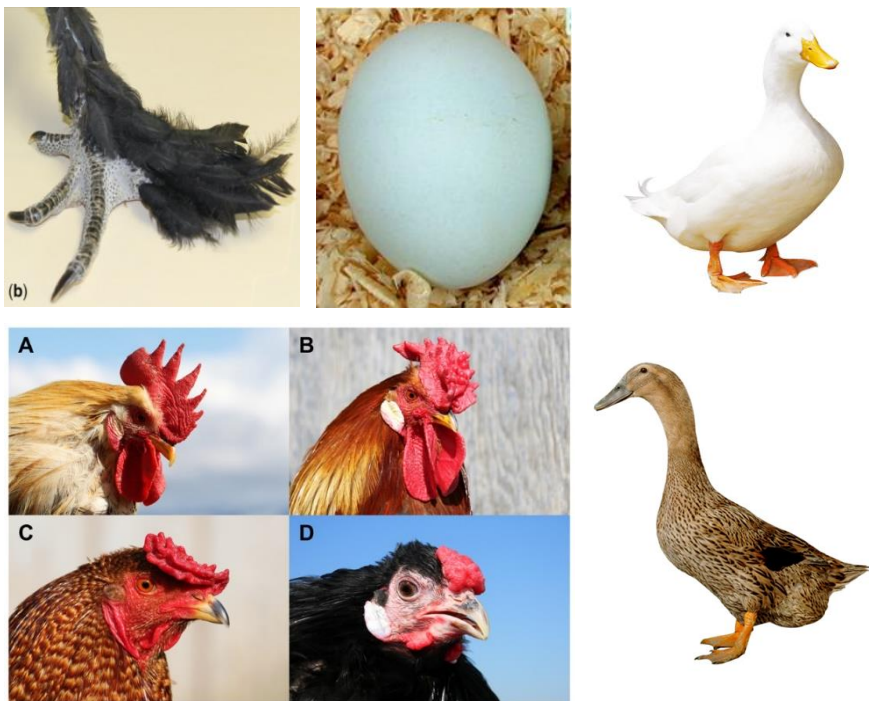
Kejun Wang, Guoying Hua, Jingyi Li, Yu Yang, Chenxi Zhang, Lan Yang, Xiaoyu Hu, Armin Scheben, Yanan Wu, Ping Gong, Shuangjie Zhang, Yanfeng Fan, Tao Zeng, Lizhi Lu, Yanzhang Gong, Ruirui Jiang, Guirong Sun, Yadong Tian, Xiangtao Kang, Haifei Hu, Wenting Li

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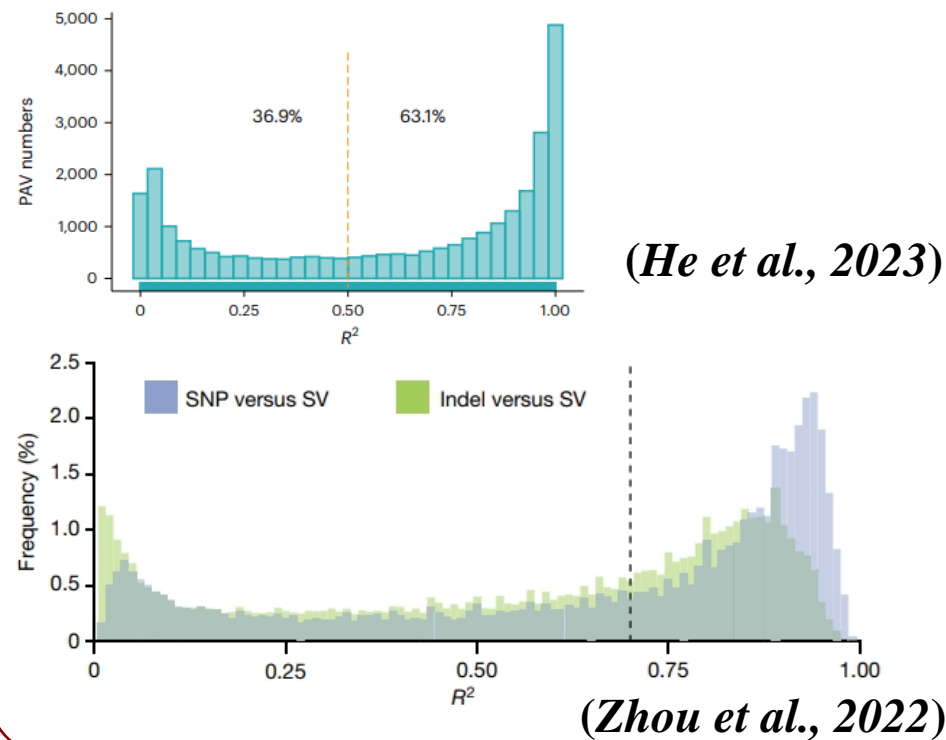
Background



(Imsland et al., 2012)

