

retroCNVs

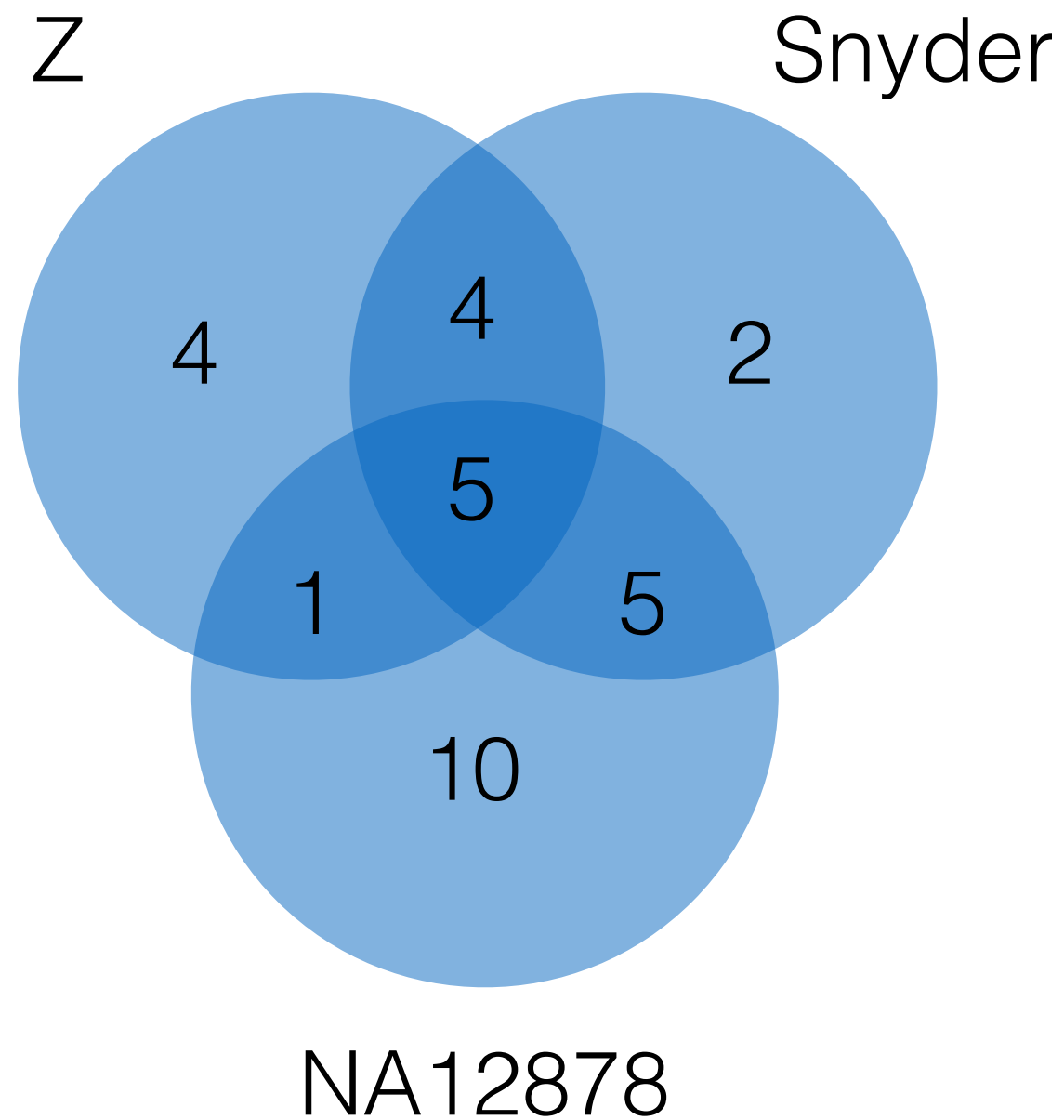
CZ; MS; NA12878

FN - Tech

	<b>Pseudogenes</b>	<b>Processed pseudogenes</b>	<b>Human specific processed pseudogenes</b>
<b>Human</b>	~14,000	7,831	127

