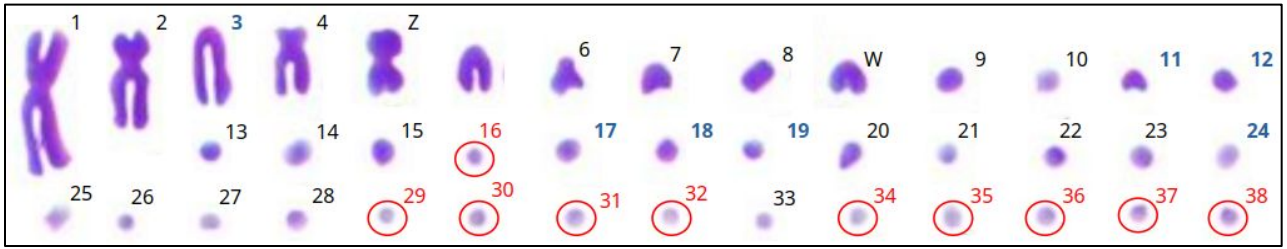
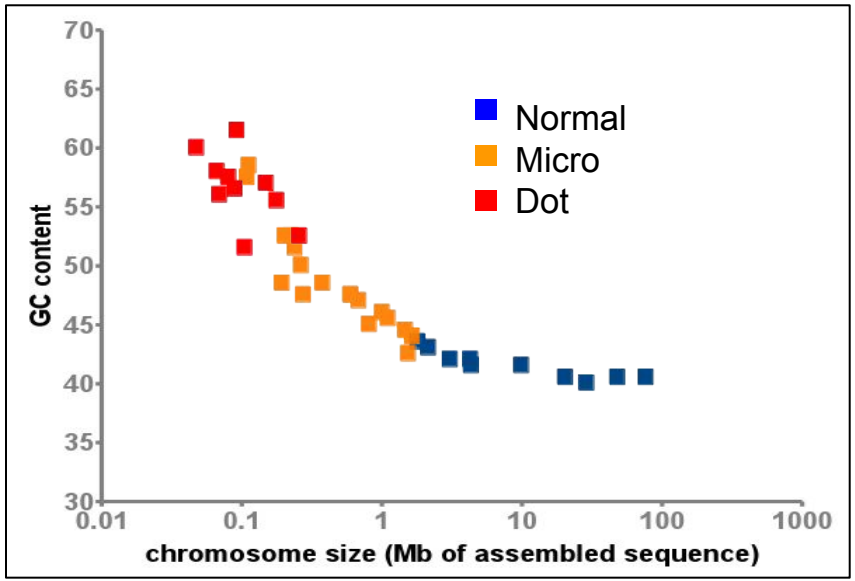
Crazy avian genome, stuttering genes, and why we love nanopore

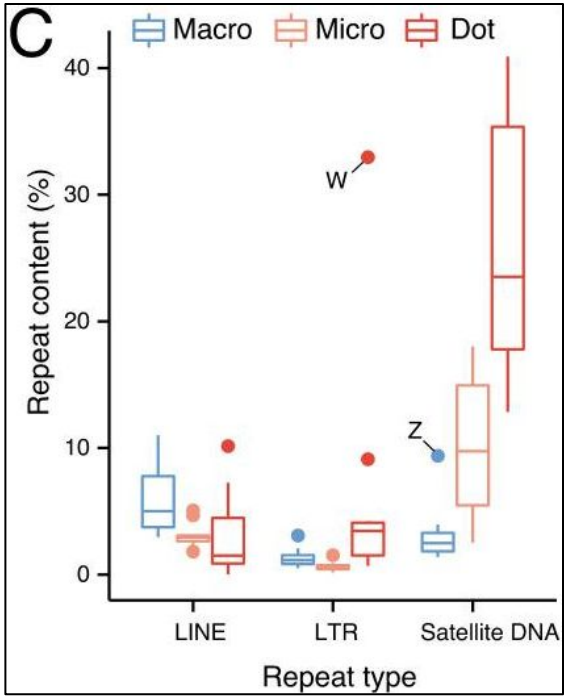
Tomáš Hron

ENBIK 2025

Crazy avian genome







Proc Natl Acad Sci U S A. 2023 Feb 13;120(8):e2216641120. doi: 10.1073/pnas.2216641120

