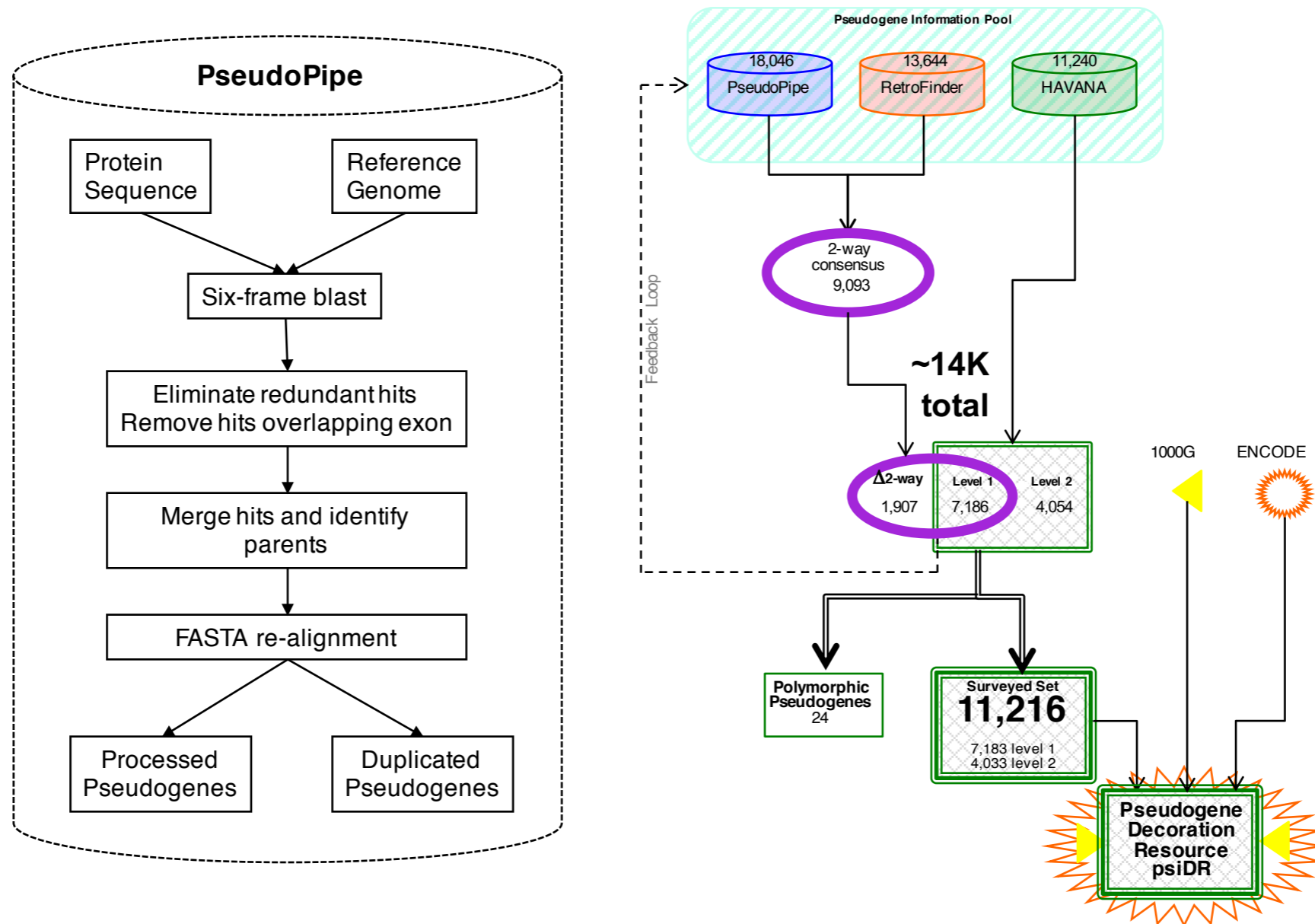


retroCNVs

CZ; MS; NA12878

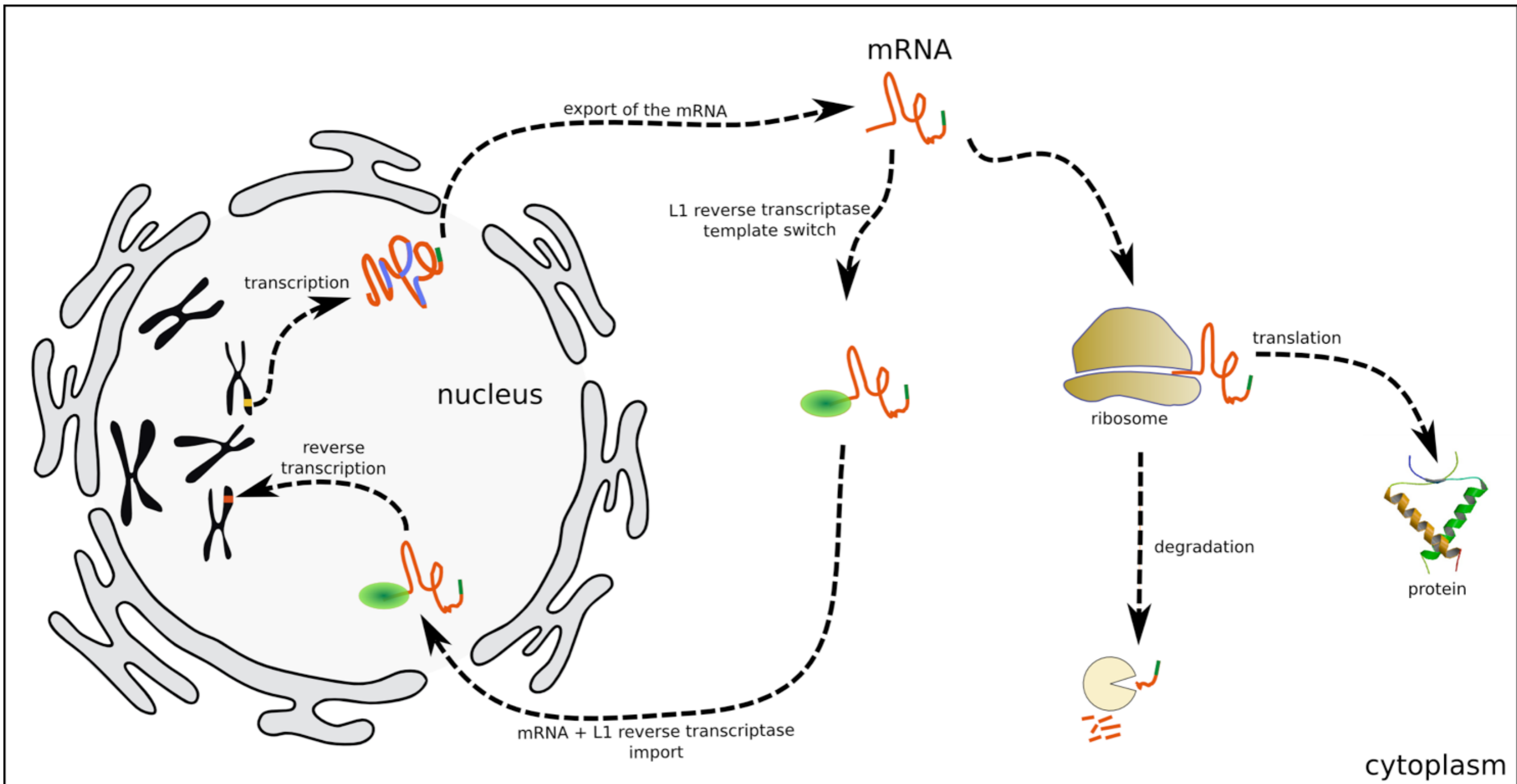
FN - Tech

# Pseudogenes in the Human