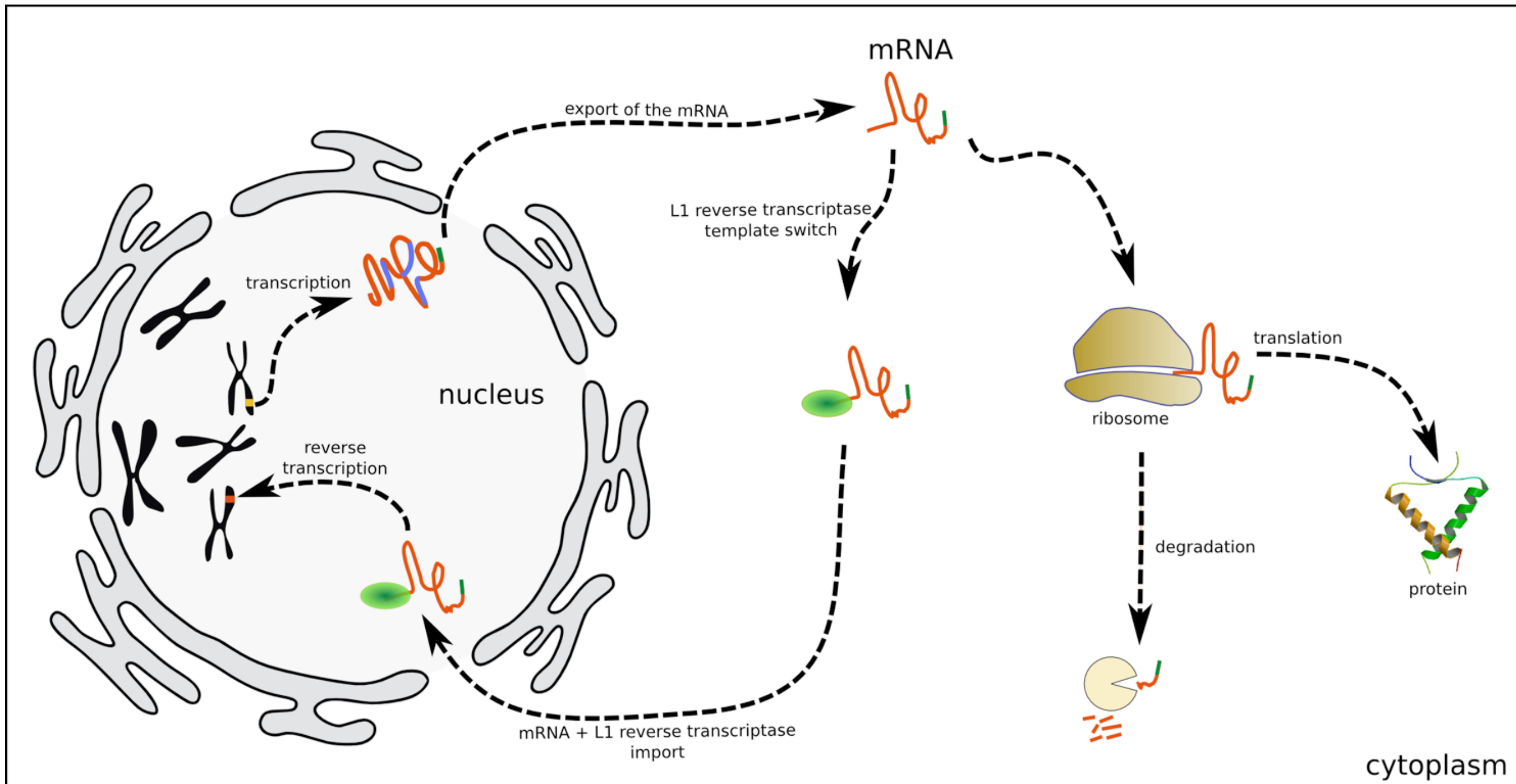
	<b># of ALUs in the genome</b>	<b>ALUY</b>	<b>Human specific ALUs</b>
<b>Human</b>	1,238,995	146,308	?