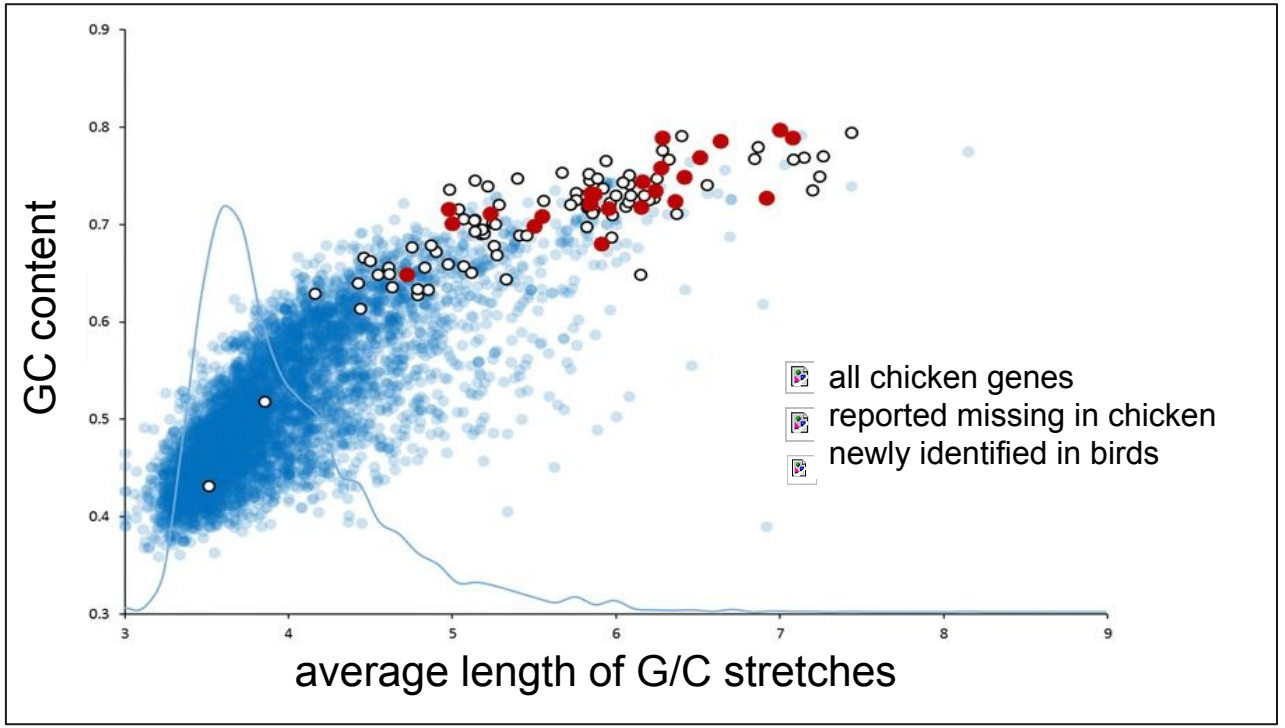
Avian hidden genes

Erythropoietin
glycoprotein hormone that controls erythropoiesis

Leptin
hormone that regulates energy balance by inhibiting sense of hunger

Nephrin
protein necessary for the functioning of the renal filtration barrier

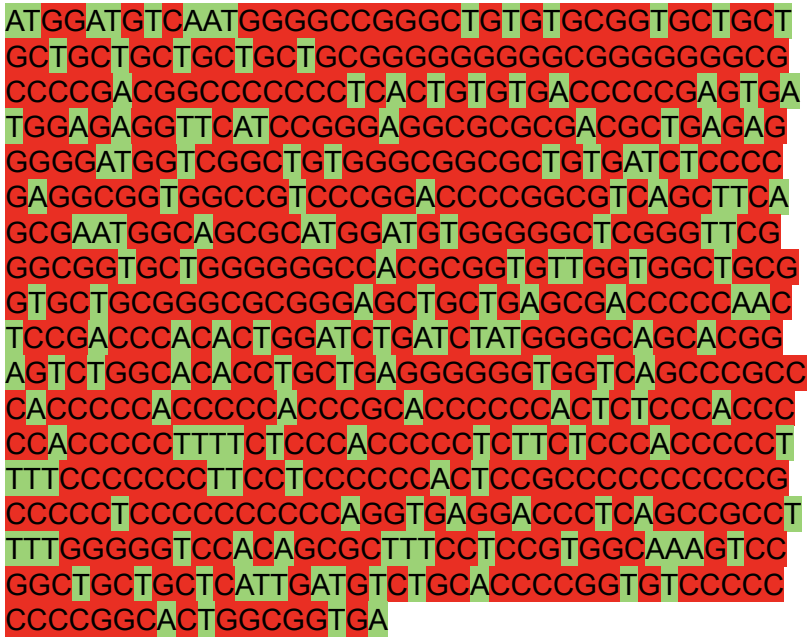
TNF-alpha
cytokine involved in systemic inflammation...



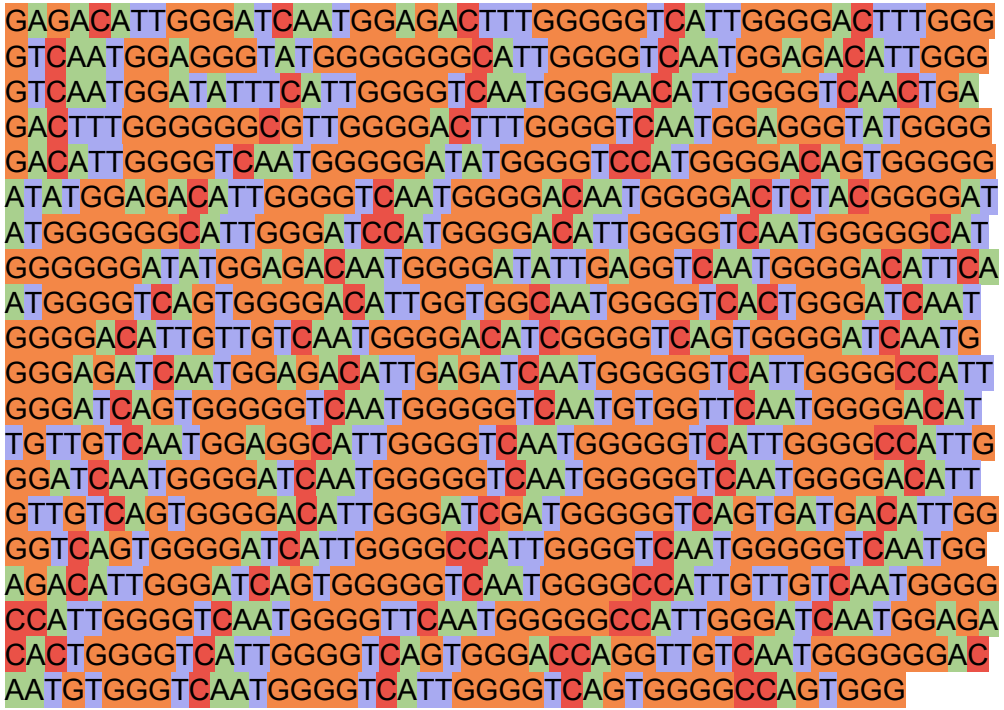
Hron et al. Genome Biology (2015). Hidden genes in birds

Avian hidden genes

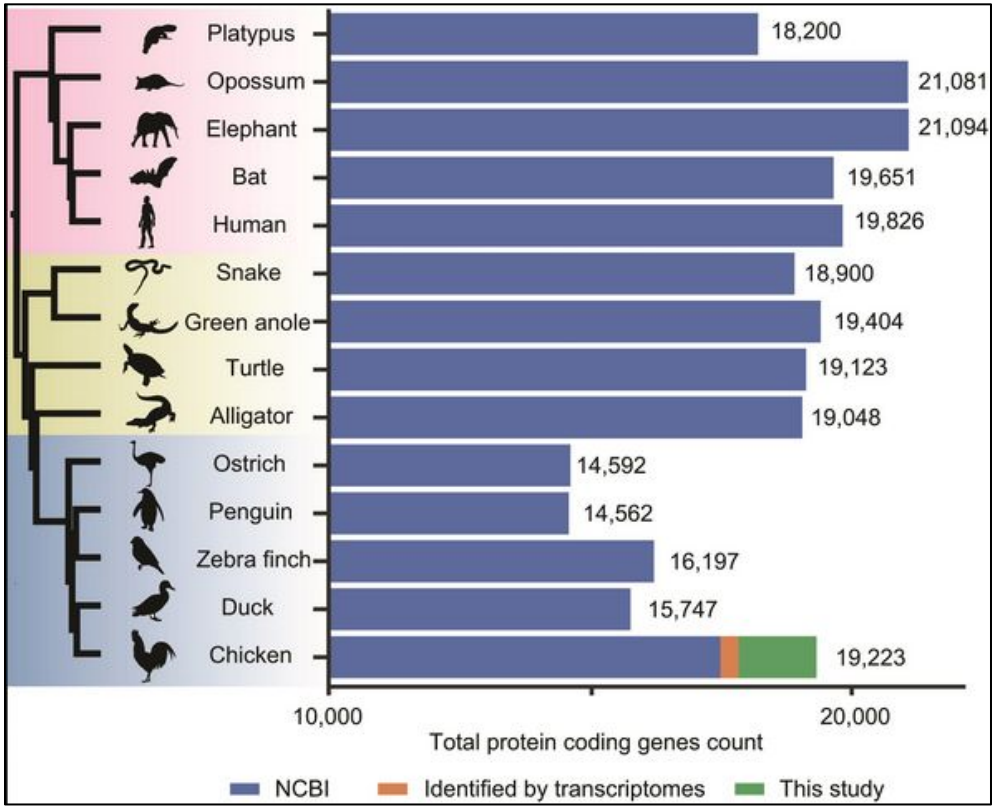
Chicken EPO - CDS (72% GC)