Genome

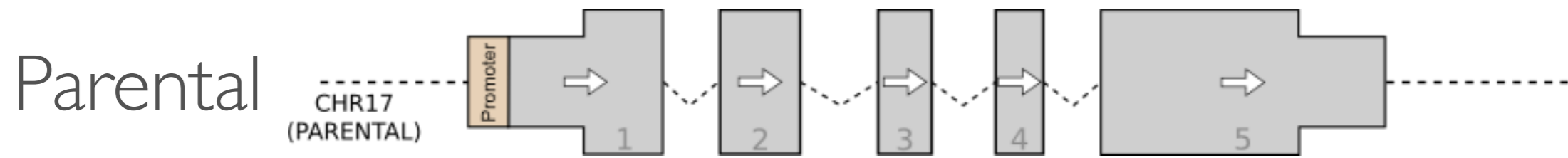


# Retrotransposition of mRNAs

origin of processed pseudogenes and retrogenes



# Involved sequences



mRNA



Retrocopy  
processed pseudogene  
and **retrogene**



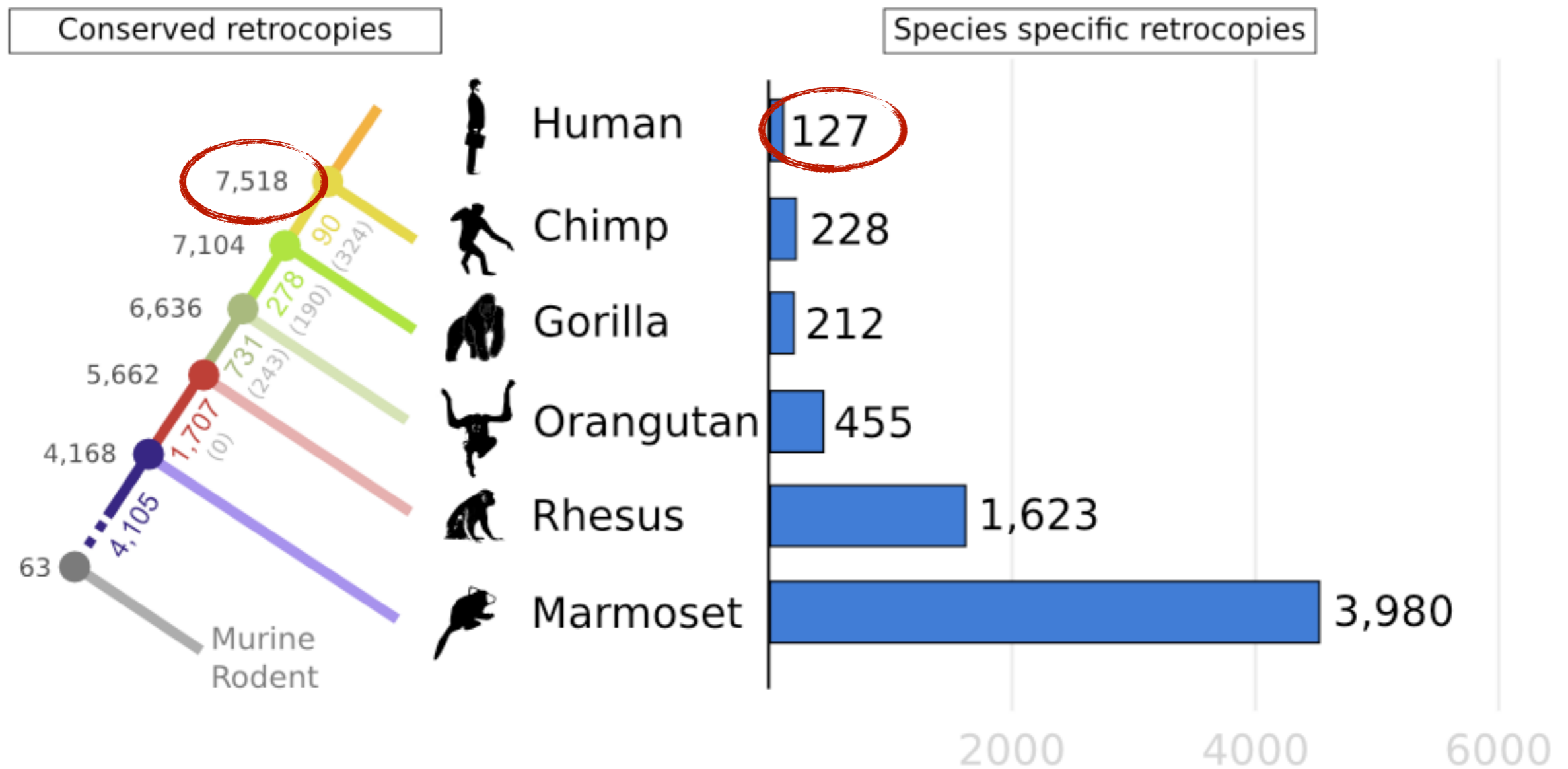
context

intergenic

intragenic  
(host gene)

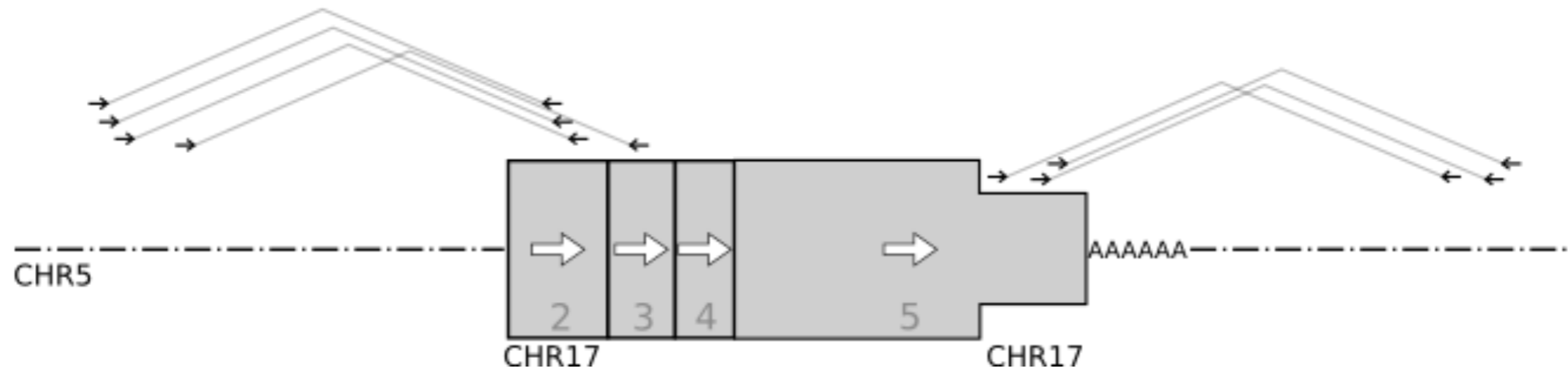
# Primate retrocopy evolution

(reference genome analysis)

