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Retrocopies Expression in Normal and Tumoral Tissue



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DE ENSINO E PESQUISA

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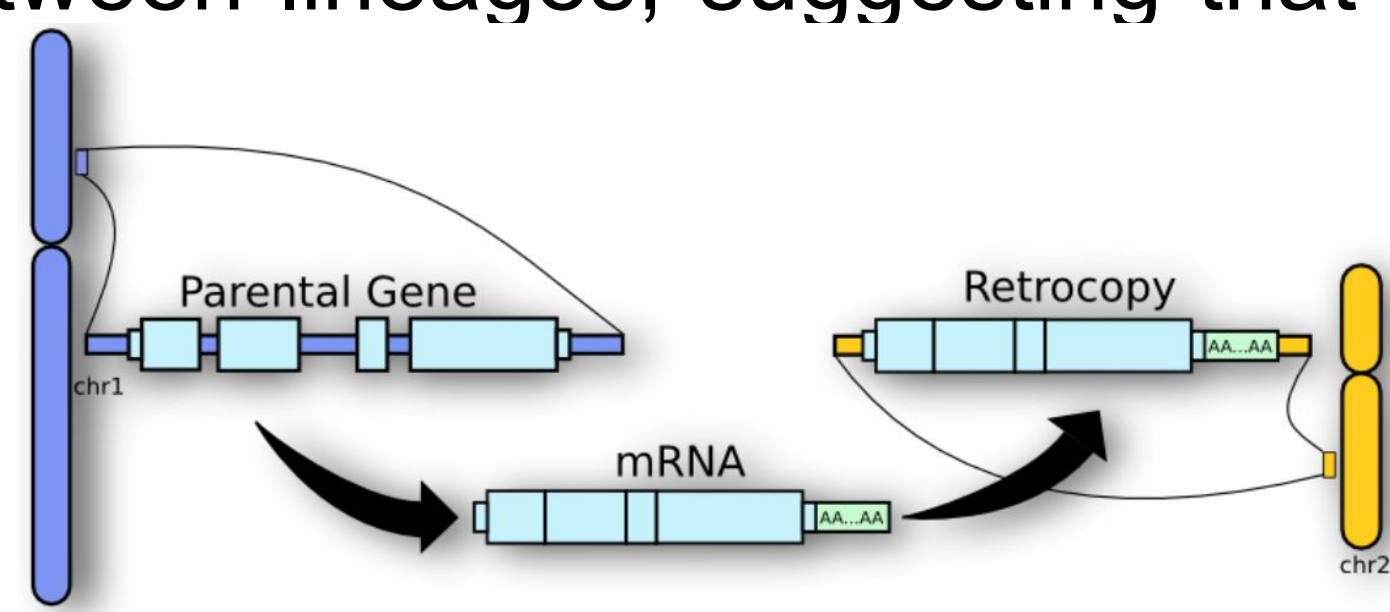
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INTRODUCTION

Retrocopies are copies of mRNAs reverse transcribed into the genome, characterized by a lack of introns and containing only exons of the parental gene. As a consequence of the lack of promoter regions for the expression, the first works describing the retrocopies classified them as "dead on arrival". An unexpected number of functional retrocopies have been identified thanks to the increasing practice of complete genome sequencing. Usually, new functional retrocopies (retrogenes) are initially expressed in the testicles, but their increased complexity allows them to display functions in other organs. The mechanism of retrotransposition of messenger RNA is active during the evolution of the primates and introduces a great genetic variety between lineages, suggesting that the retrocopies create a reservoir of new potentially functional genomic loci and, therefore, can assume important activities from the evolutionary point of view.



METHODOLOGY

Although some studies have investigated the functional evolution of primate retrocopies, transcriptome data from retrocopies remain largely unexplored. To explore the possible implications of retrocopies (transcribed) in the genome of humans and other mammals, we used the information from RCPedia, a database developed by our research group. RCPedia contains a complete catalog of retrocopies present in humans and five other primates, their genomic context, inter-species conservation, and expression data in six different tissues. Our objective was to study retrocopies with expression in humans, in order to (i) analyze the expression level of each retrocopy; (ii) to study retrocopies with species-specific expression and those shared among all primates; (iii) to evaluate the functional potential of each retrocopy based on its conservation with respect to the parental gene and the potential coding of retrocopies.