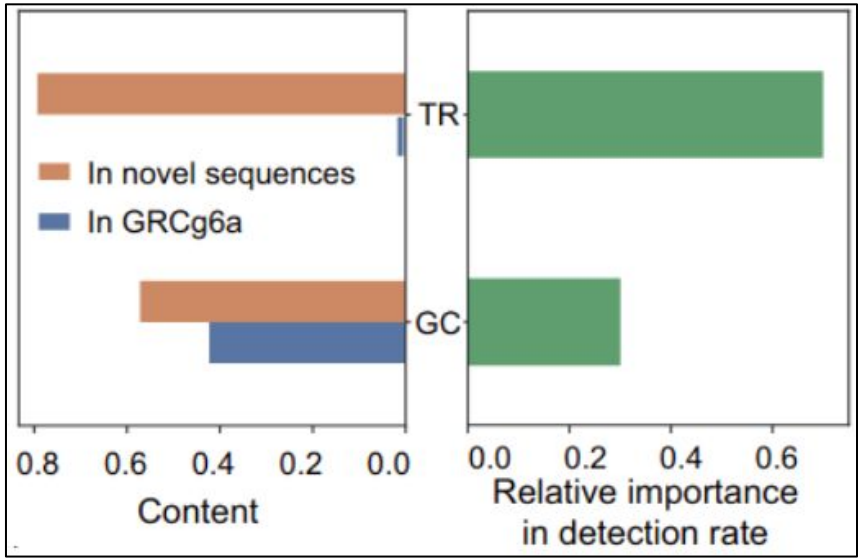


Chicken EPO - part of intron 1



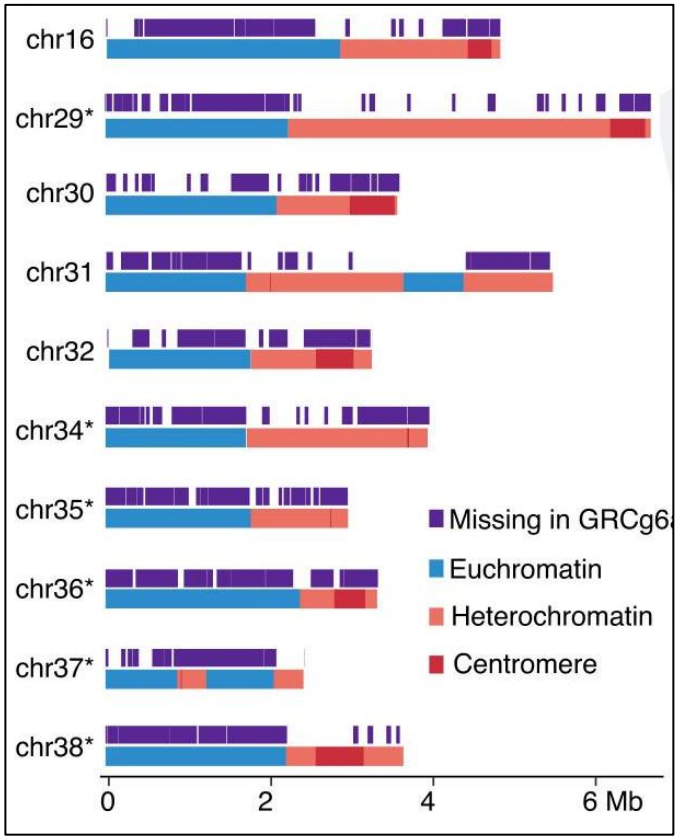
Thousands of hidden genes identified on microchromosomes

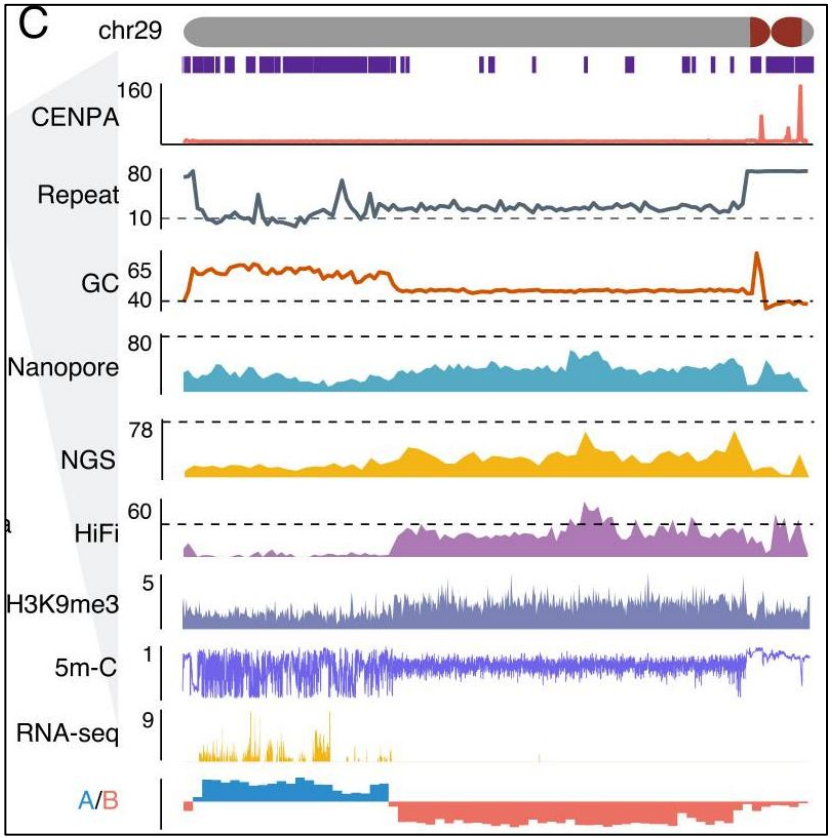




Molecular Biology and Evolution, Volume 39, Issue 4, April 2022, msac066, https://doi.org/10.1093/molbev/msac066