# pseudogene absence

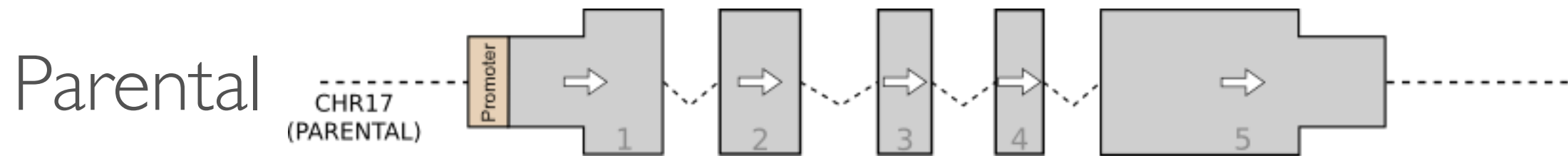


# Retrotransposition of mRNAs

origin of processed pseudogenes and retrogenes



# Involved sequences



mRNA



Retrocopy  
processed pseudogene  
and retrogene



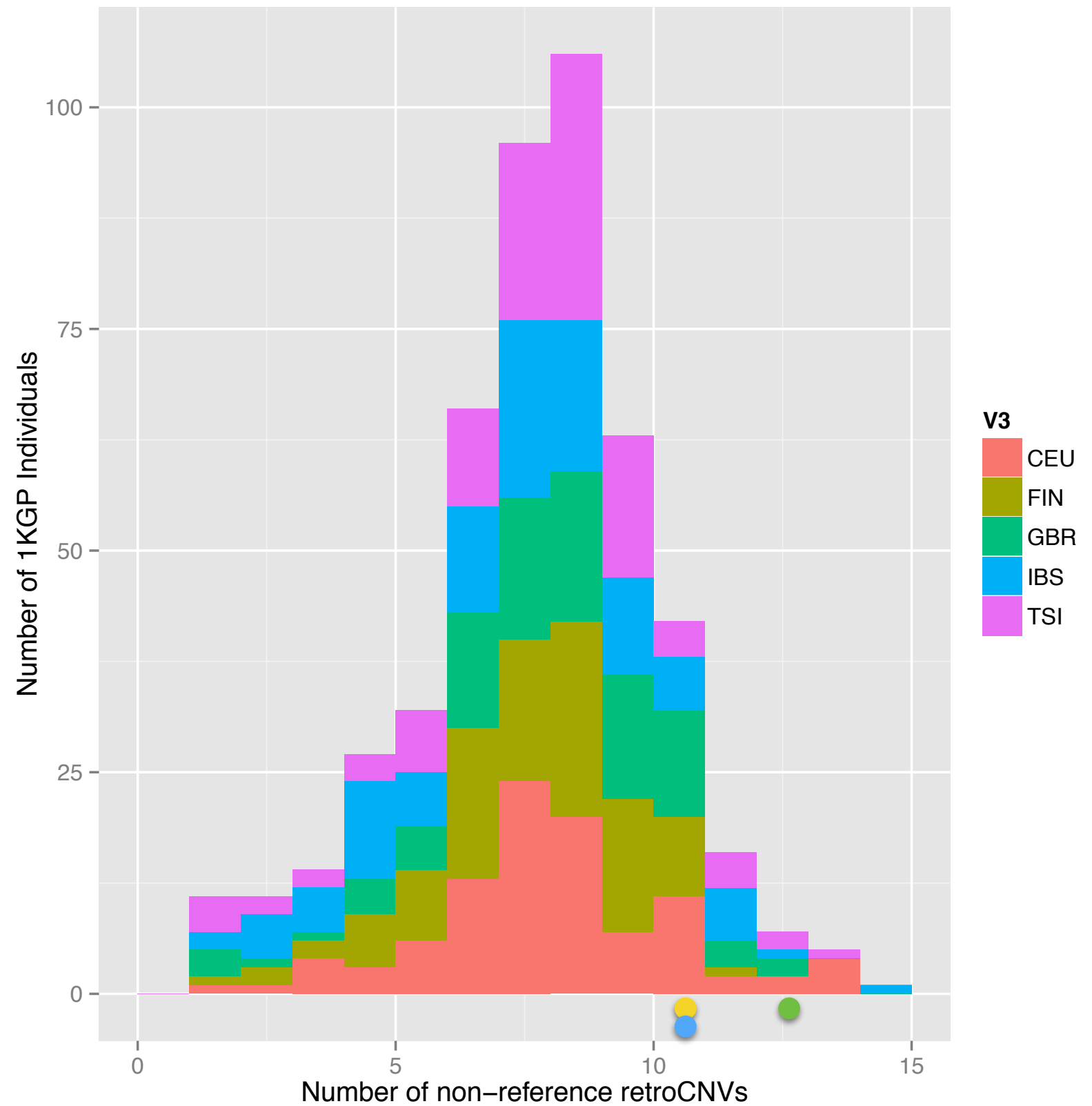
context

intergenic

intragenic  
(host gene)