- **Determined by Structural variation**

Many structural variation showed weaker linkage with adjacent SNP



Background



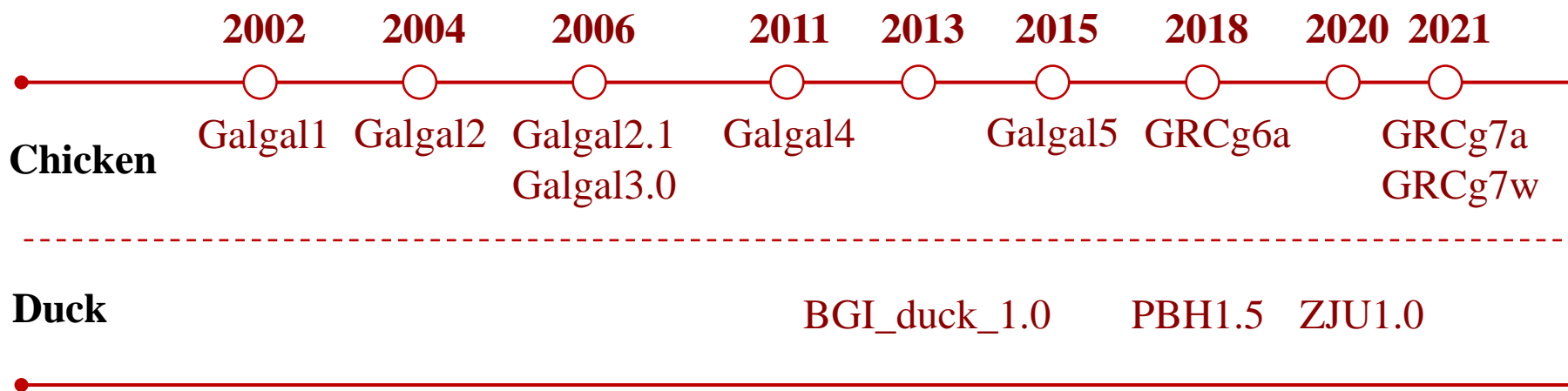
Anas platyrhynchos



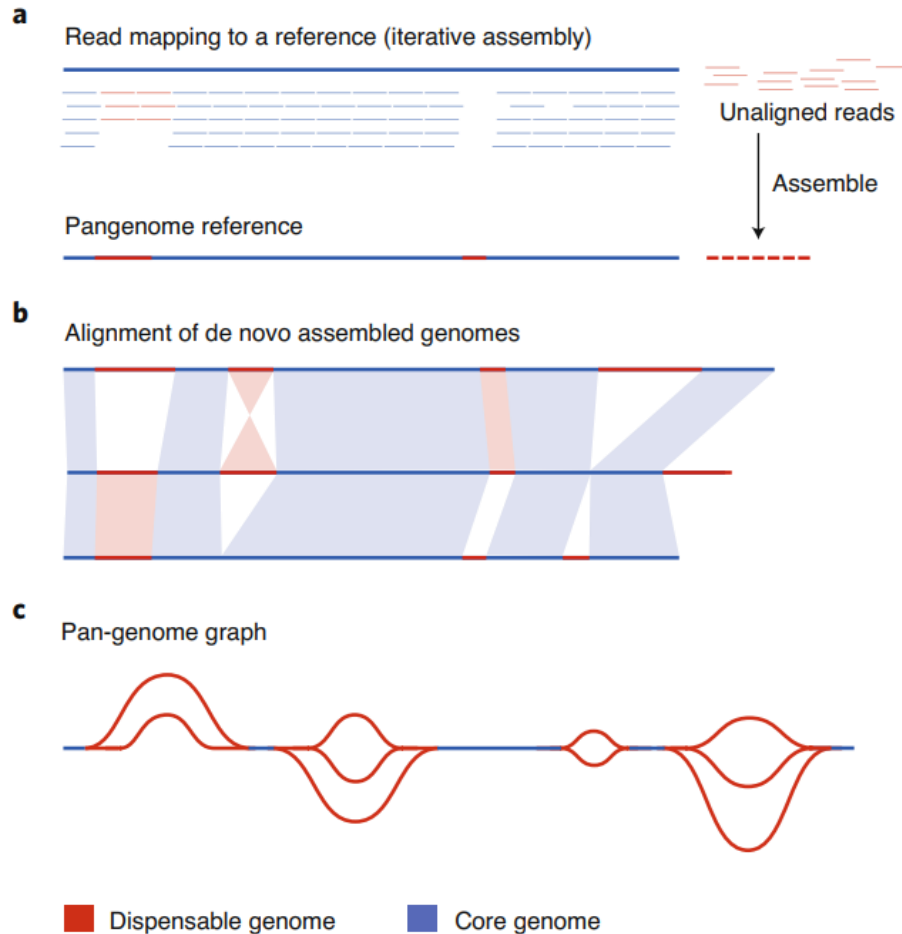
Anas zonorhyncha



Pekin duck



Background



■ Construction strategy of Pan-genome

(1) Linear pan-genome

