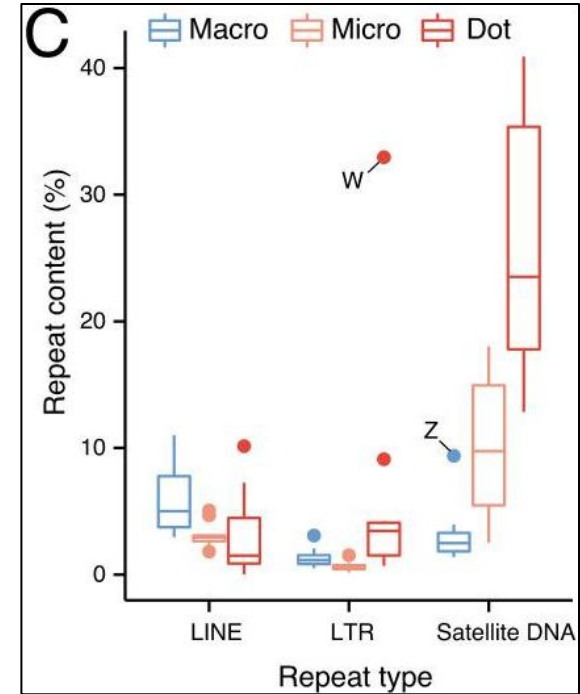
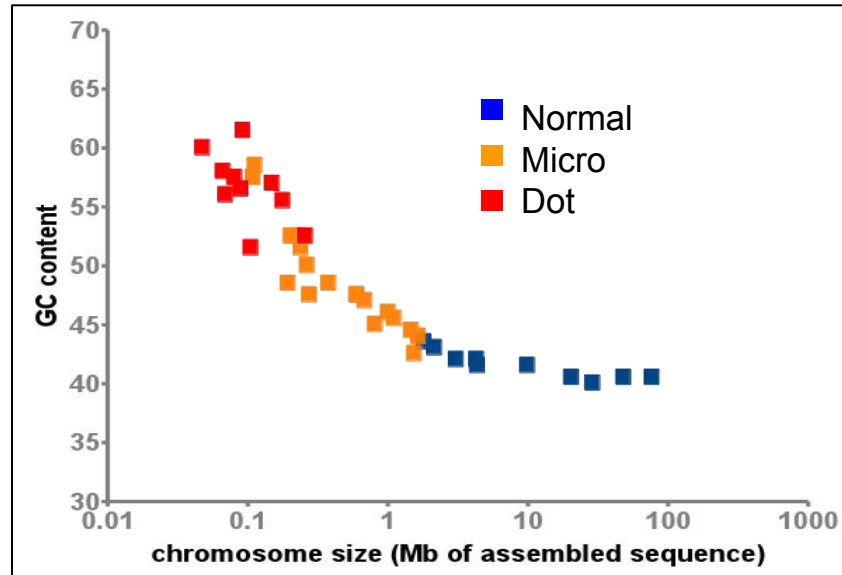
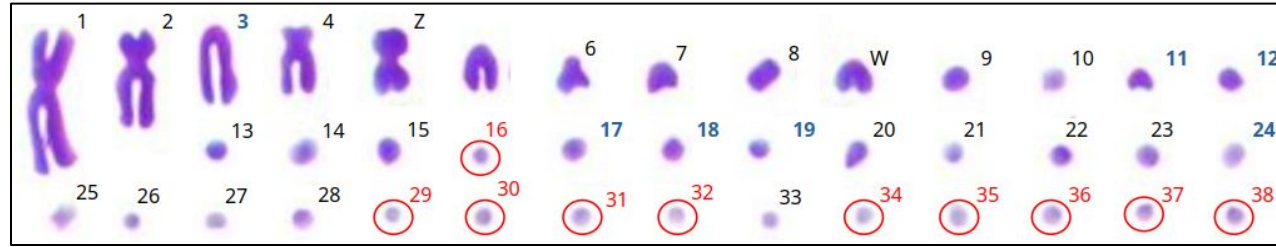


Crazy avian genome, stuttering genes, and why we love nanopore

Tomáš Hron

ENBIK 2025

Crazy avian genome



Avian hidden genes

Erythropoietin

glycoprotein hormone that controls erythropoiesis

Leptin

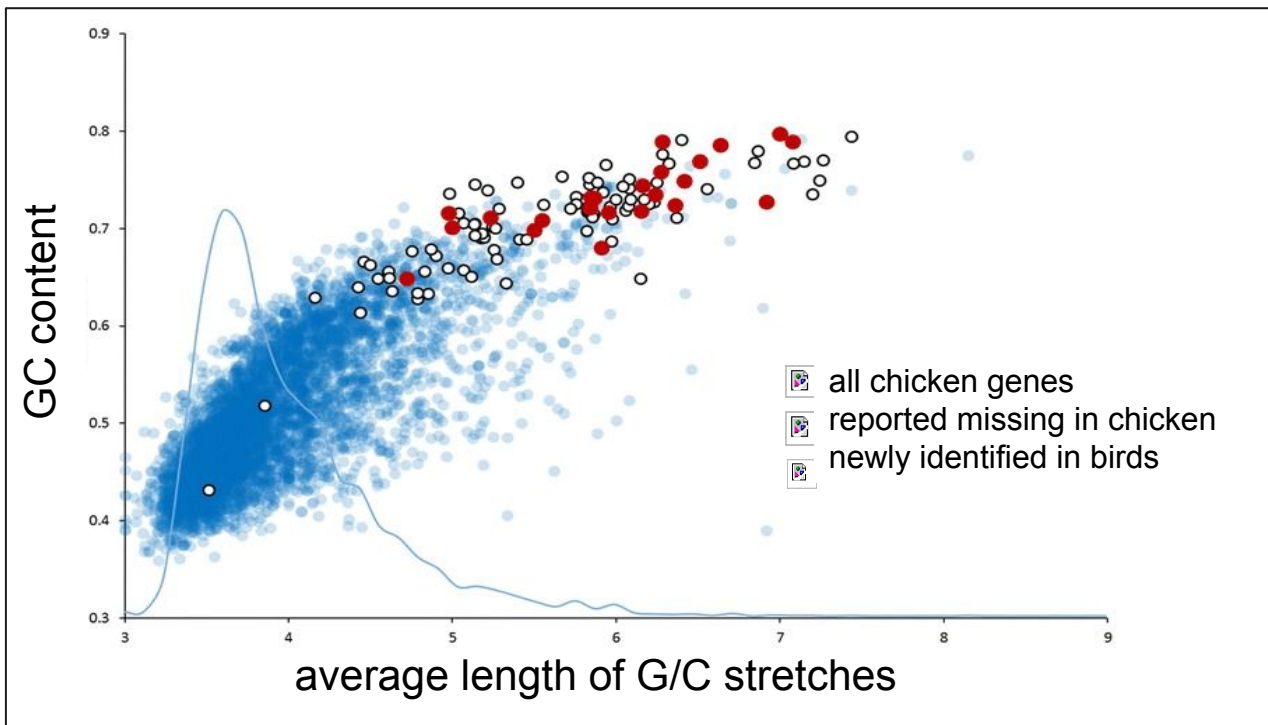
hormone that regulates energy balance by inhibiting sense of hunger