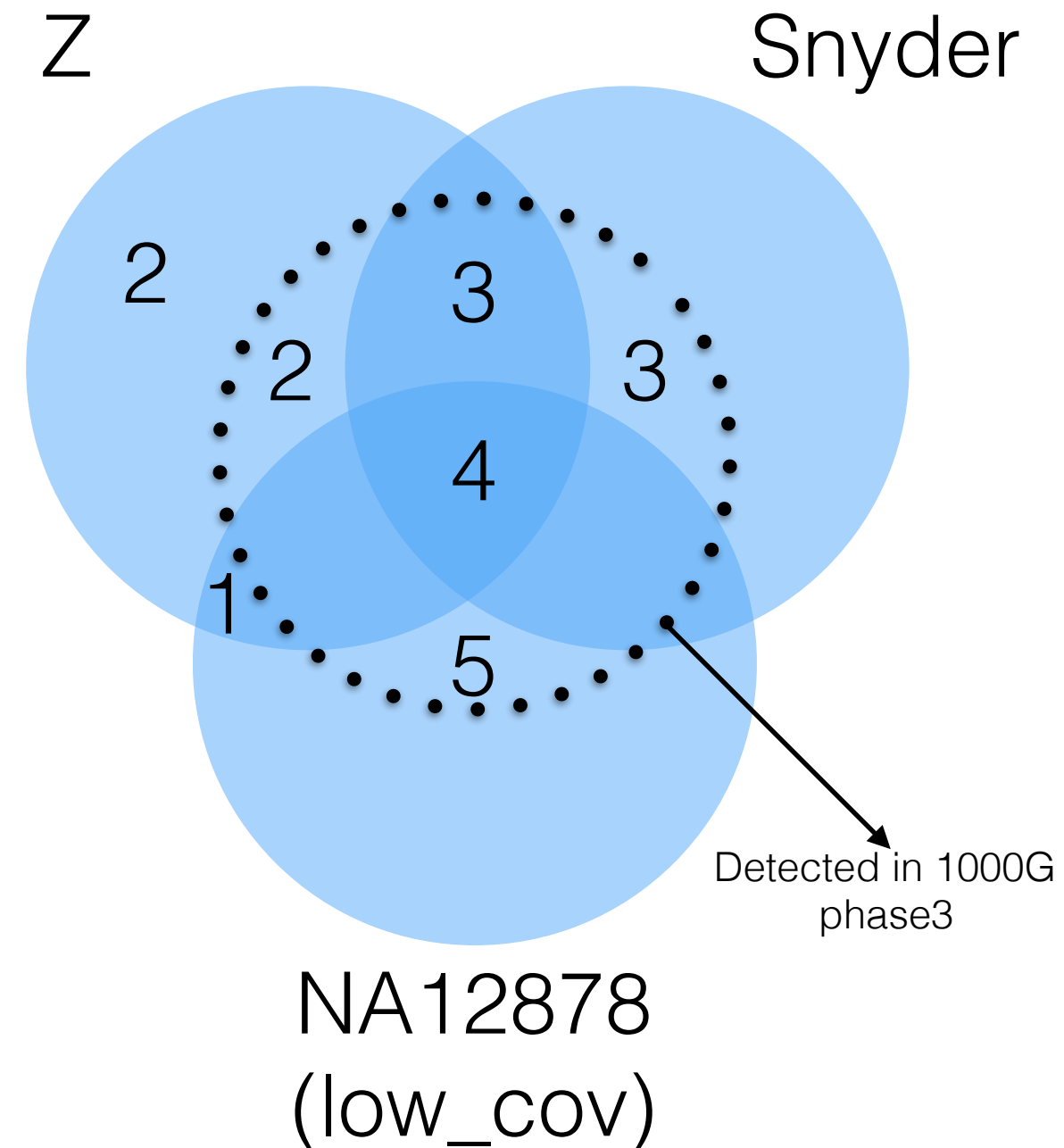
# 1000 Genomes retroCNVs

- Z: 12 retroCNVs
- M. Snyder: 10 retroCNVs
- NA12878: 10 retroCNVs



# Shared and Specific retroCNVs

- Z 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

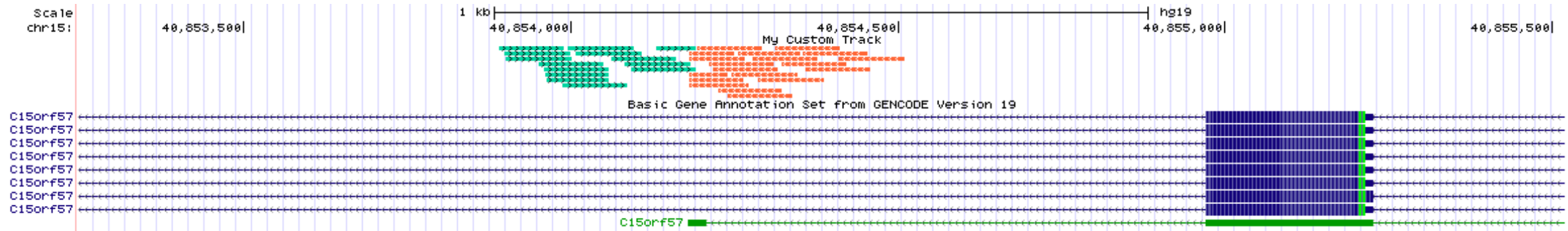


# New retroCNVs

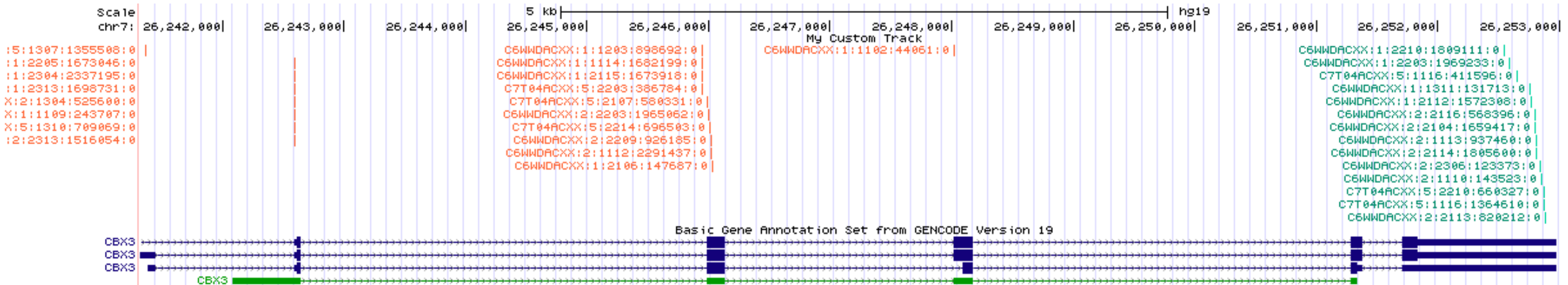
- TENM4 (Z and NA12878)  
Small segment 3'UTR  
Intergenic insertion close to ORs (3kb)  
"neural development, regulating the establishment of proper connectivity within the nervous system"
- FAM230  
Could be a huge deletion (only evidence from one extremity)  
Intergenic - gene desert  
unknown function
- ANKDR36  
3'UTR (only evidence from one extremity)  
Intragenic (NBPF25P)