God bless Nanopore - new complete chicken genome





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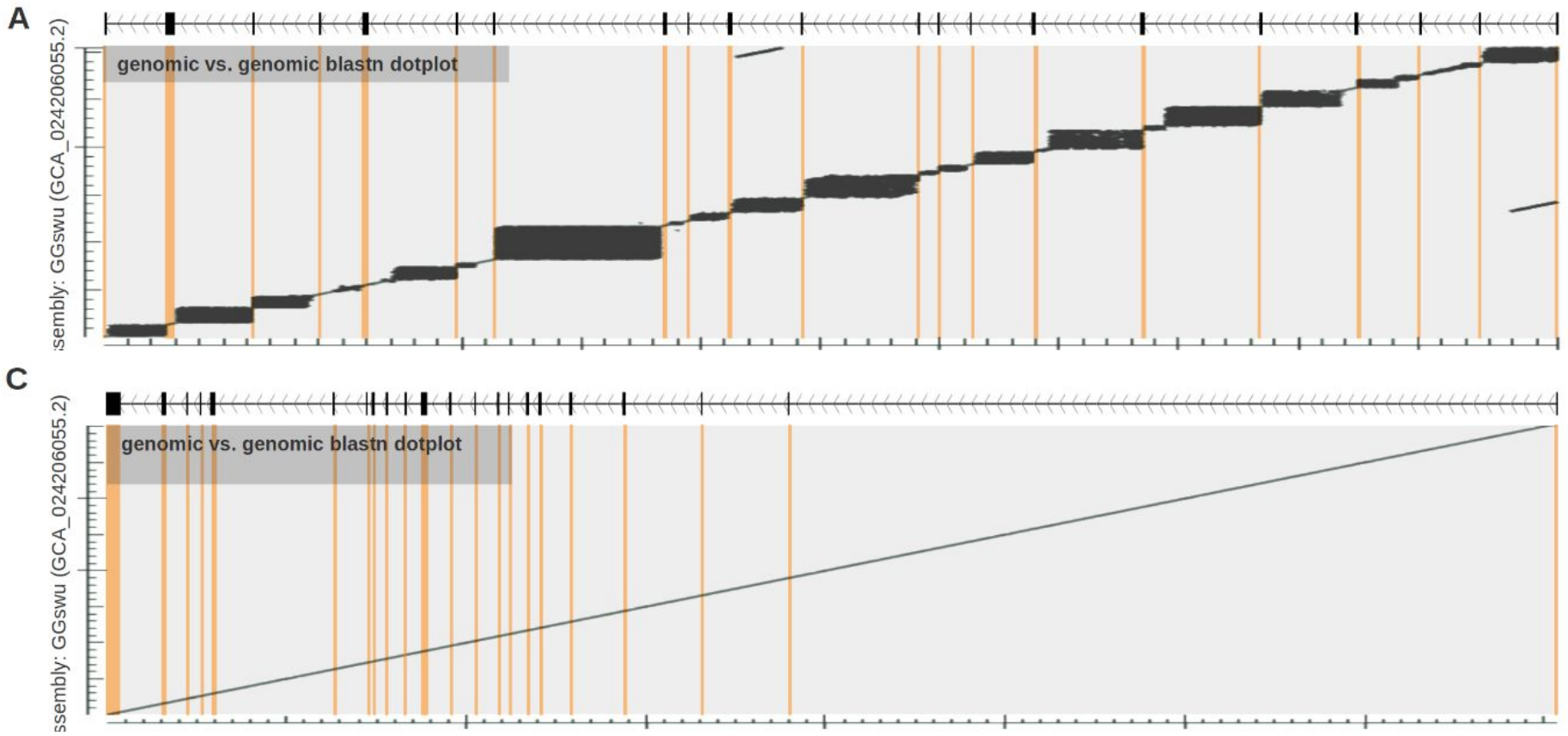
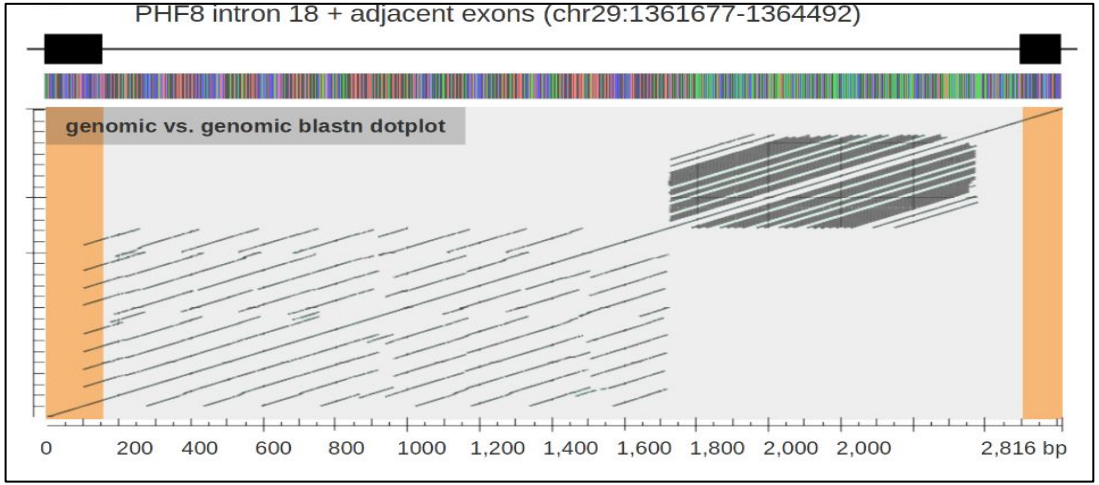
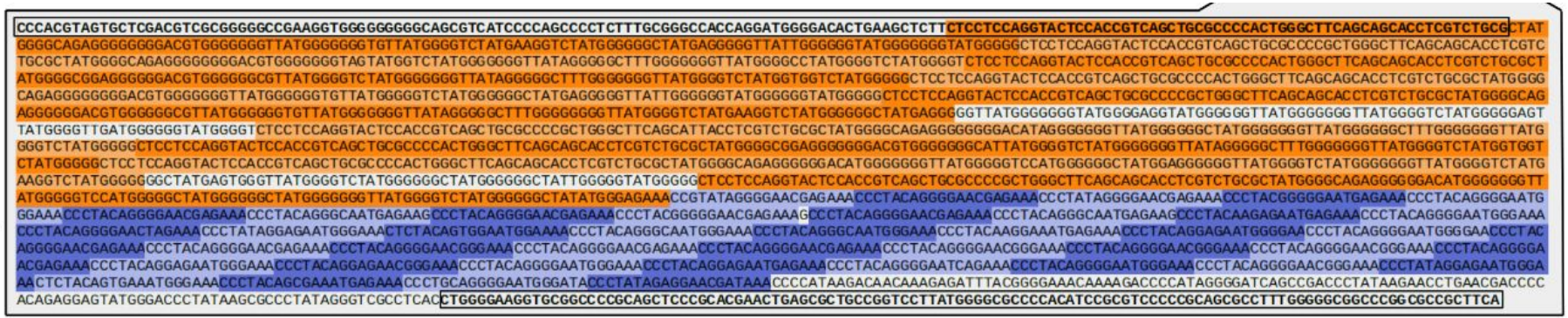
Local expansion of repeats on dot chromosomes