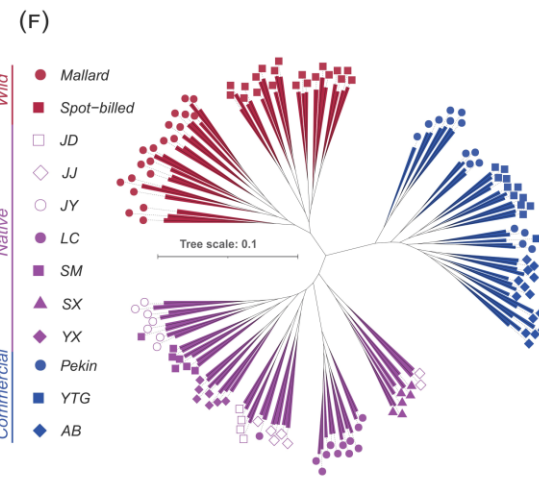
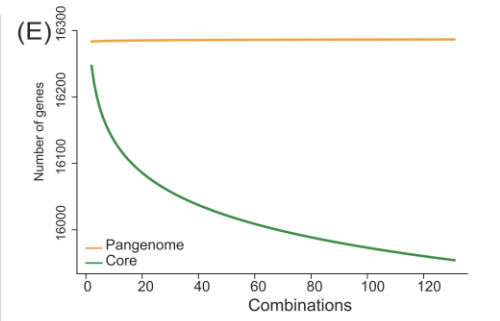
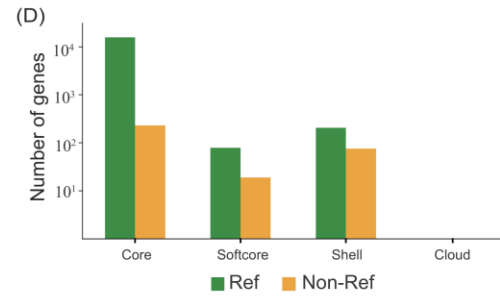
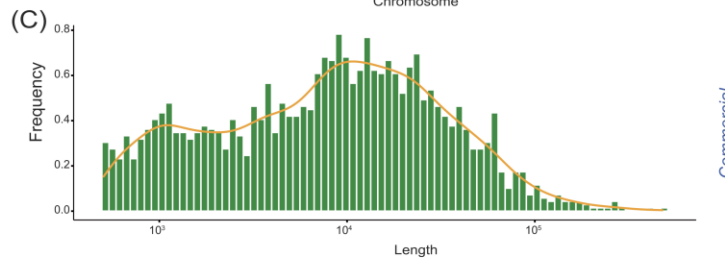
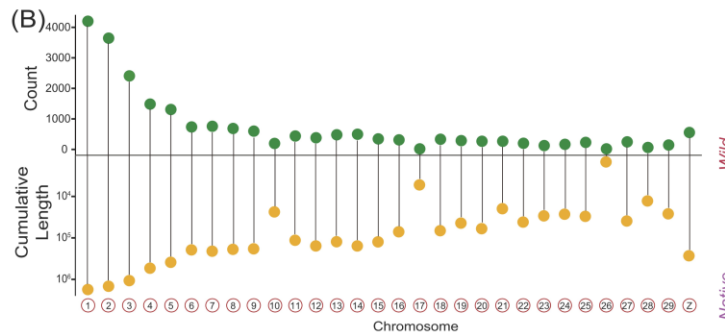
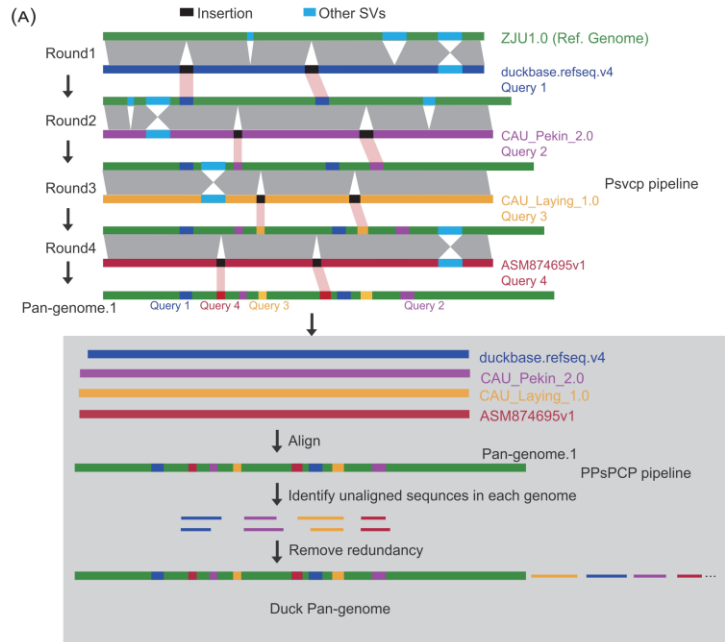
- ✓ Iterative mapping and assembly approach
- ✓ Assembly alignment approach

(2) Graph pan-genome

- ✓ Assembly alignment-based approach
- ✓ Reference + VCF approach



Results



01. Duck pan-genome

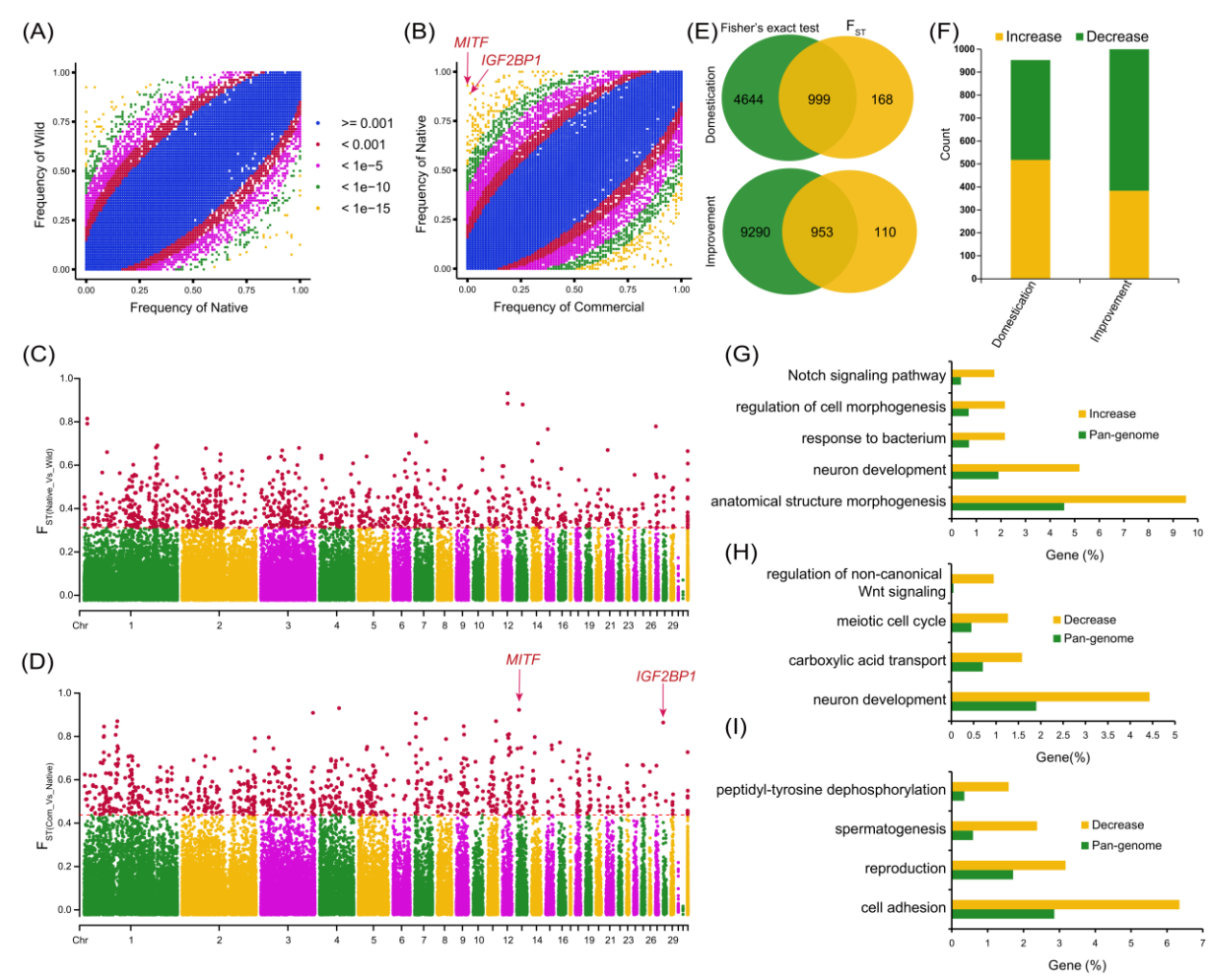
- **Type:** Linear pan-genome
- **Method:** Assembly alignment
- Five published duck genome assembly
- Identified ~40.98Mb novel contig
- **advantage:** novel contig with genome location



Results

02. The impact of domestication and improvement on genomic structural variation.

- **Method:** Fixation index (F_{ST}) 和 Fisher's exact test
- **Domestication:** 999 SVs ~ nervous system development, bacterial response, and morphogenesis
- **Improvement:** 953 SVs ~ reproduction and spermatogenesis function
- **Two Significant SVs adjacent to *IGF2BP1*, *MITF* gene**



Results

03. Transposon-derived structural variations are linked to duck domestication and improvement

● **TE: 24.37% ~ novel contig**

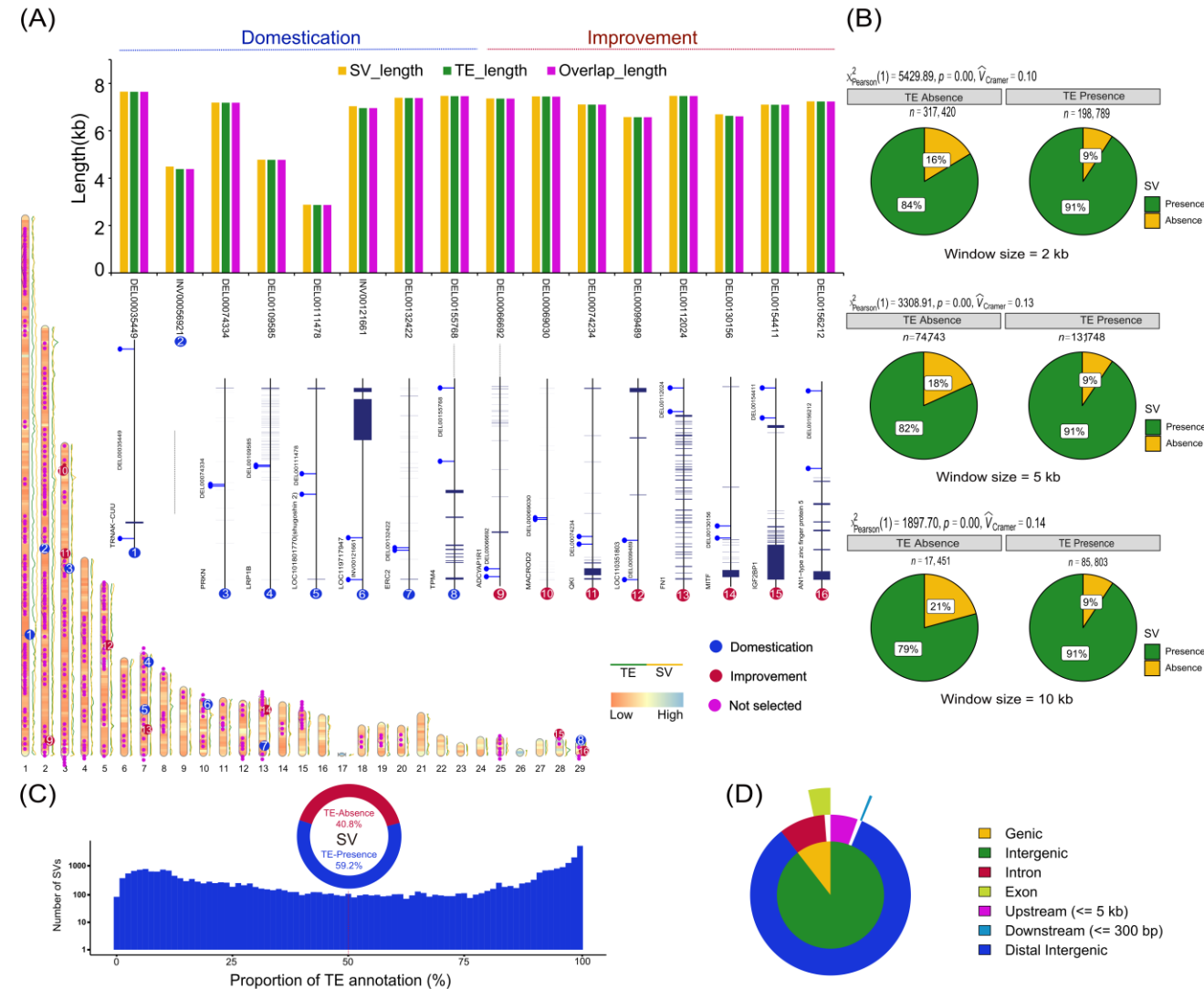
9.49% ~ whole genome

➤ **Chi-square test: SV presence ~ TE presence, 59.2% SVs match at least one TE, of which 58.7% overlapped with TE more than 50%.**

➤ **Intact TE : 392 SV presented 95% matching to TE (included 16 significant)**

***IGF2BP1* ~ DEL00154411**

***MITF* ~ DEL00130156**



Results

04. Insertion of a 6,945 bp Gypsy transposon into the promoter region of *IGF2BP1* increases duck bodyweight

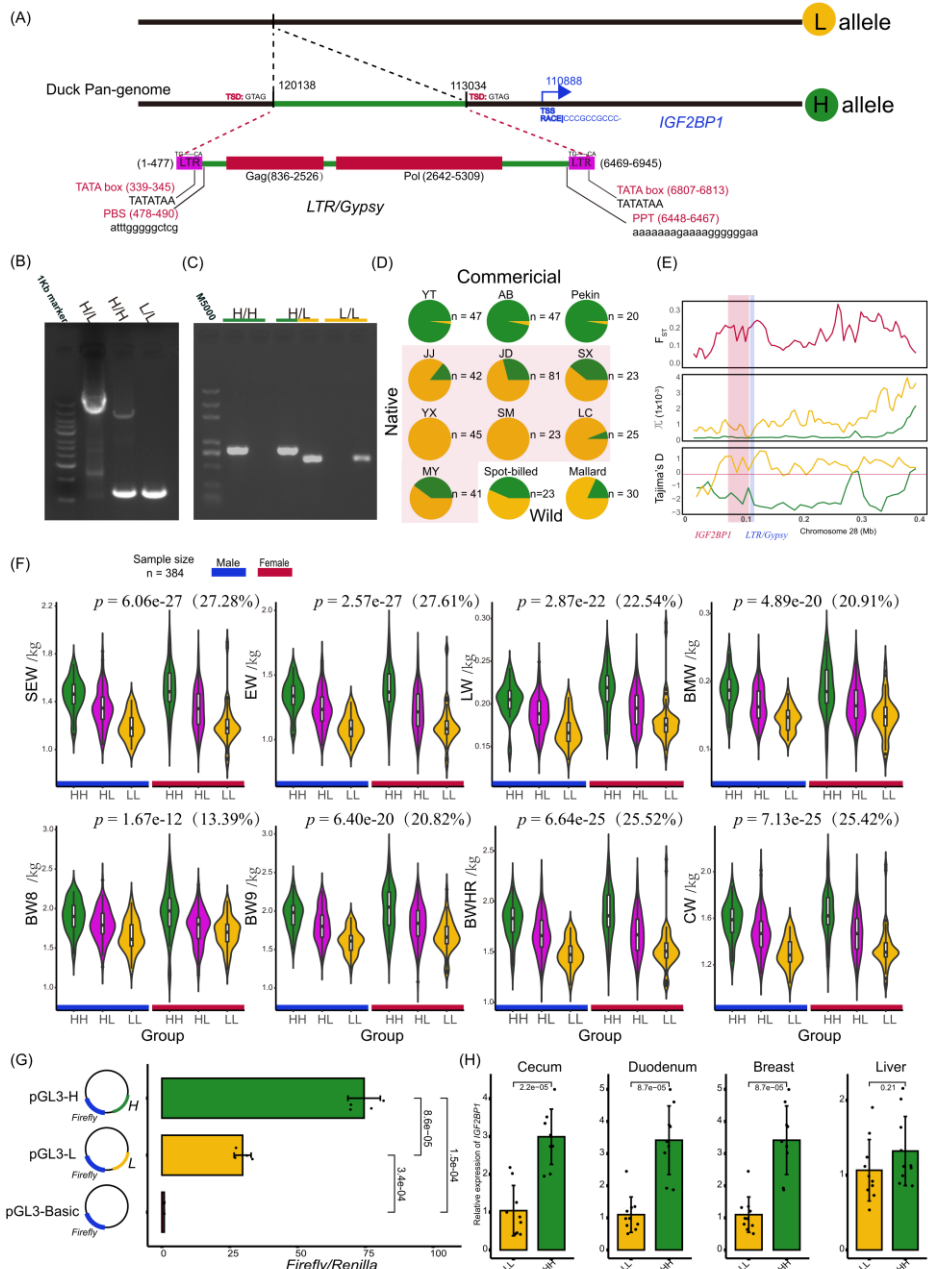
Variant: 6945 bp Gypsy TE located the upstream of *IGF2BP1*

GWAS: Related to BW8, BW9, BWHR, CW, EW, EW, BMW, LW,

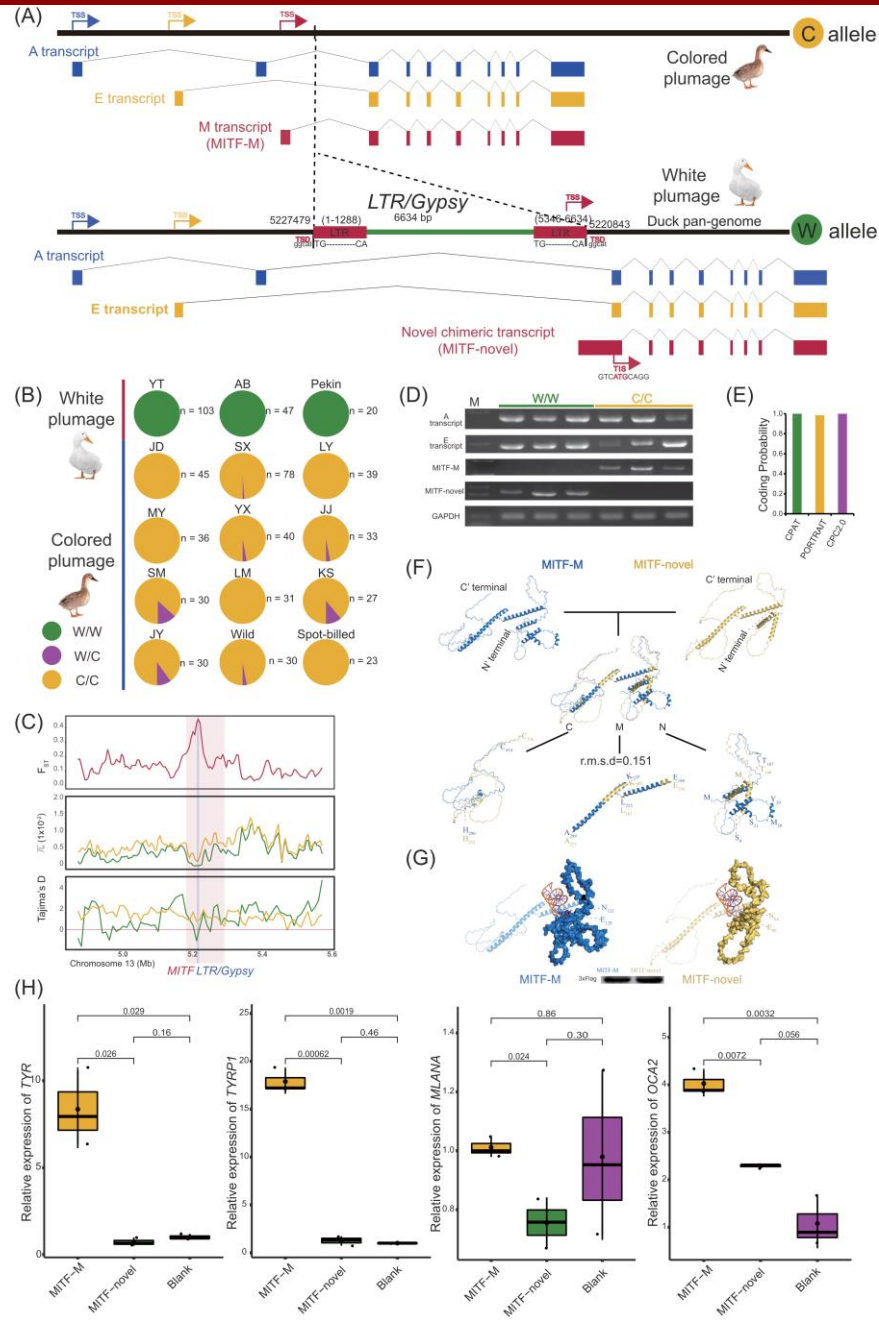
27.61% of phenotypic variation for EW trait.

(Largest effect on bodyweight among avian species)

Pattern: Enhancer for *IGF2BP1* expression regulating the bodyweight



Results



05. A 6,634 bp Gypsy element inserted into the intron of MITF generates a chimeric transcript that contributes to the white plumage phenotype in duck

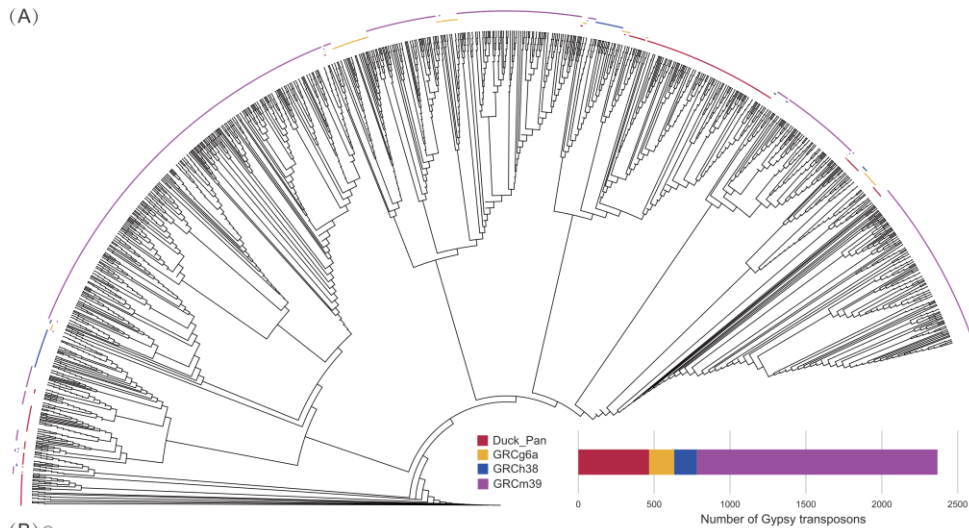
● **Variation:** 6634bp Gypsy TE located at intron of MITF

➤ **Effect:** Generating a chimeric transcript with protein coding ability.

➤ **Pattern:** Loss of function, blocking the melanogenesis pathway and thus causing white plumage.

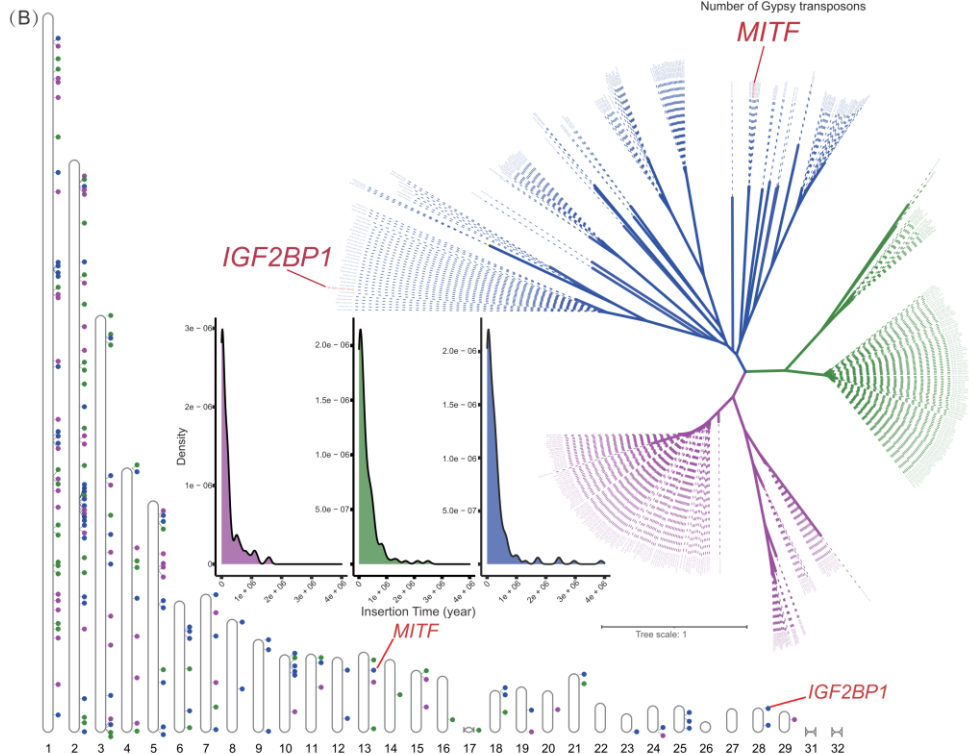


Results



06. Phylogenetic analysis of Gypsy TE

- **TE:** Compared to chicken, an **obvious expansion** in the quantity of Gypsy was identified in the duck genome (467 pan-genome and 431 ZJU1.0 Vs. 165 in GRCg6a)



➤ Phylogenetic analysis :

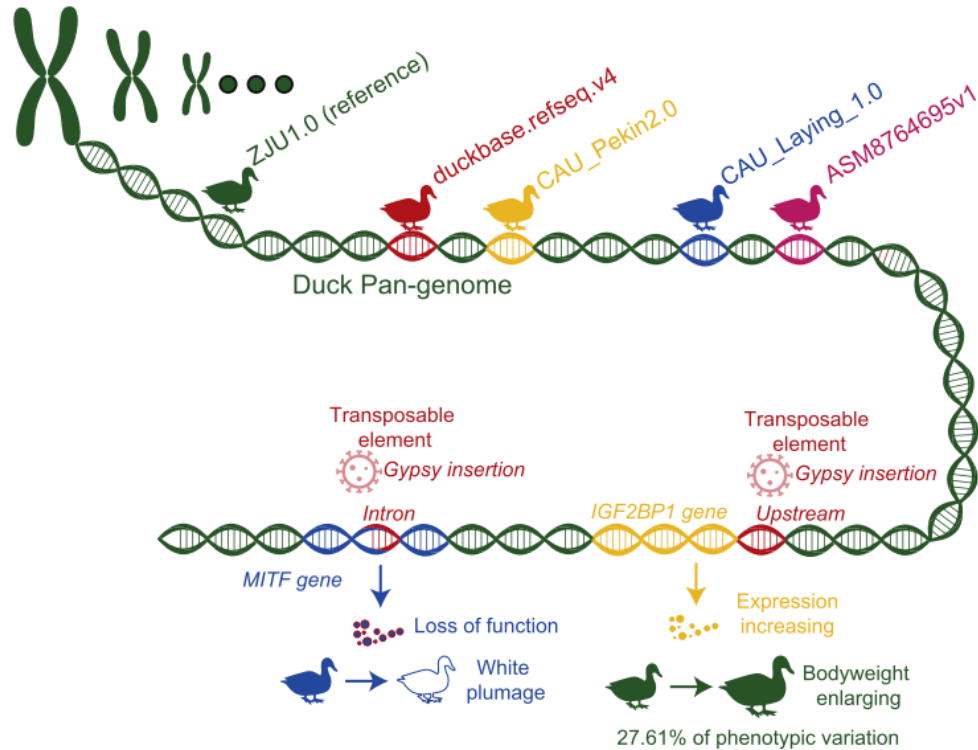
- **Three clades**
- **Ancient Gypsy copies**

IGF2BP1 ~3.88 million years ago

MITF ~0.6 million years ago



Summary



- We present the first duck pan-genome capturing ~40.98 Mb new sequences.
- A substantial number of these SVs were associated with traits related to domestication and improvement.
- We take the transposon insertions in the promoter of IGF2BP1 and the intron region of MITF as examples to analyze the phenotypic effect of transposons. We decipher the regulatory mechanisms of IGF2BP1 in bodyweight and MITF in white plumage.
- The Gypsy insertion located at the promoter of IGF2BP1, to our knowledge, explains the largest effect on bodyweight among avian species (27.61% of phenotypic variation).

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