Nephrin

protein necessary for the functioning of the renal filtration barrier

TNF-alpha

cytokine involved in systemic inflammation...



Avian hidden genes

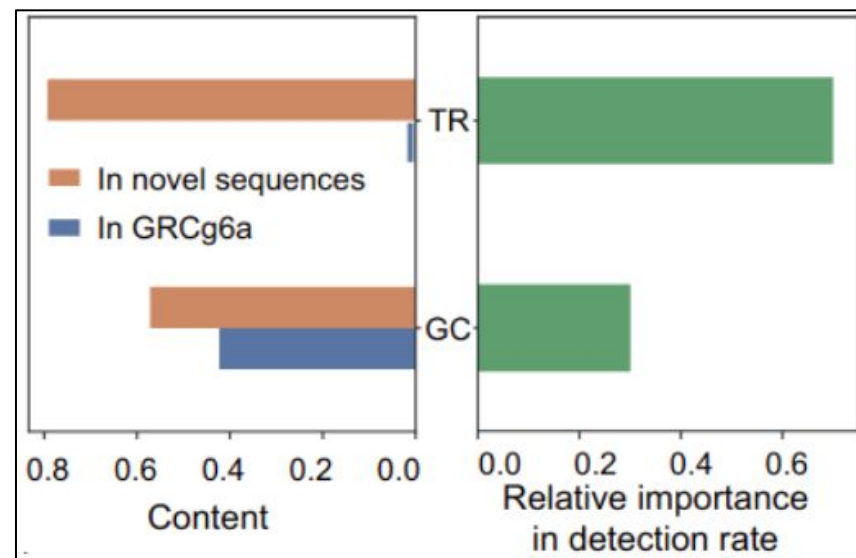
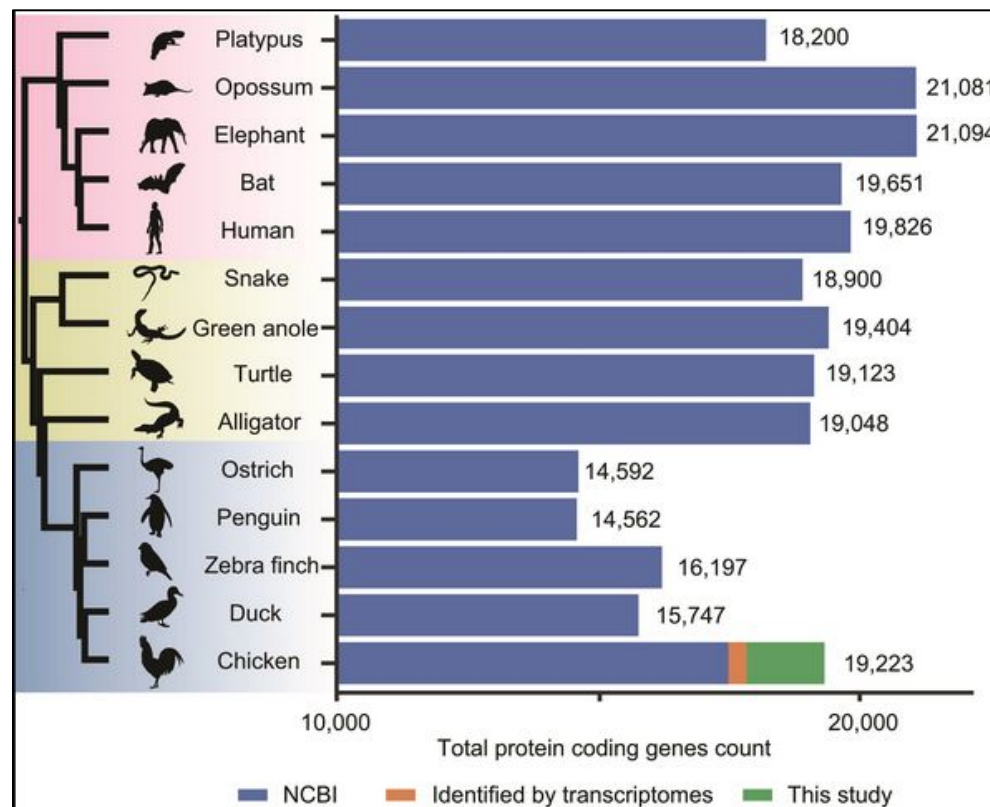
Chicken EPO - CDS (72% GC)

ATGGATGTC AATGGGGCCGGGCTGTGTGCGGTGCTGCT
GCTGCTGCTGCTGCTGCGGGGGGGGGCGGGGGGGGGCG
CCCCGACGGCCCCCCTCACTGTGTGACCCCGAGTGA
TGGAGAGGTT CATCCGGGAGGCGCGCGACGCTGAGAG
GGGGATGGTCGGCTGTGGGCGGCGCTGTGATCTCCCC
GAGGCGGTGGCCGTCCCGGACCCCGGCGTCAAGCTTCA
GCGAATGGCAGCGCATGGATGTGGGGGCTCGGGTTCTG
GGCGGTGCTGGGGGGGCCACGCGGTGTTGGTGGCTGCG
GTGCTGCGGGCGCGGGAGCTGCTGAGCGACCCCAAC
TCCGACCCACACTGGATCTGATCTATGGGGCAGCACGG
AGTCTGGCACACCTGCTGAGGGGGGTGGTCAGCCCGCC
CACCCCAACCCCAACCCGCACCCCACTCTCCACCC
CCACCCCTTTTCTCCACCCCTCTTCTCCACCCCT
TTTCCCCCCTTCTTCCCCCACTCCGCCCCCCCCCG
CCCCCTCCCCCCCCCAAGTGAGGACCTCAGCCGCCT
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CCCCGGCACTGGCGGTGA

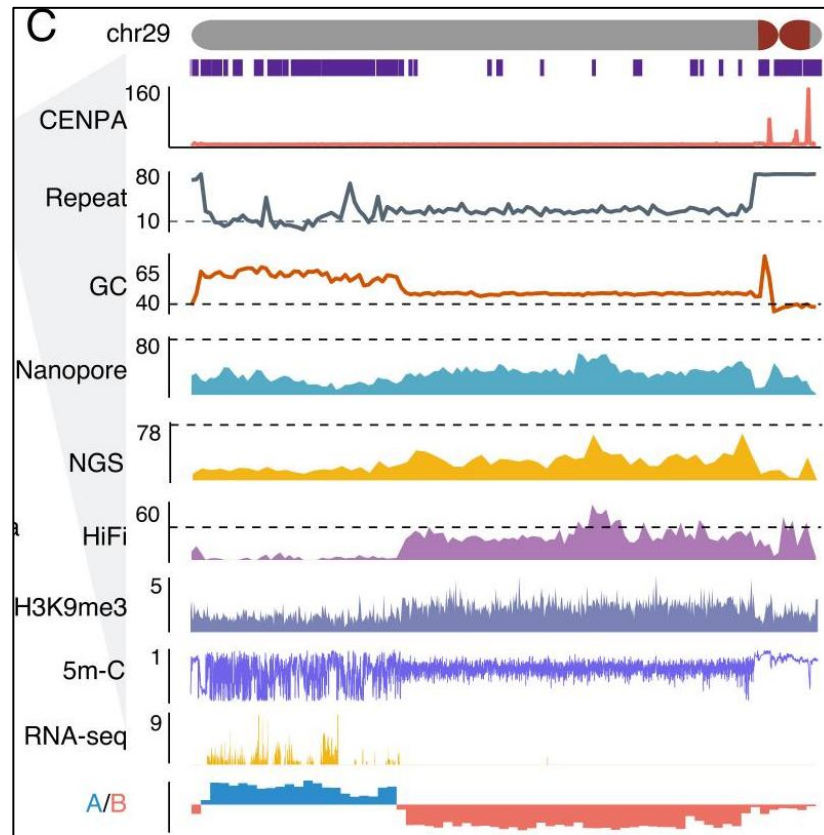
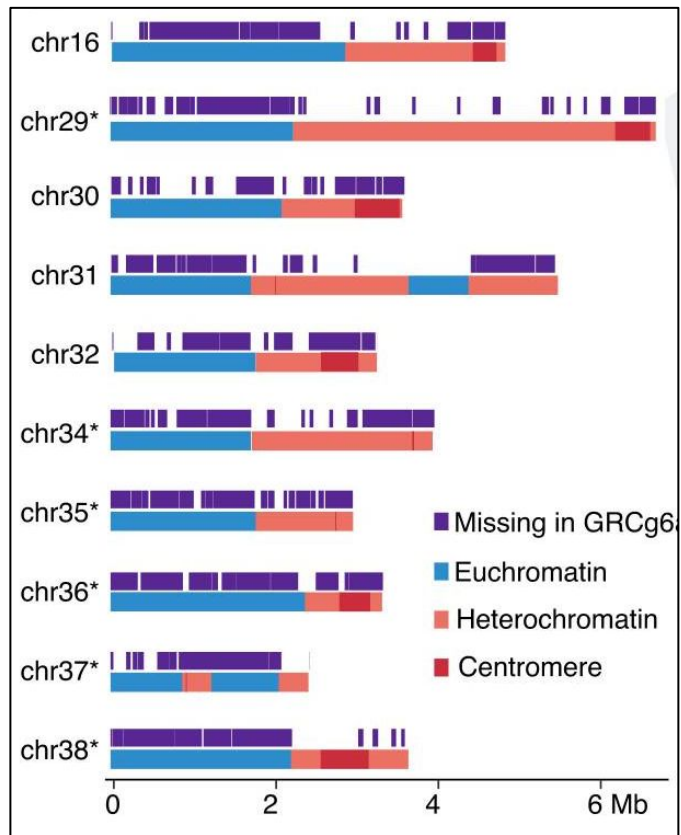
Chicken EPO - part of intron 1

GAGACATTGGGATCAATGGAGACTTTGGGGGTCAATTGGGGACTTTGGG
GTCAATGGAGGGTATGGGGGGGCATTGGGGTCAATGGAGACATTGGG
GTCAATGGATATTTCAATTGGGGTCAATGGGAACATTGGGGTCAACTGA
GACTTTGGGGGGCGTTGGGGACTTTGGGGTCAATGGAGGGTATGGGG
GACATTGGGGTCAATGGGGGATATGGGGTCCATGGGGACAGTGGGGG
ATATGGAGACATTGGGGTCAATGGGGACAATGGGGACTCTACGGGGAT
ATGGGGGGCATTGGGATCCATGGGGACATTGGGGTCAATGGGGGCAT
GGGGGGATATGGAGACAATGGGGATATTGAGGTCAATGGGGACATTCA
ATGGGGTCAGTGGGGACATTGGTGGCAATGGGGTCACTGGGATCAAT
GGGGACATTGTTGTCAATGGGGACATCGGGGTCACTGGGGATCAATG
GGGAGATCAATGGAGACATTGAGATCAATGGGGGTCAATTGGGGCCATT
GGGATCAGTGGGGGTCAATGGGGGTCAATGTGGTTCAATGGGGACAT
TGTTGTCAATGGAGGCATTGGGGTCAATGGGGGTCAATTGGGGCCATTG
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GTTGTCACTGGGGACATTGGGATCGATGGGGGTCACTGATGACATTGG
GGTCACTGGGGATCATTGGGGCCATTGGGGTCAATGGGGGTCAATGG
AGACATTGGGATCAGTGGGGGTCAATGGGGCCATTGTTGTCAATGGGG
CCATTGGGGTCAATGGGGTTCAATGGGGGCCATTGGGATCAATGGAGA
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AATGTGGGTCAATGGGGTCAATTGGGGTCACTGGGGCCAGTGGG