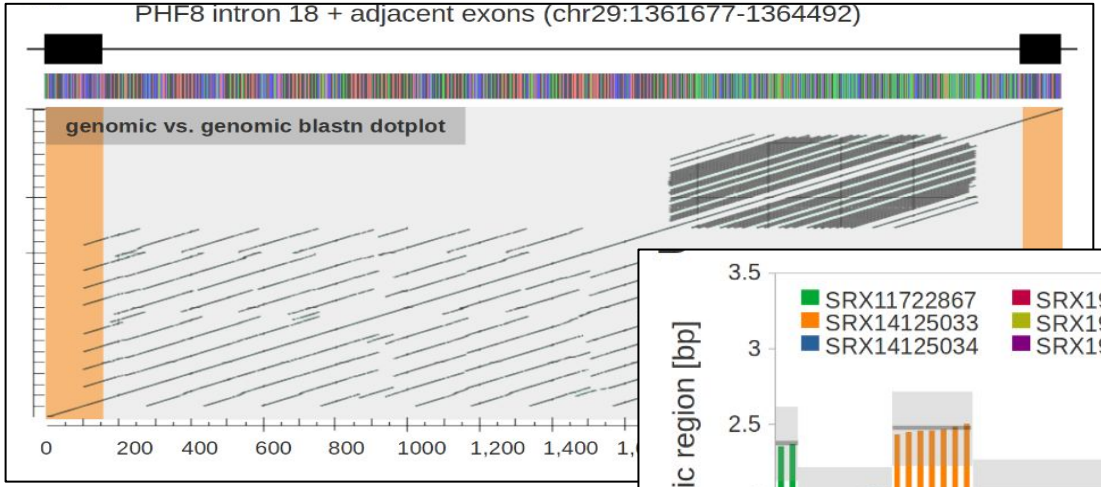


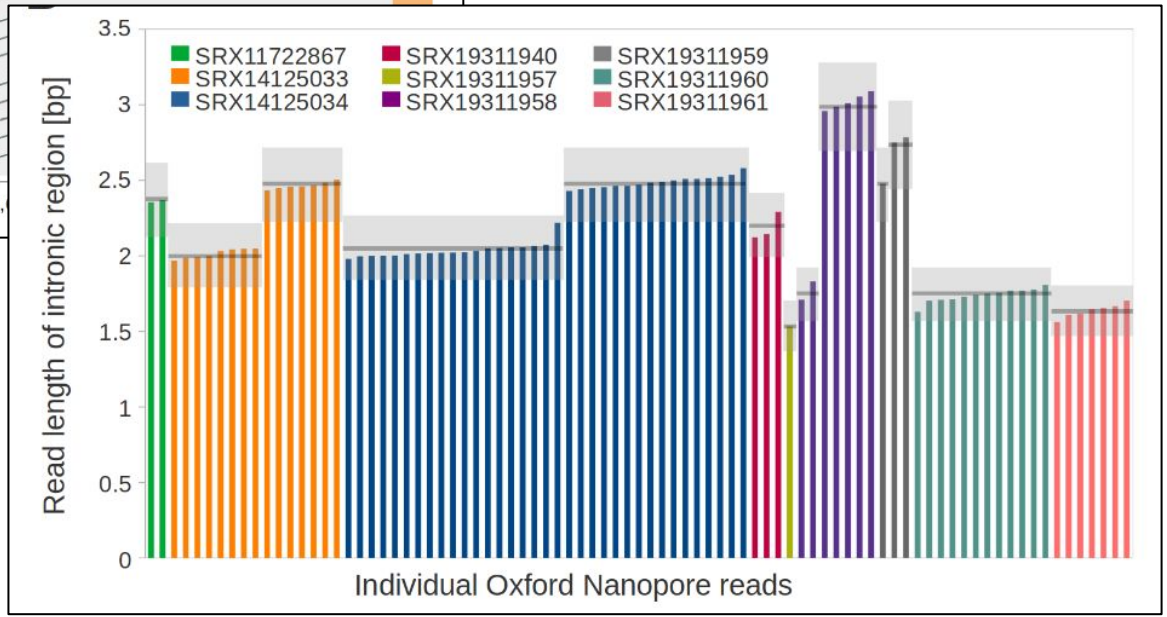
Local expansion of repeats on dots



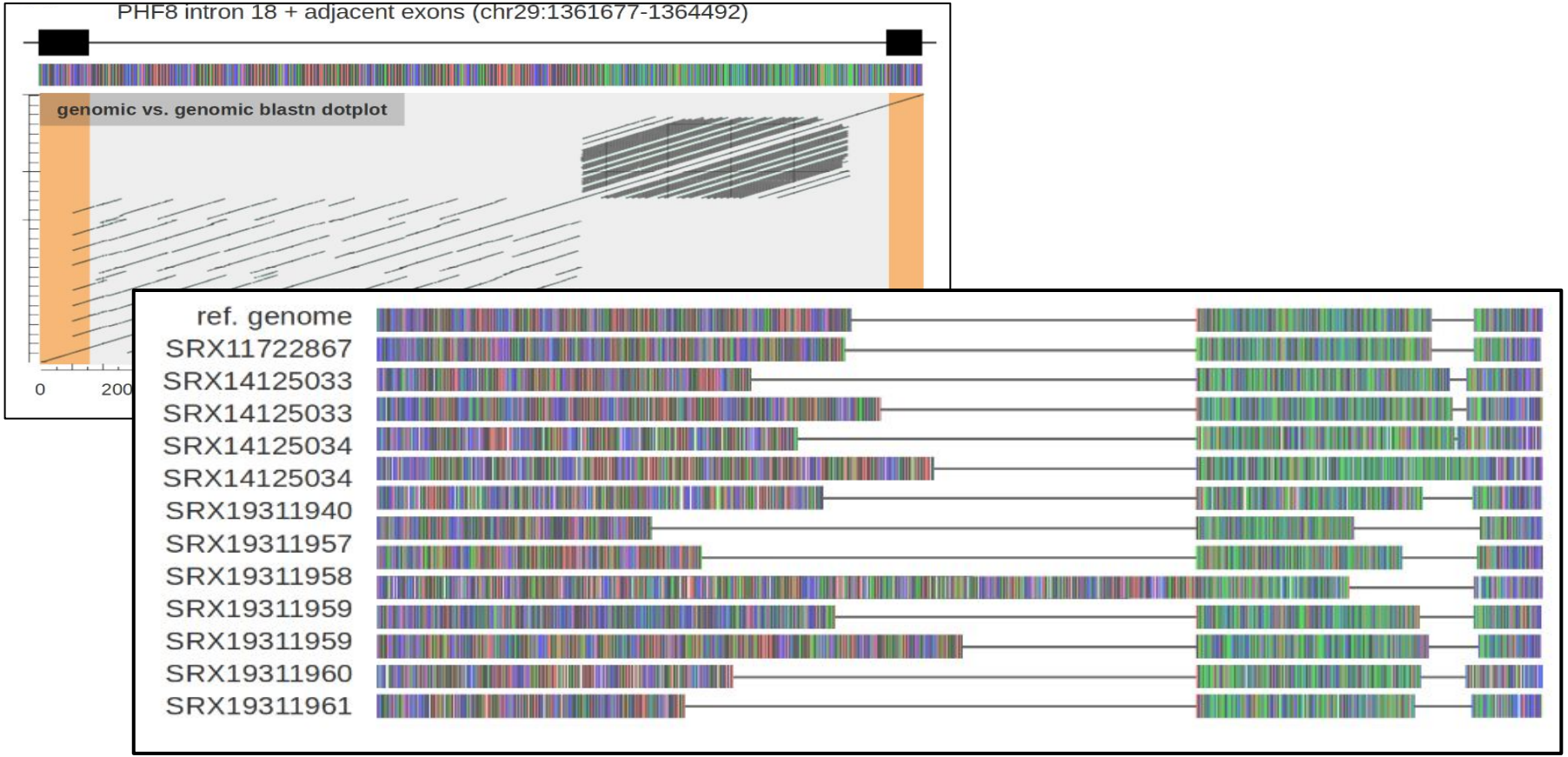


Local expansion of repeats on dots - length variability



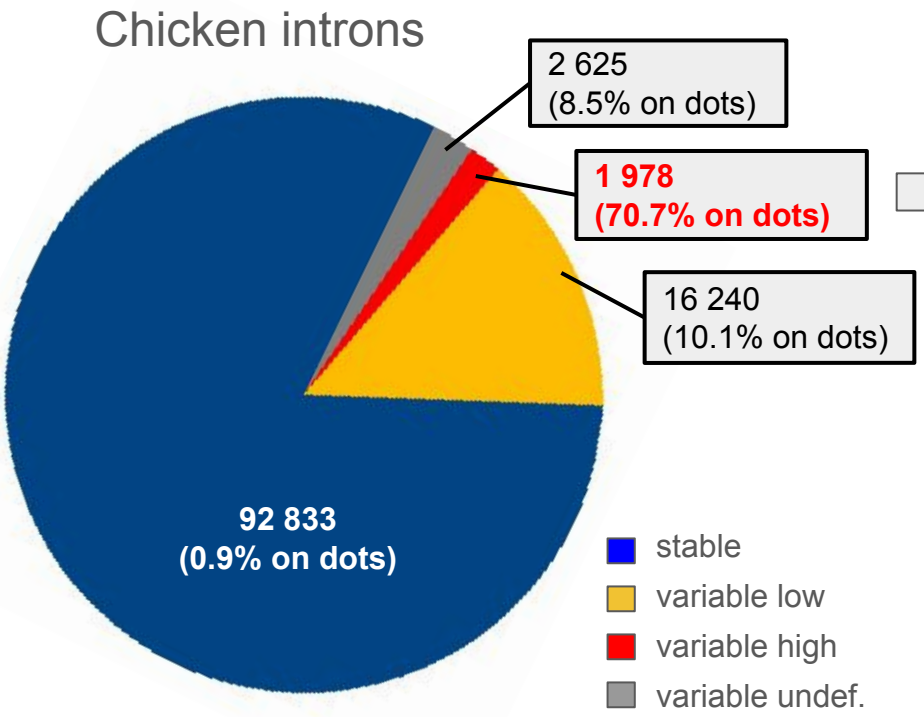


Local expansion of repeats on dots - length variability