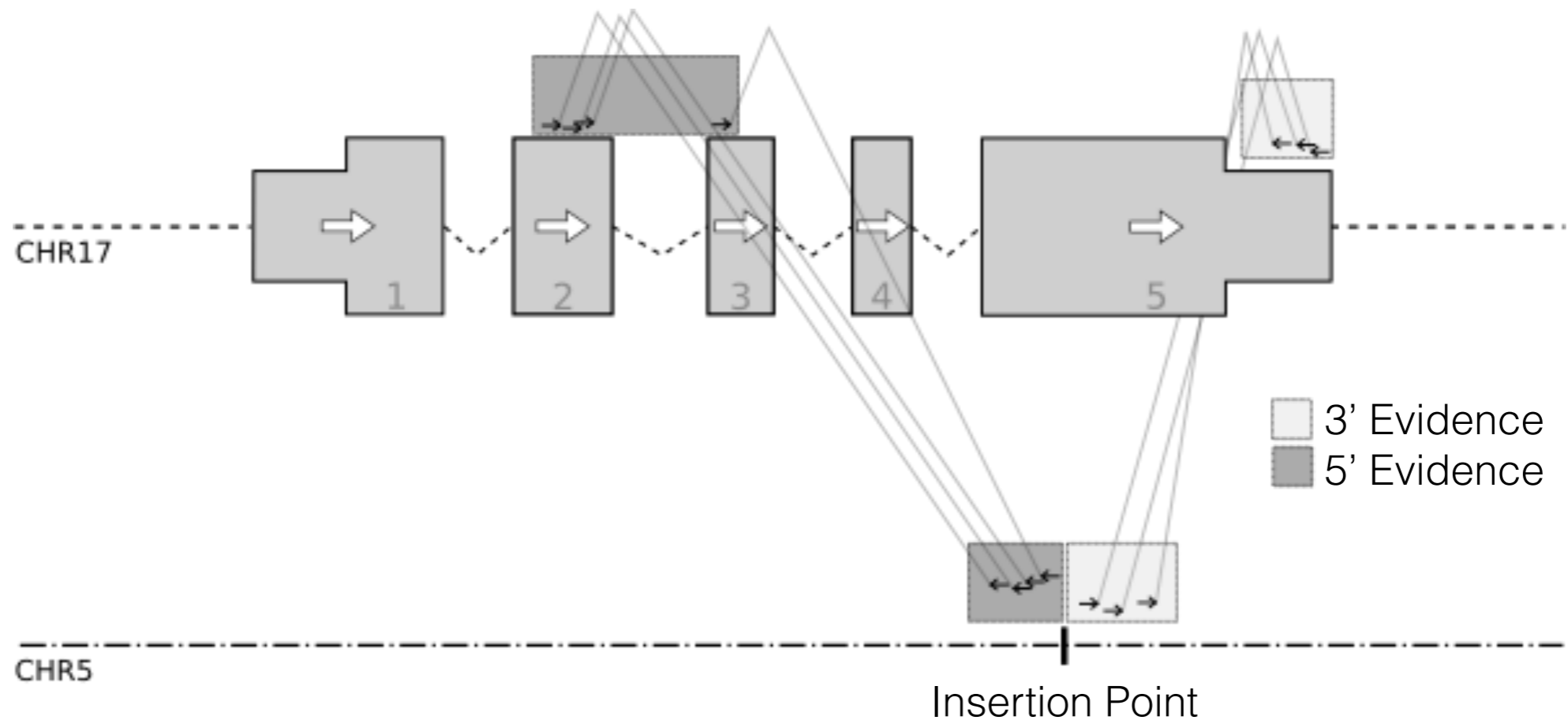


# “New retrocopies”

population/individual specific retrocopies

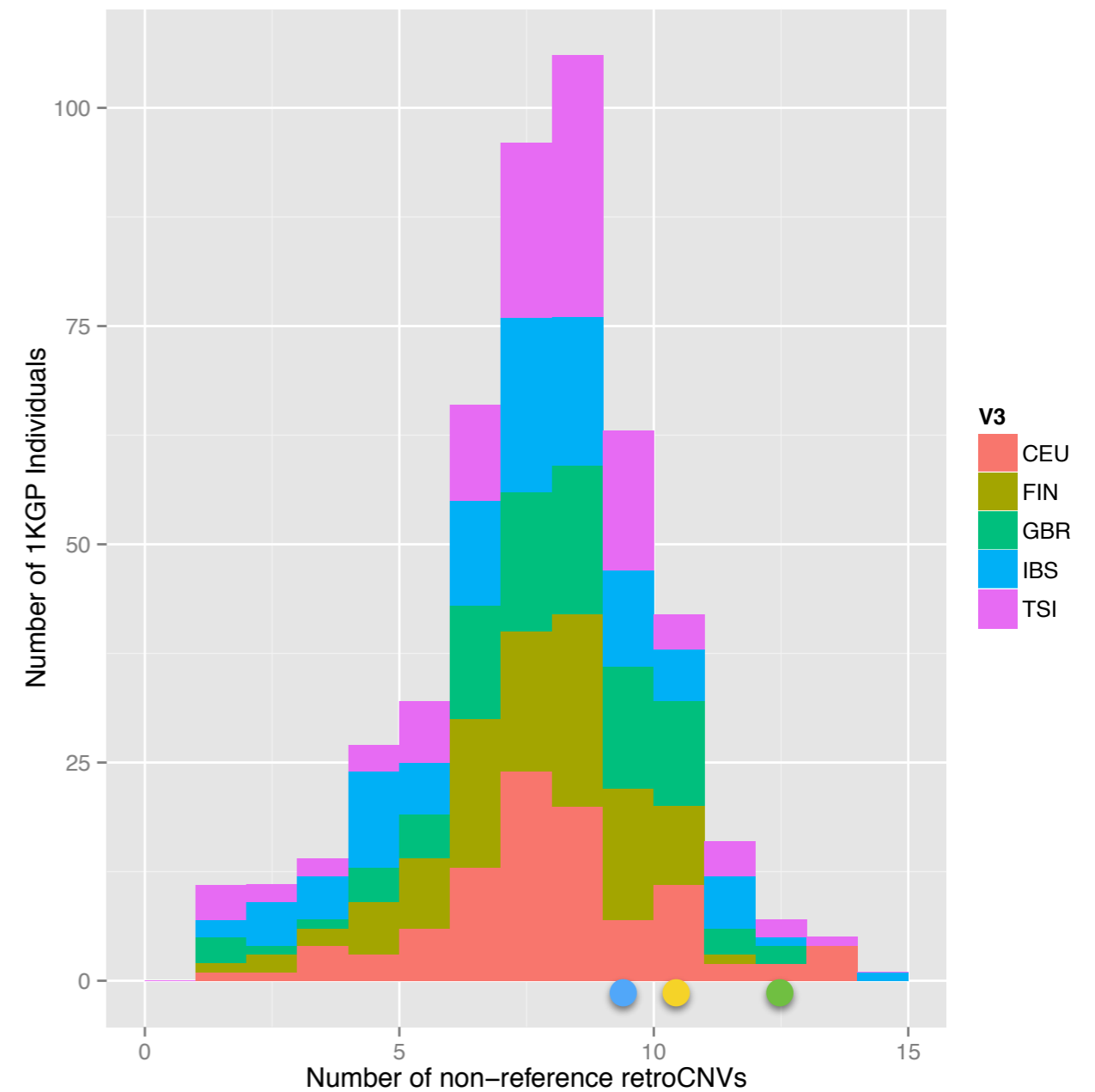


Alignment in the reference genome:



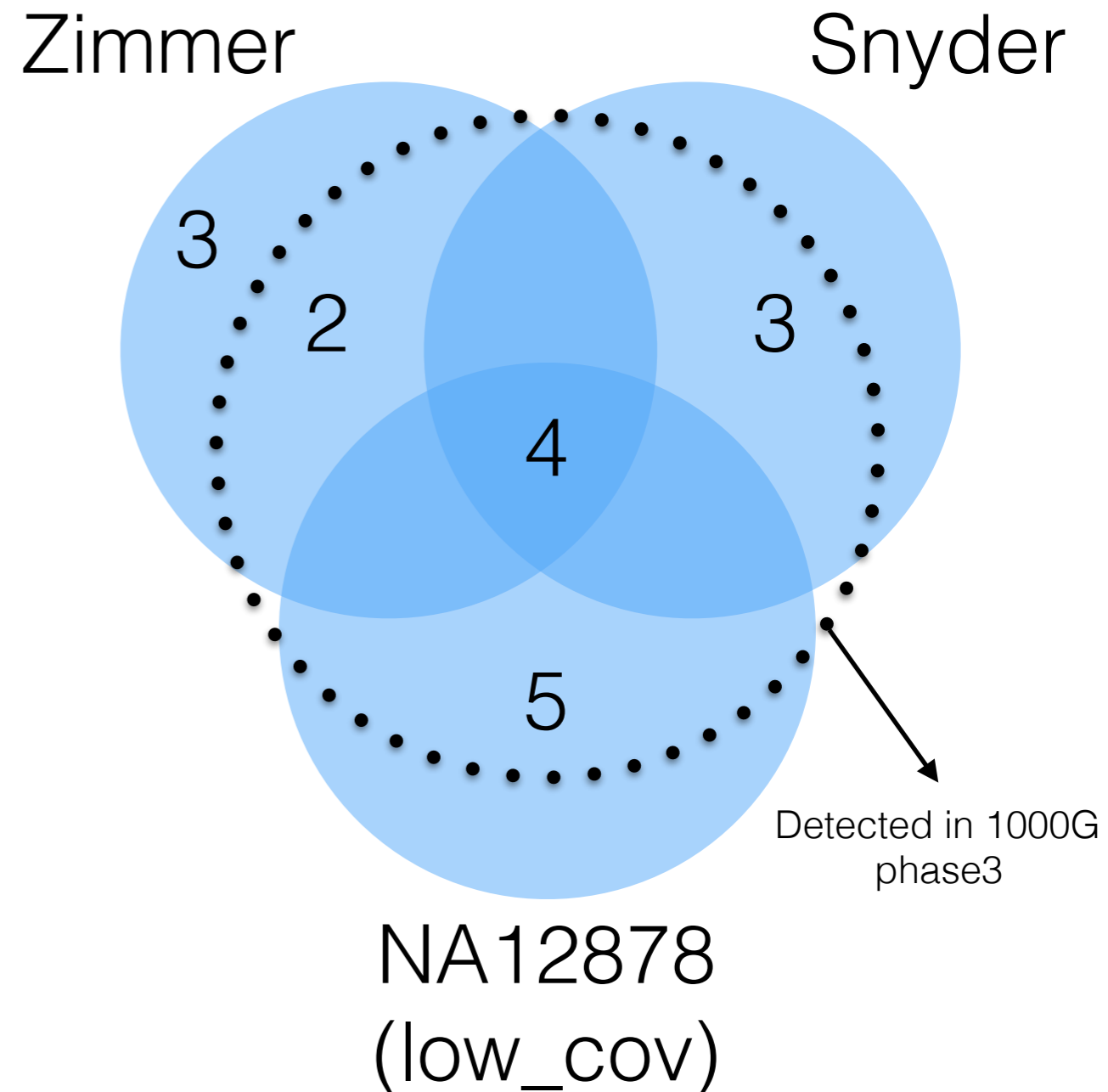
# 1000 Genomes retroCNVs

- C. Zimmer: 12 retroCNVs
- M. Snyder: 10 retroCNVs
- NA12878 (low-cov): 9 retroCNVs
- NA12878 (high-cov): X retroCNVs



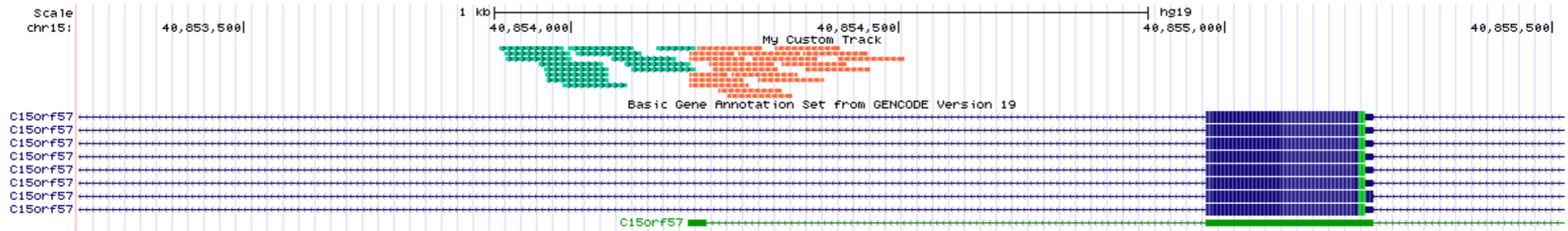
# Shared and Specific retroCNVs

- Zimmer specific insertions:
  - MFF\_chr15: freq. in AFR
  - FAM136A\_chrA: rare variant
- Snyder specific insertions:
  - ANPEP\_chr13: freq. in ASN
  - PPIA\_chr4: freq. south EUR
  - PRKRA\_chr3: freq. in ASN

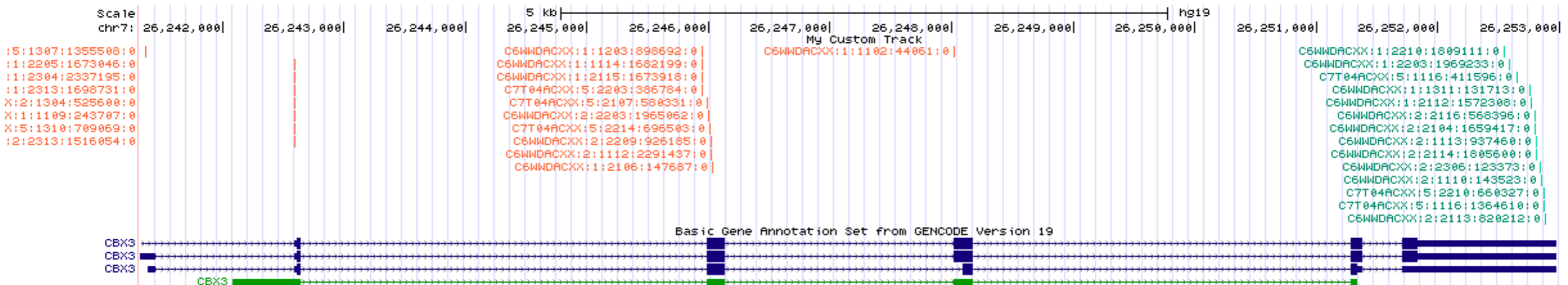




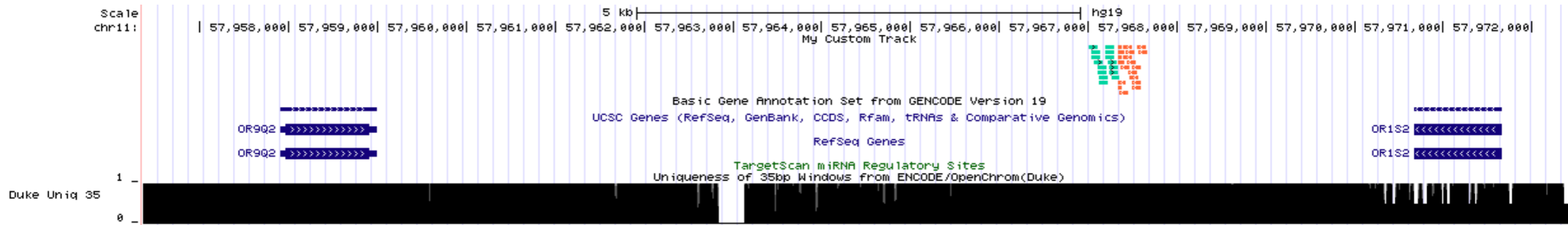
# CBX3: Insertion Point



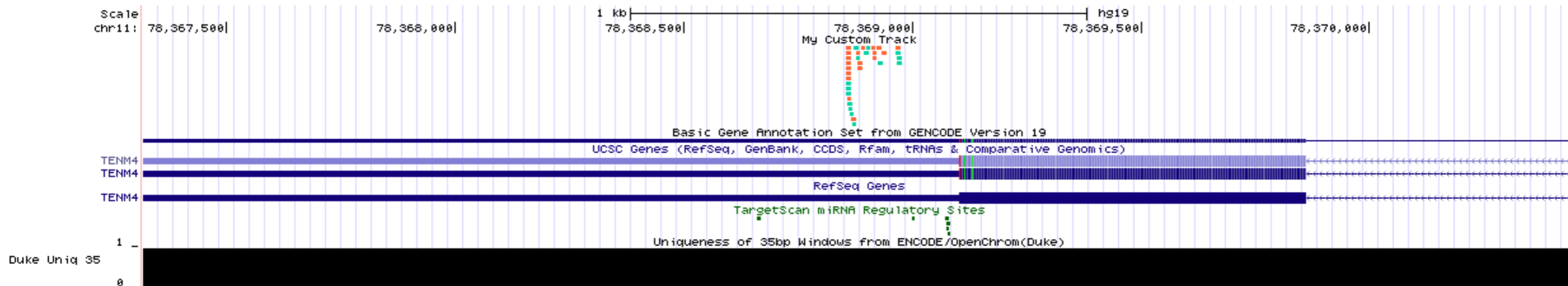
# CBX3: Parental



# TENM4: Insertion Point



# TENM4: Parental



# New retroCNVs

- TENM4  
Small segment 3'UTR  
Intergenic insertion close to ORs (3kb)  
"neural development, regulating the establishment of proper connectivity within the nervous system"
- FAM230  
Could be a huge deletion (only evidence from one extremity)  
Intergenic - gene desert  
unknown function
- ANKDR36  
3'UTR (only evidence from one extremity)  
Intragenic (NBPF25P)