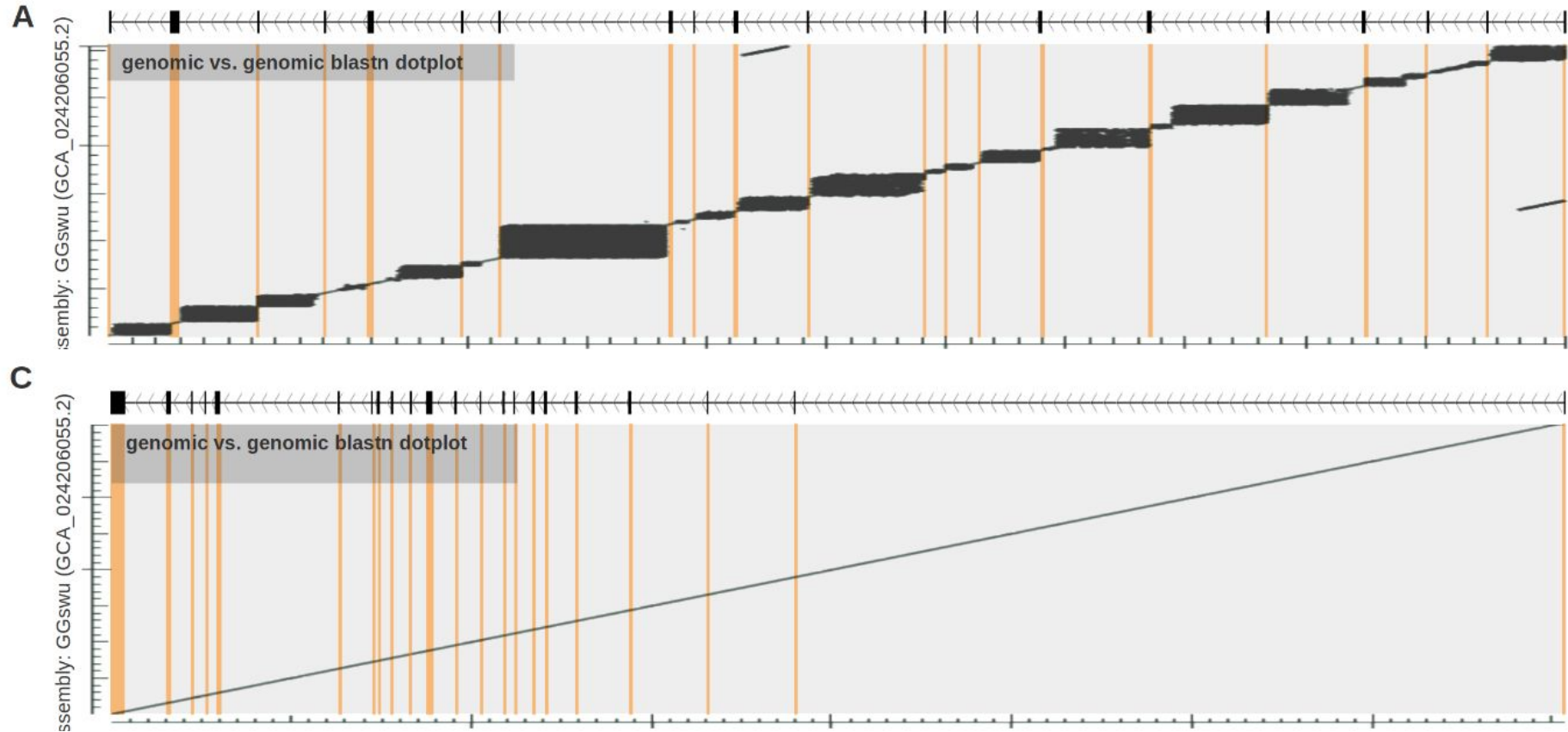
Thousands of hidden genes identified on microchromosomes



God bless Nanopore - new complete chicken genome



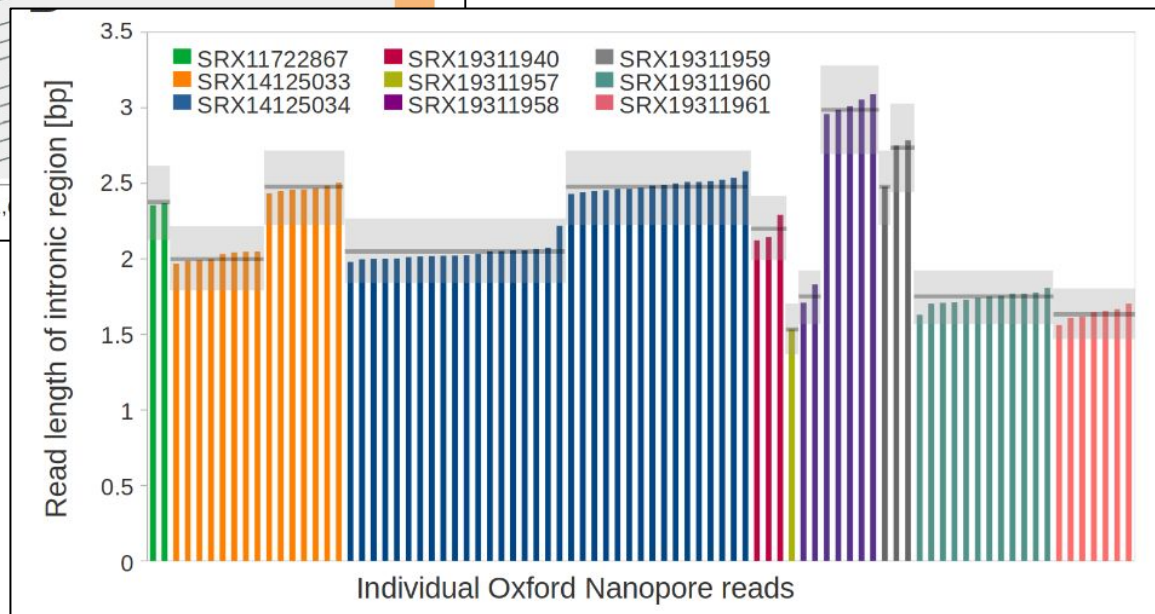
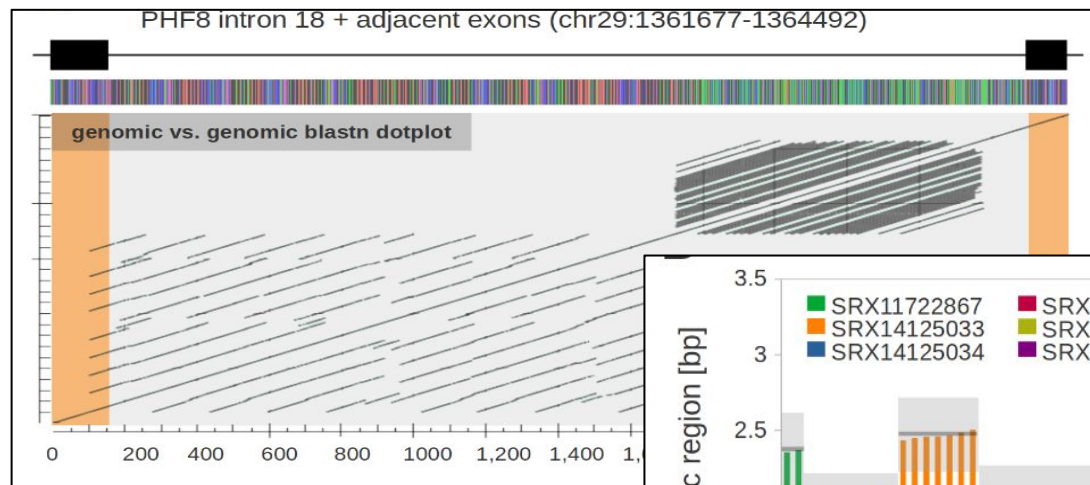
Local expansion of repeats on dot chromosomes



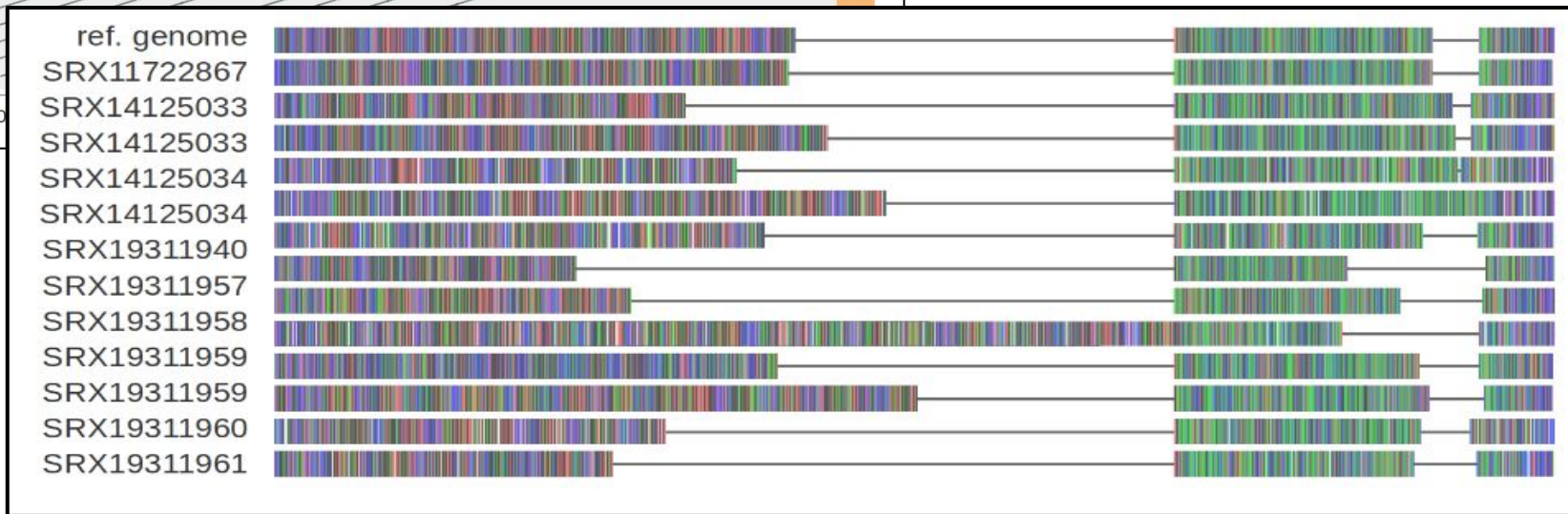
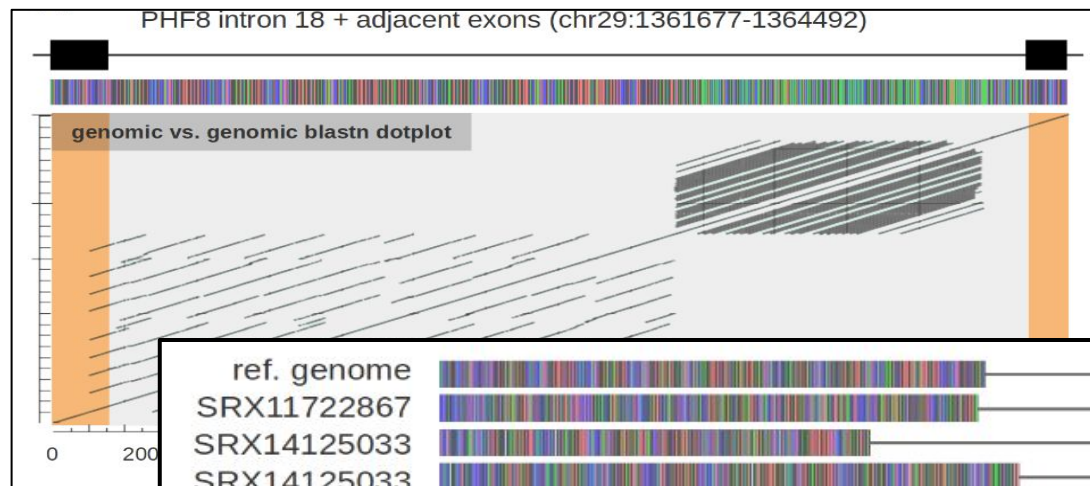
Unpublished