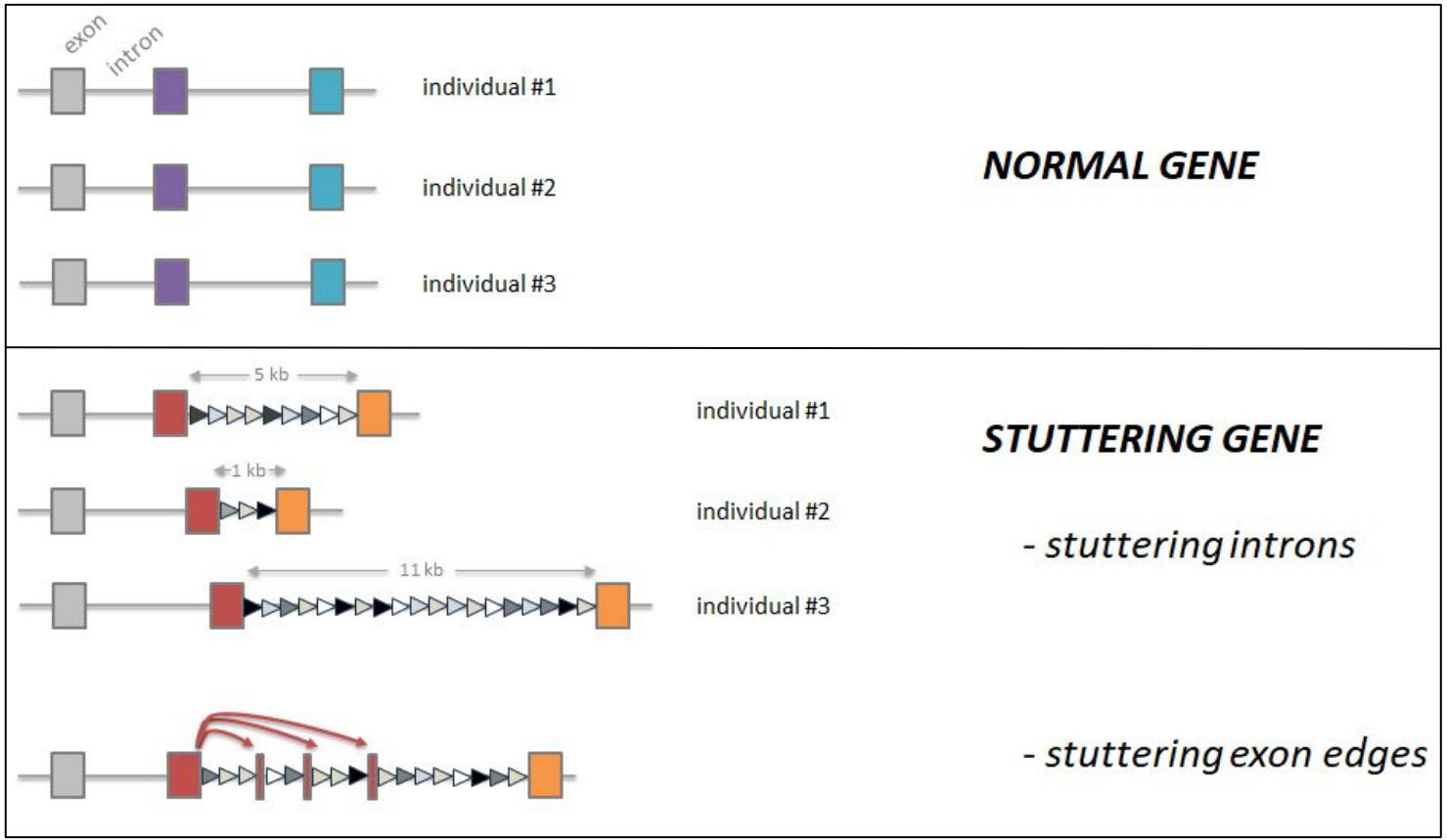
Unpublished

Local expansion of repeats on dots - Recent changes in chickens

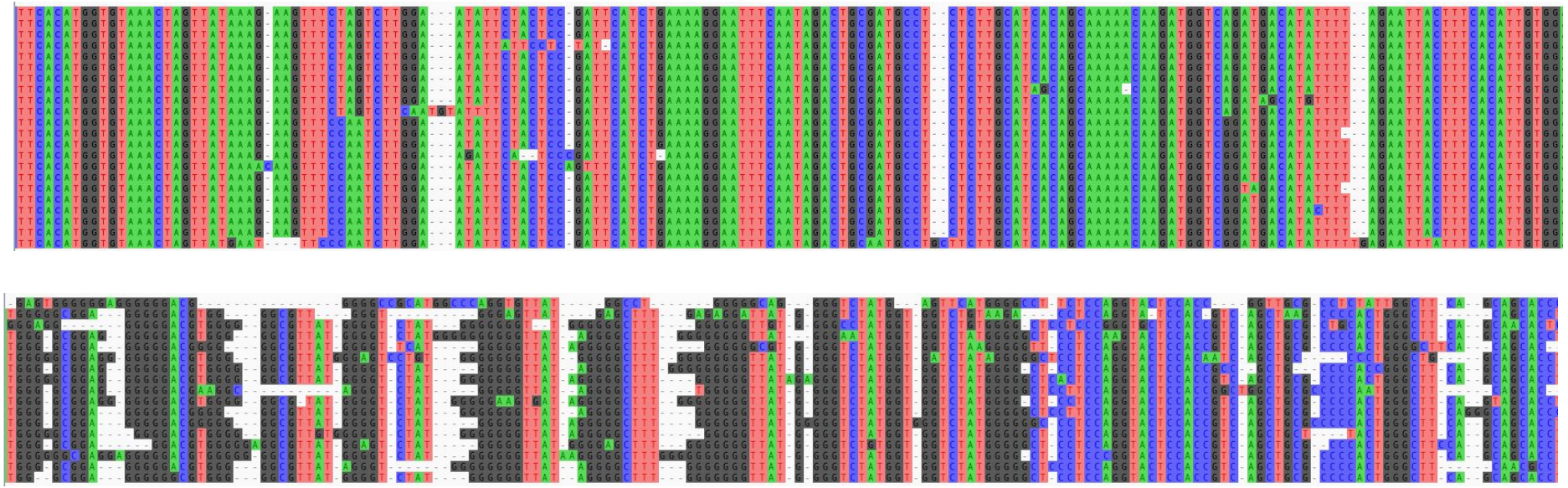


- highly variable in chicken population
- massive occurrence of repeat clusters
- almost exclusively on dot chrome
- present also in other birds
- nothing like this in other vertebrates