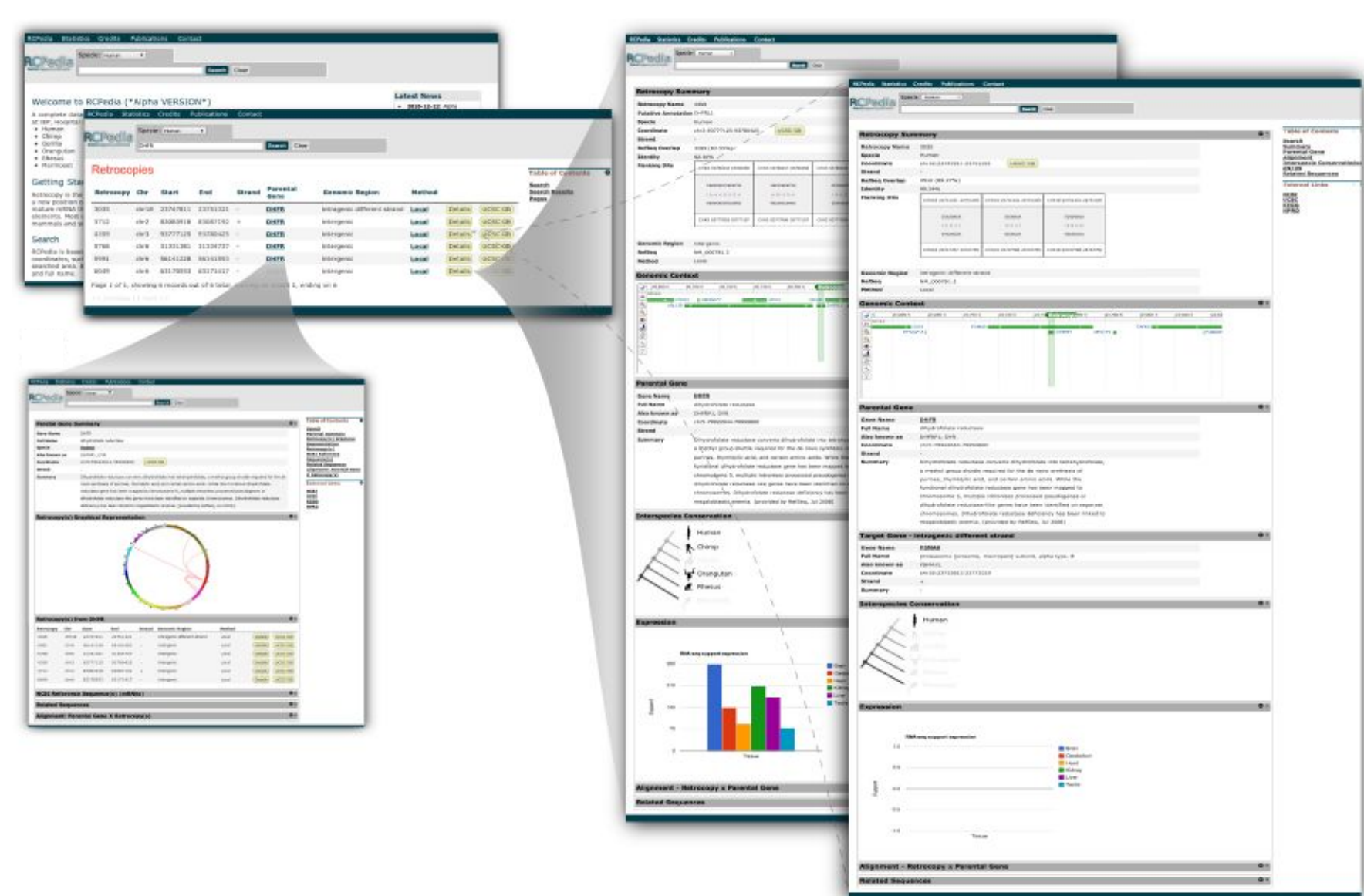


Figure 1. RCPedia summary. A schematic representation of RCPedia results for a sample query using the human DHFR gene. For the full dataset, please access <https://www.bioinfo.mochsl.org.br/rcpedia/>

RESULTS

This work revealed important expression profiling of retrocopies in primates, such as the predominance of tissue-specific expression (especially in testis), acquisition of new regulatory sequences, and divergence in terms of sequence and expression of retrocopies relative to their respective genes.

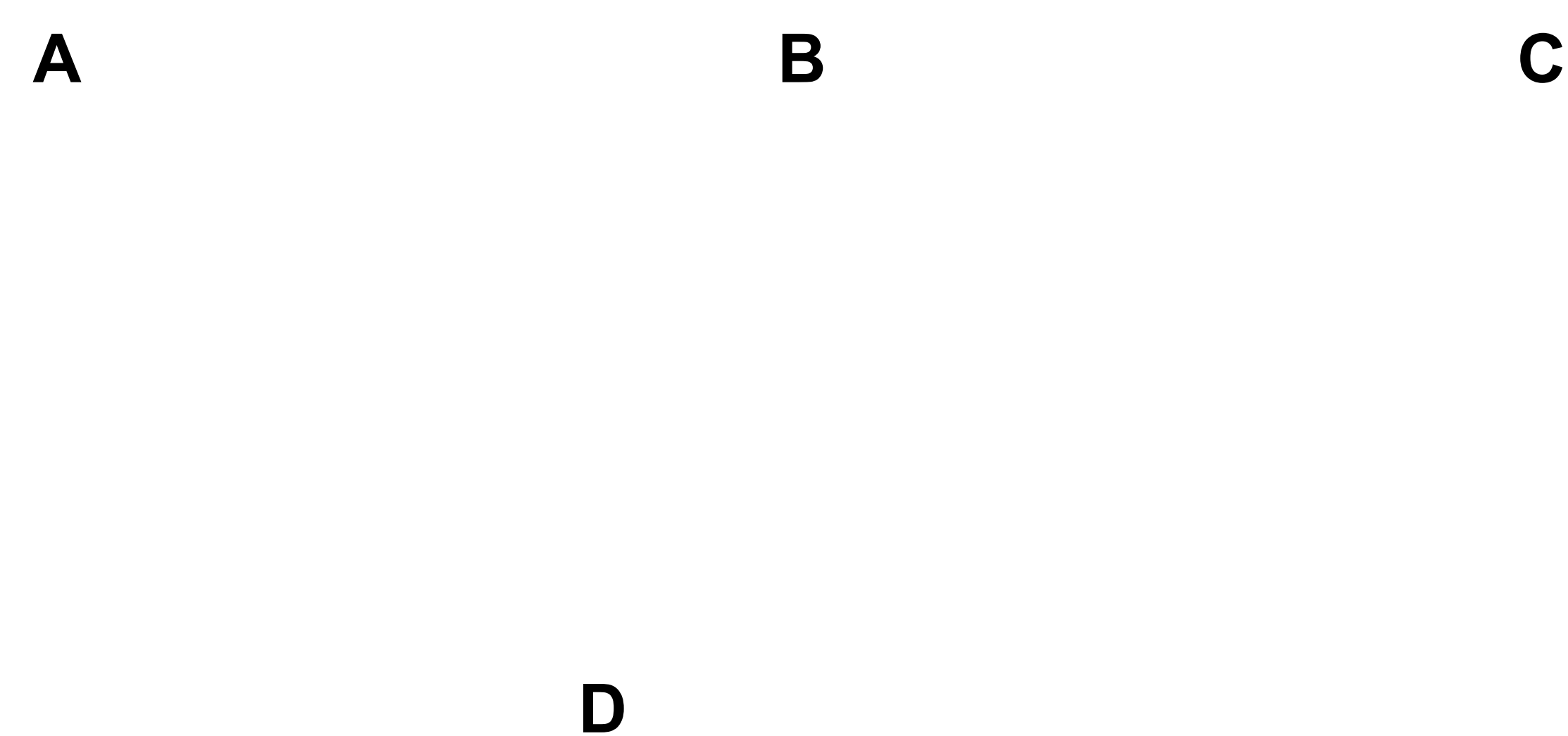


Figure 2. Summary of Retrocopies' Features A) Number of retrocopies and parental genes for each specie. B) Proportion of expressed retrocopies and parental genes that generate expressed retrocopies. C) Size distribution of retrocopies and gene transcripts for each specie. D) Retrocopies location in the genome for each specie.



Figure 3. Retrocopies' expression pattern in all species. A) Number of retrocopies testicle specific per species. B) Number of retrocopies brain and cerebellum specific per species. C) Number of retrocopies that are expressed only in 1, 2, 3, 4, 5 or 6 tissues.



Figure 4. Retrocopies' expression pattern in Humans. A) Expression distribution of retrocopies that are expressed in 1, 2, 3, 4, 5 or 6 tissues. B) Same as A but without the expression in the testicles. C) Correlation of ORF identity and expression.

CONCLUSION

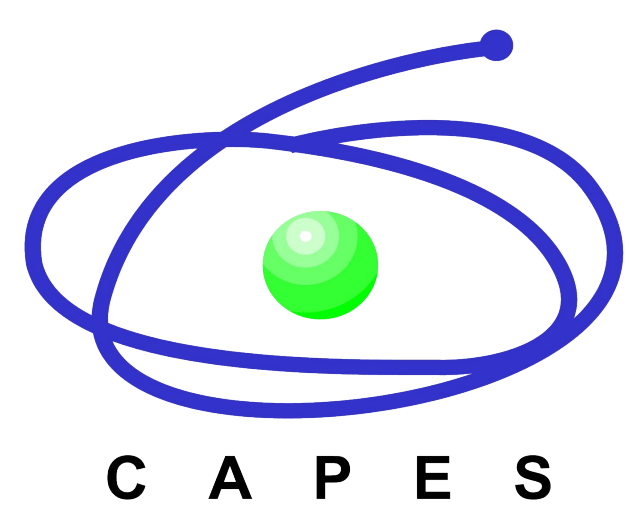
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