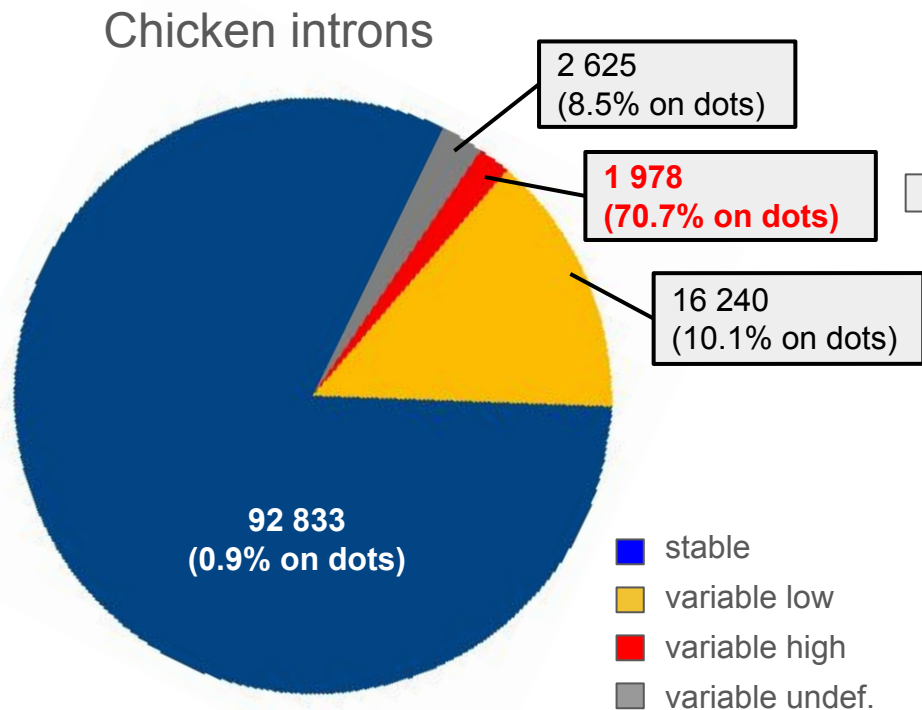
Local expansion of repeats on dots - length variability



Local expansion of repeats on dots - length variability

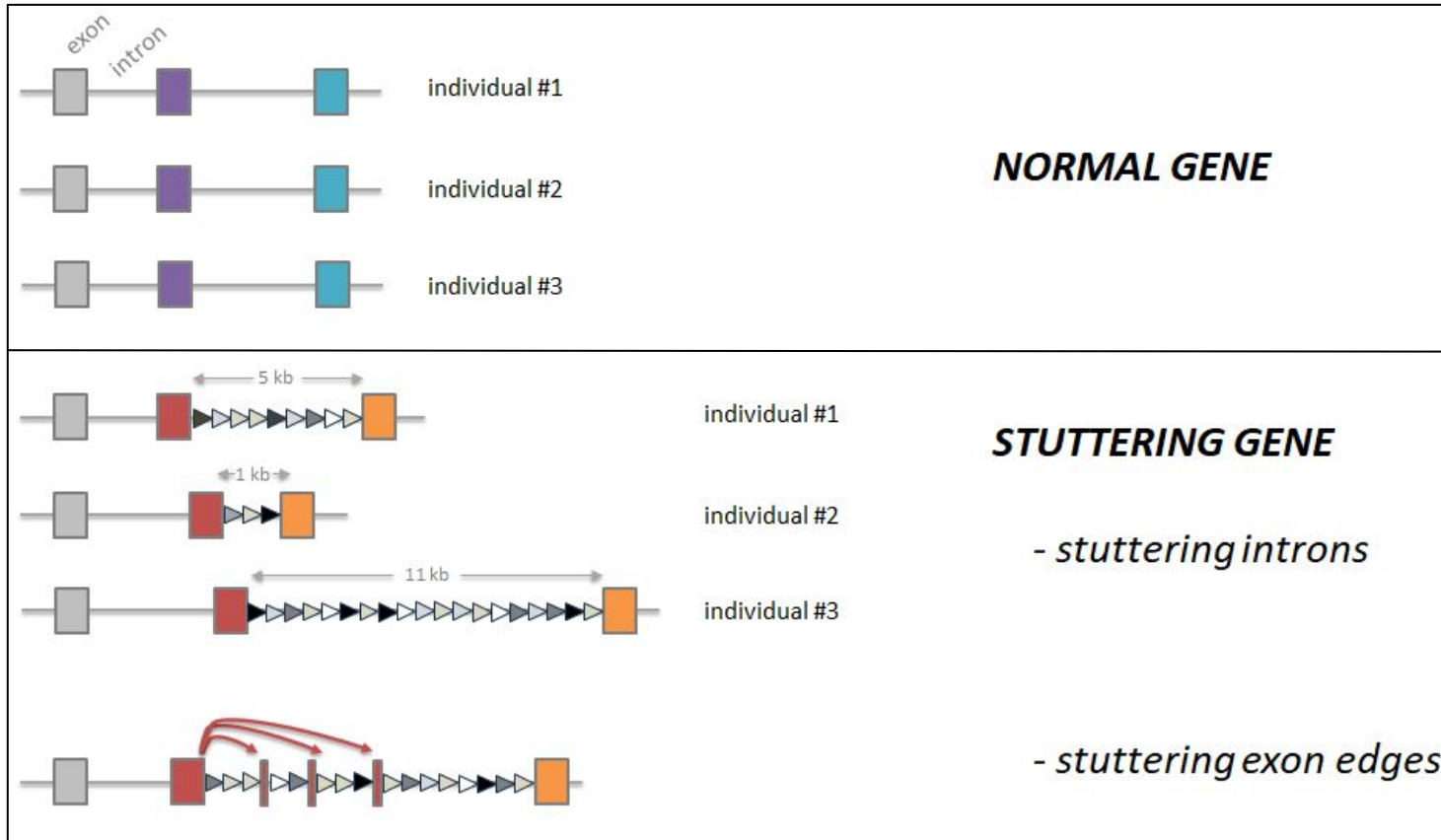


Local expansion of repeats on dots - Recent changes in chickens

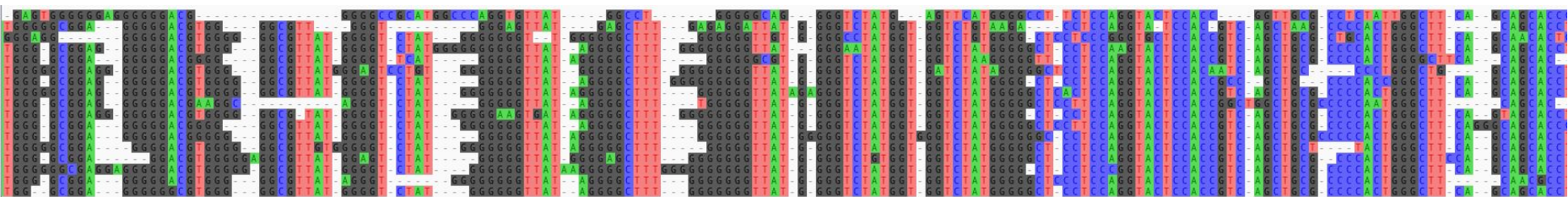
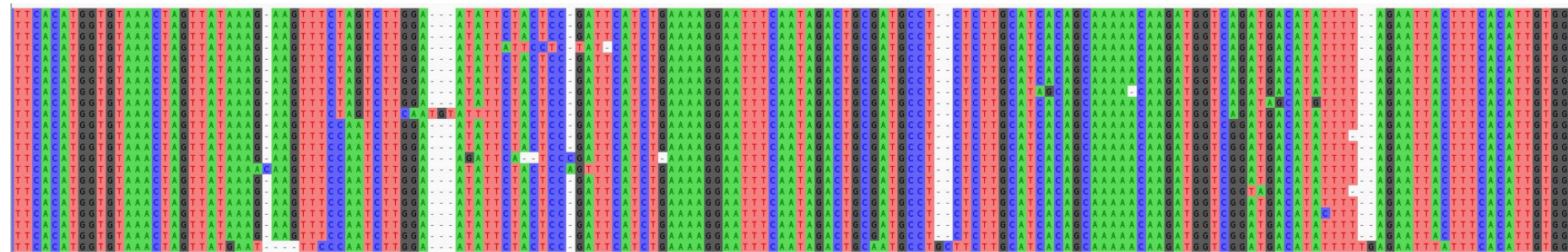


- highly variable in chicken population
- massive occurrence of repeat clusters
- almost exclusively on dot chromosome
- present also in other birds
- nothing like this in other vertebrates

Stuttering genes

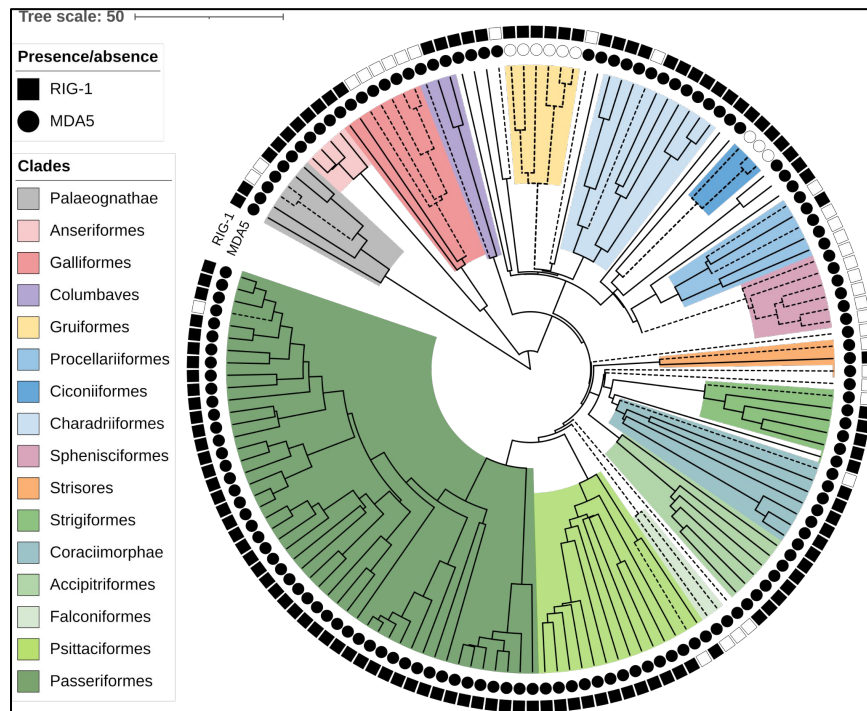


Even Nanopore data cannot go to single-base resolution

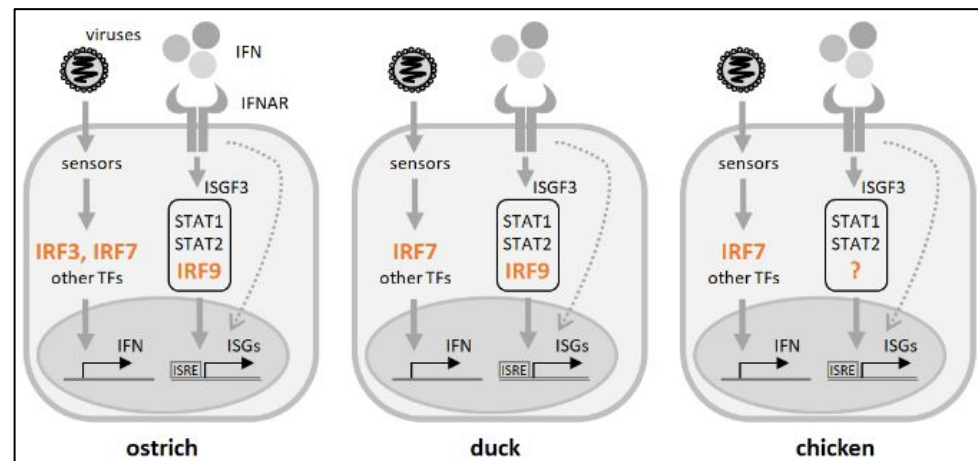


Hidden and missing genes in birds

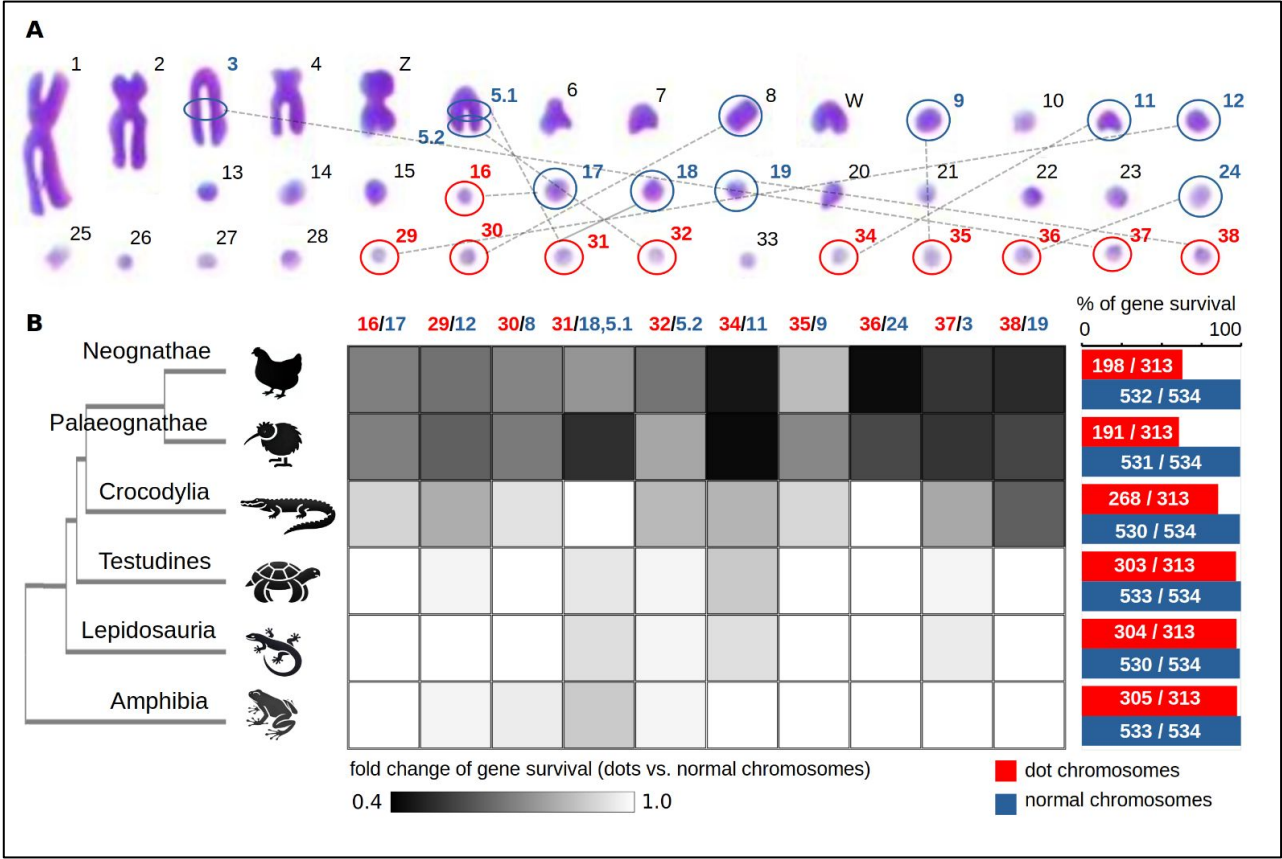
RNA virus sensors missing in birds



Interferon regulatory factors missing in birds

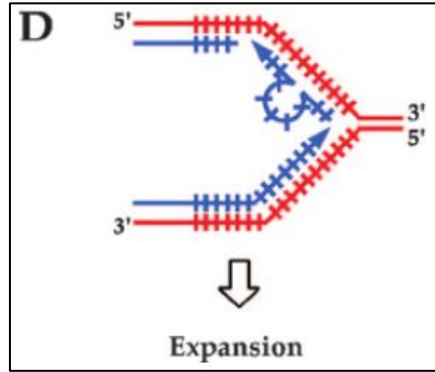


Missing genes on avian dots

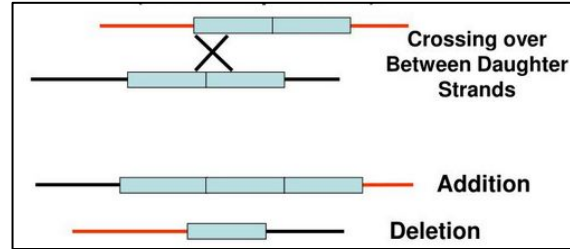


Potential mechanism of sequence stuttering on avian dots

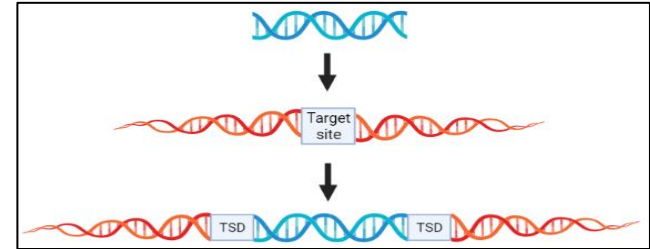
Dna replication slippage



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)

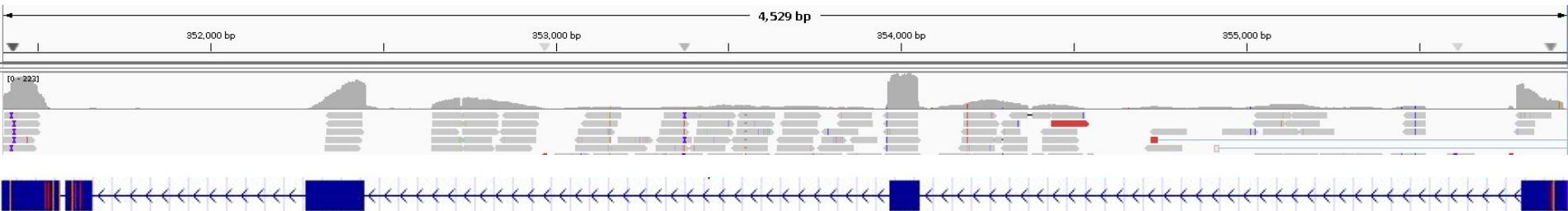
Why

G4 formation? Dot chromosome decay?

Thank you

Chicken erythropoietin (EPO)

up to 1000x coverage from RNAseq data



Avian Hidden Genes