Stuttering genes



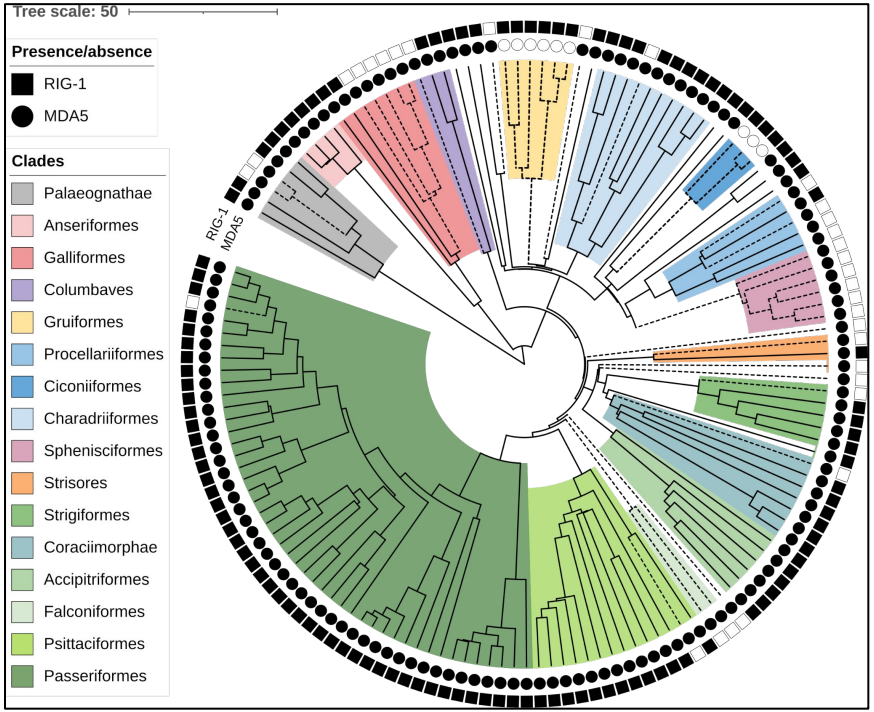
Even Nanopore data cannot go to single-base resolution



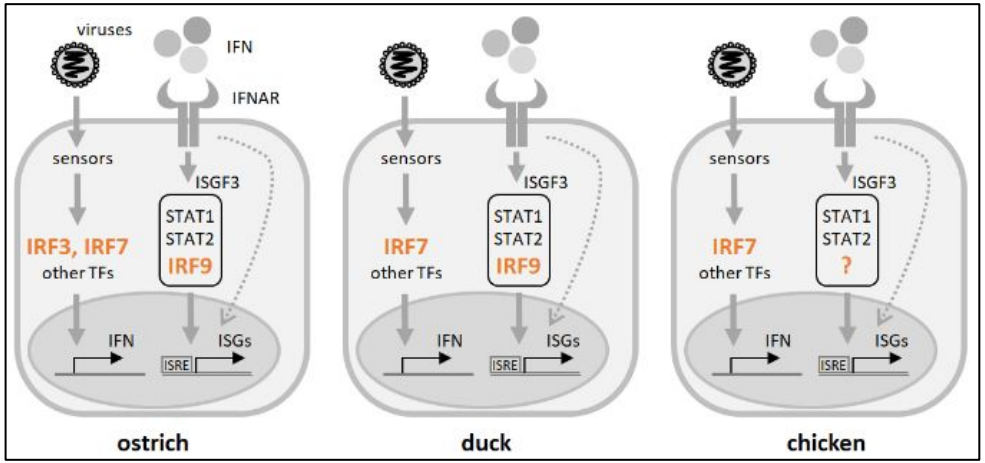
Unpublished

Hidden and missing genes in birds

RNA virus sensors missing in birds

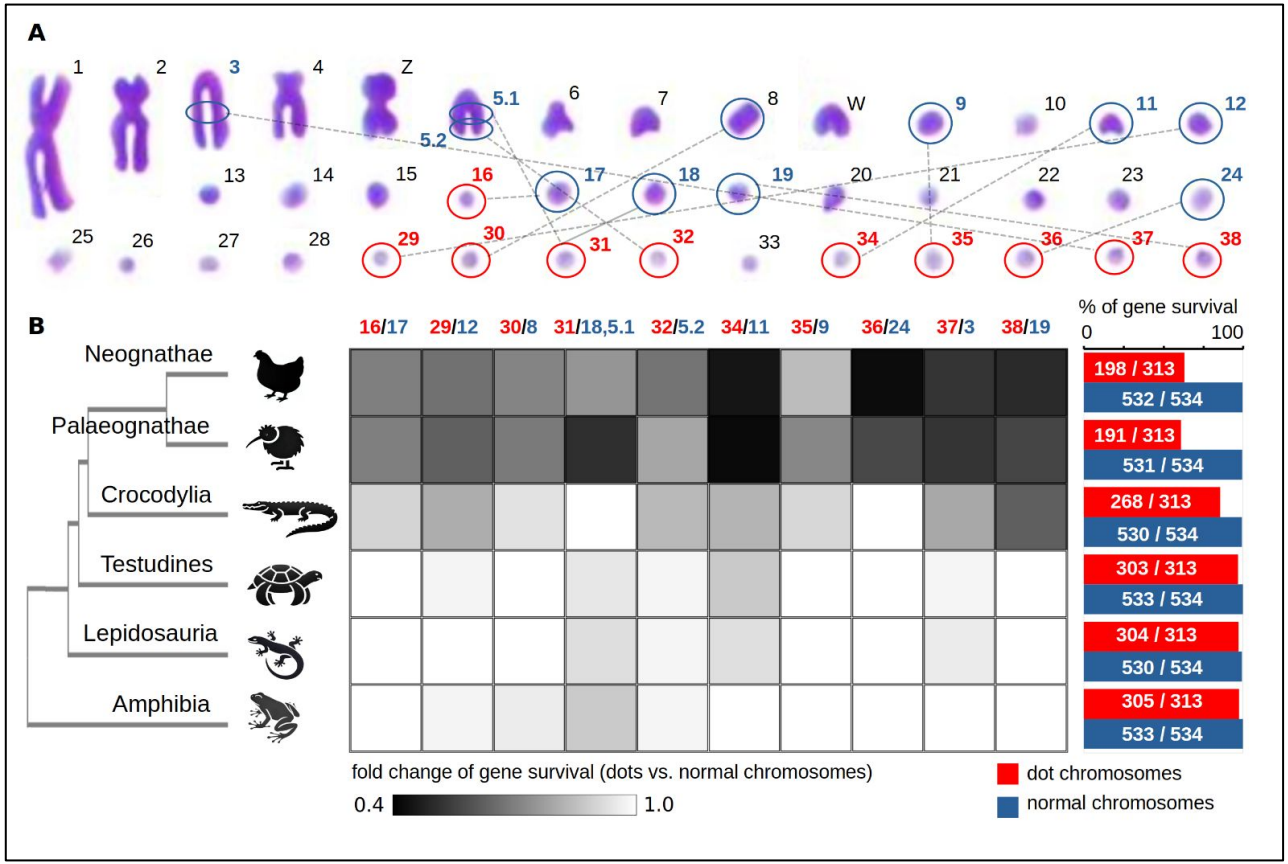


Interferon regulatory factors missing in birds



Viruses. 2022 Dec 20;15(1):3. doi: 10.3390/v15010003. ,Preprint 2024 Sep. doi: https://doi.org/10.1101/2024.09.24.613690

Missing genes on avian dots



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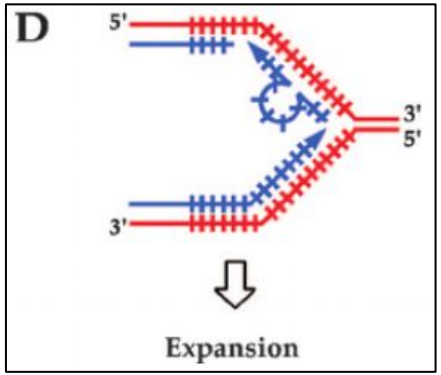
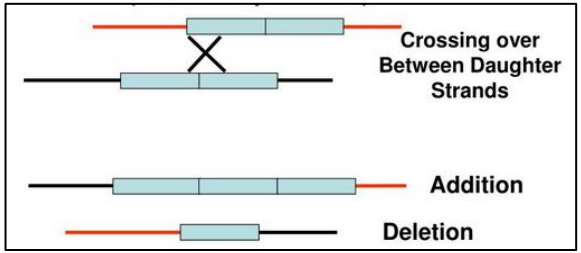
Potential mechanism of sequence stuttering on avian dots

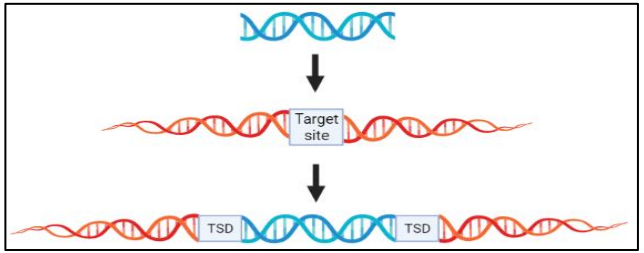
Dna replication slippage



Unequal crossing over



Mobile element insertion



Where
On dots. Where exactly? What genomic property needed?

When
in meiosis/mitosis? (sequencing cells of different passages)

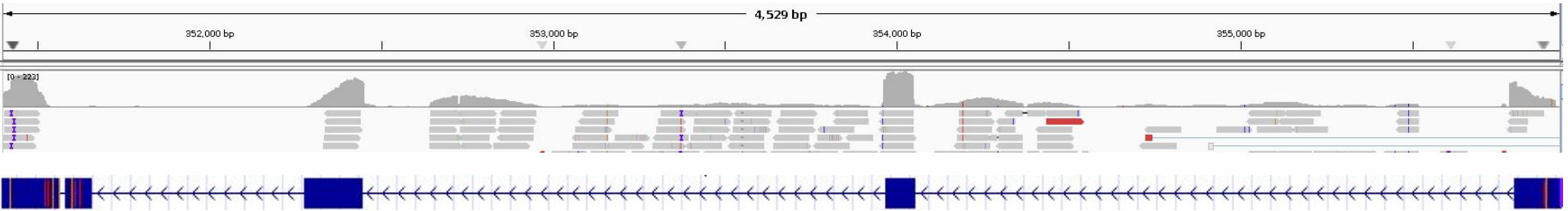
How
recombination? (local copy of ~10-1000 bp, no defined template)

Why
G4 formation? Dot chromosome decay?

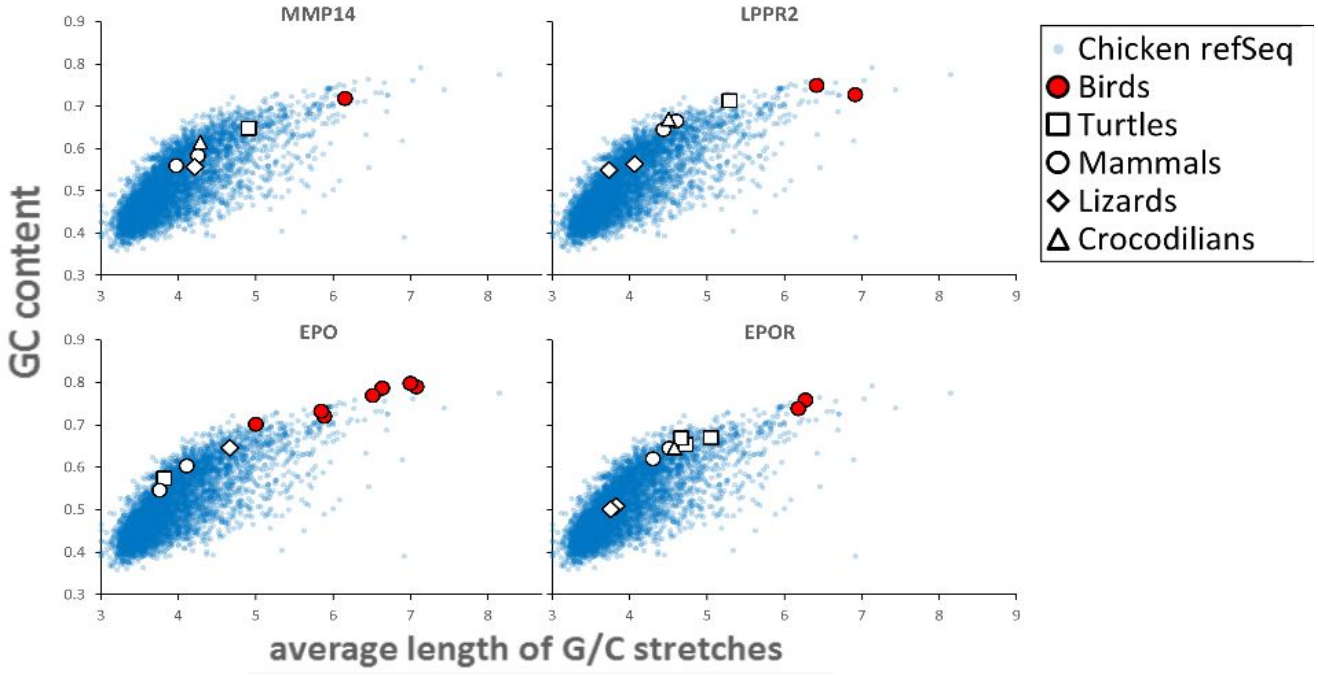
Thank you

Chicken erythropoietin (EPO)

up to 1000x coverage from RNAseq data



Avian Hidden Genes



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