# CBX3: Insertion Point

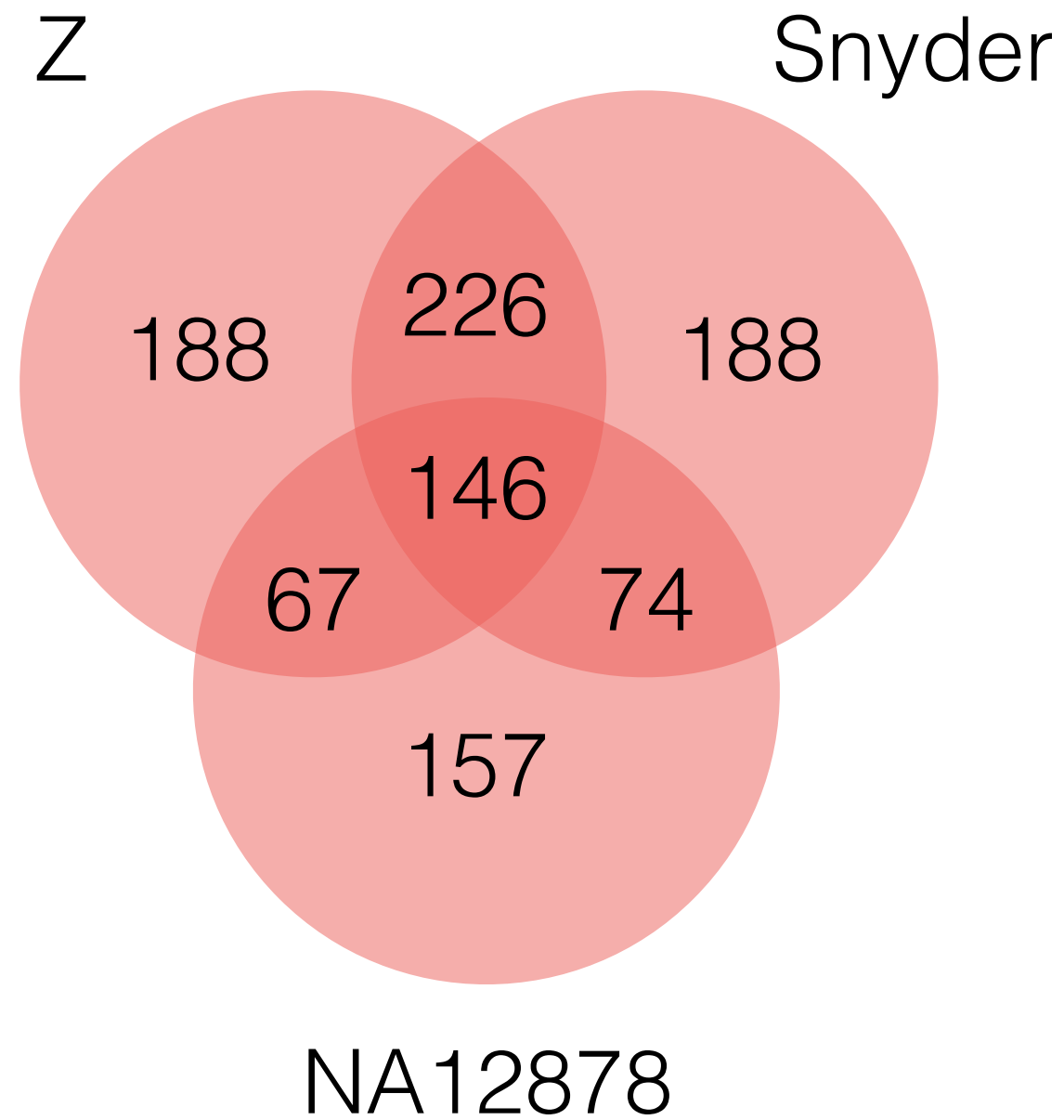


# CBX3: Parental



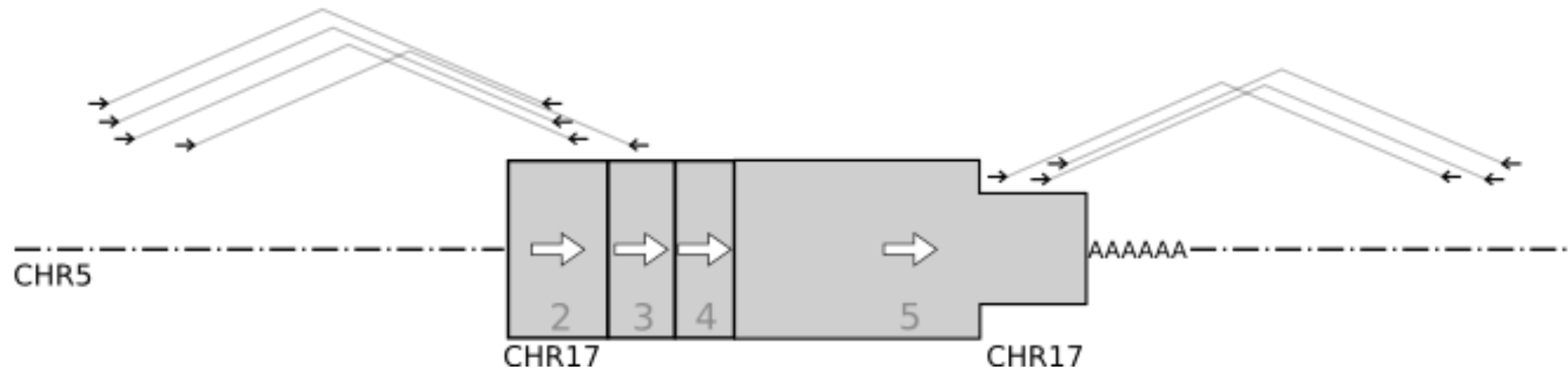


# Shared and Specific ALUs



# Detection of new retrocopies

population/individual specific retrocopies



Alignment in the reference genome:

