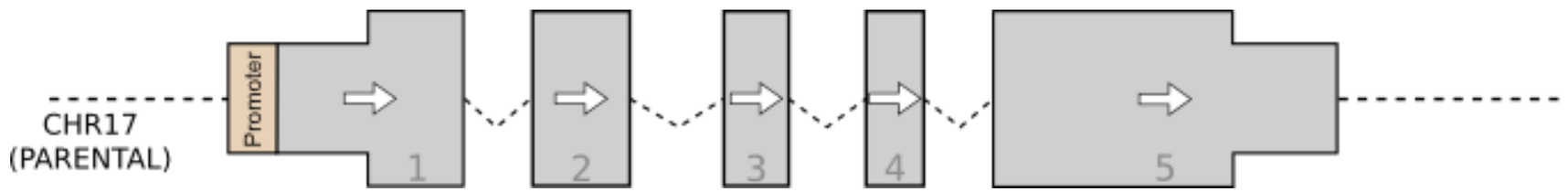
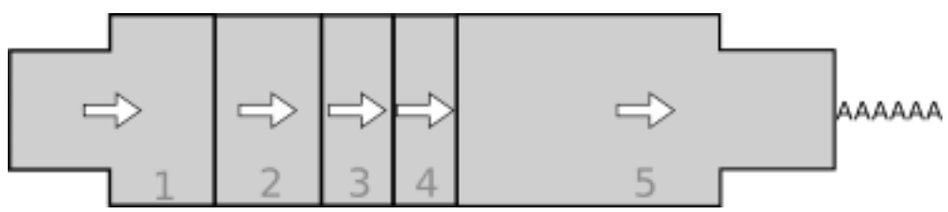
retroCNVs
CZ; MS; NA12878
FN - Tech

Involved sequences

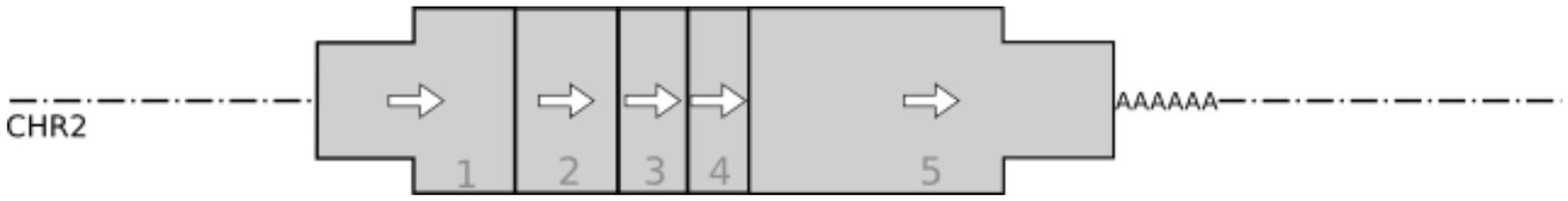
Parental



mRNA



Retrocopy

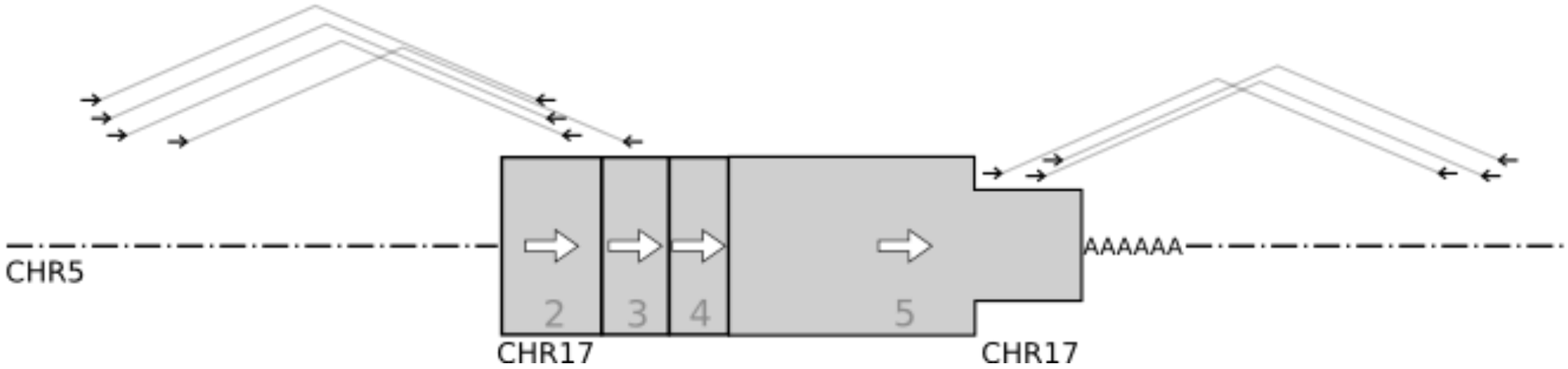


context

intergenic

intragenic
(host gene)

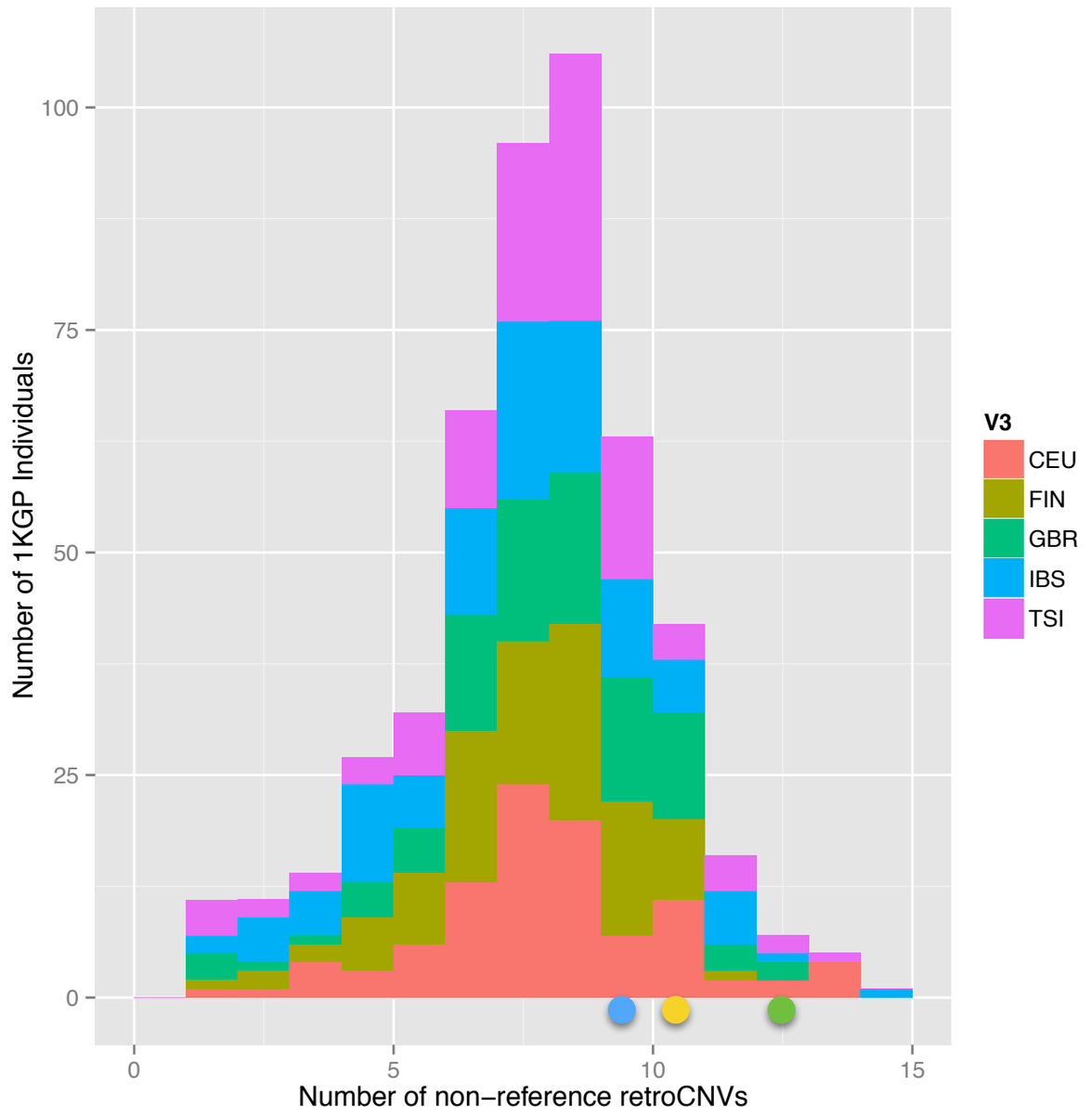
“New retrocopies”



Alignment in the reference genome:

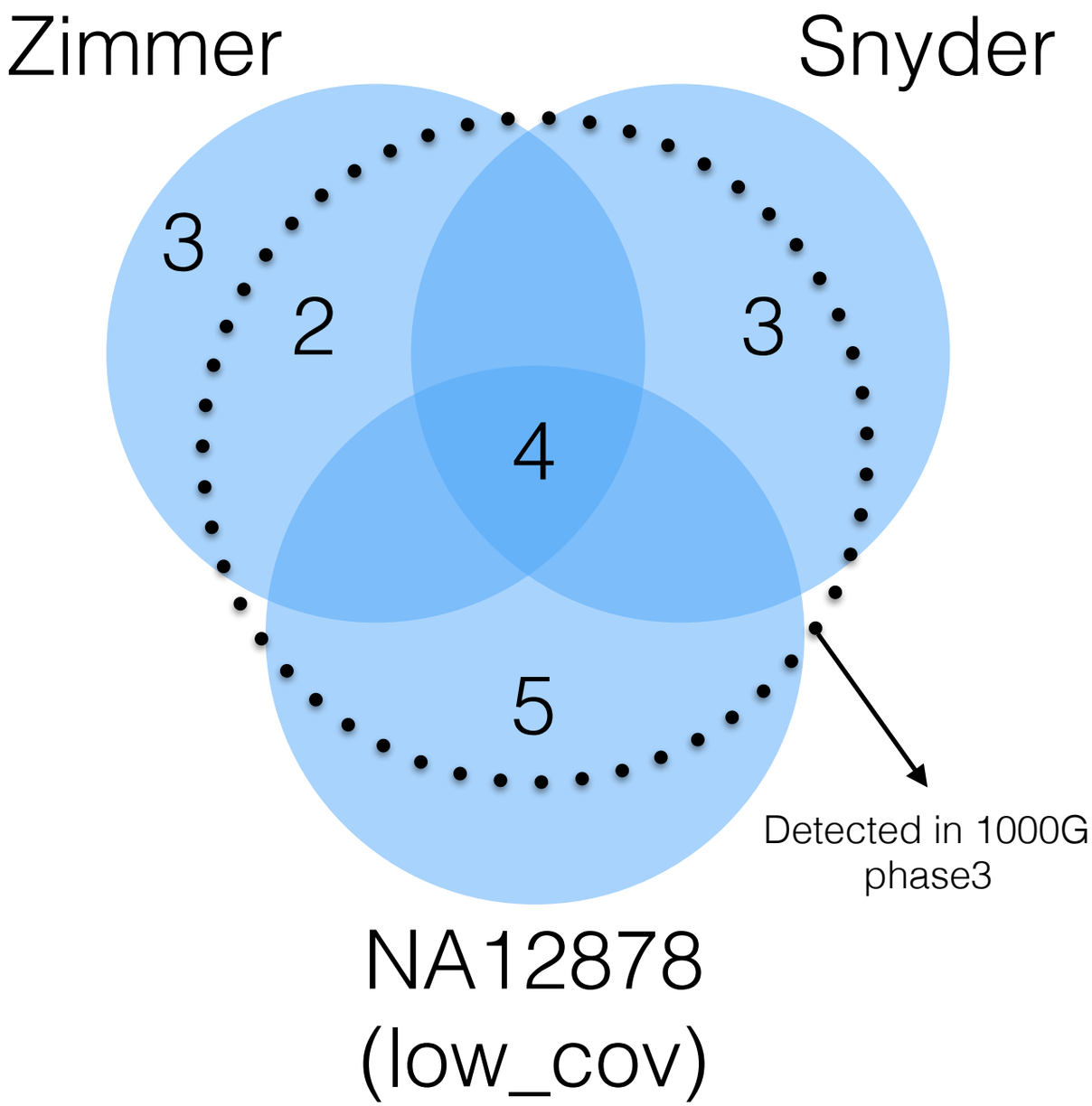
1000 Genomes retroCNVs

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

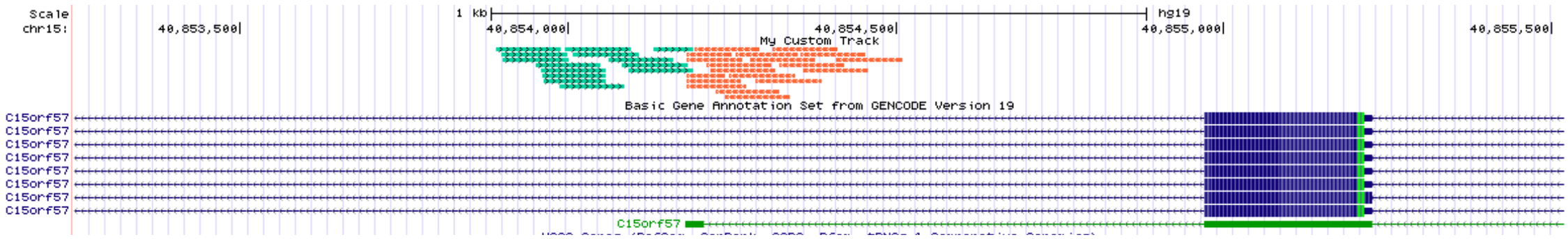


Shared and Specific retroCNVs

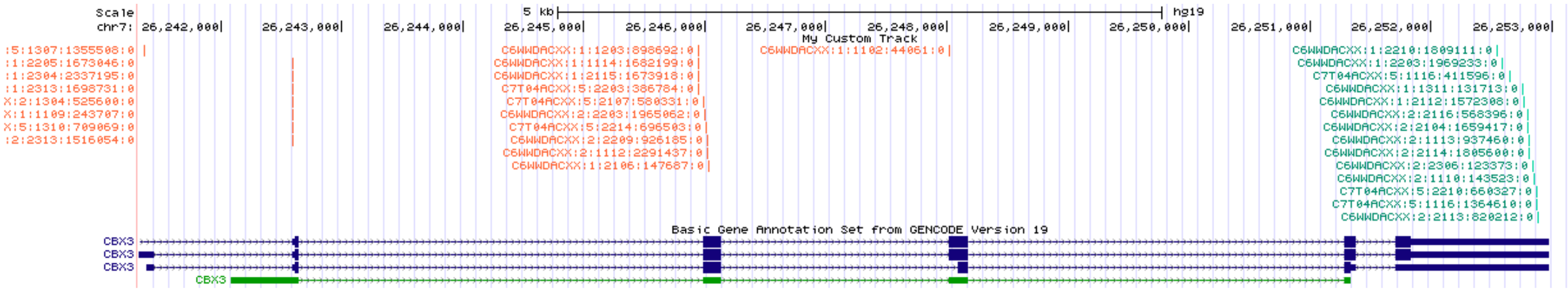
- Zimmer specific insertions:
  - MFF_chr15: freq. in AFR
  - FAM136A_chrA: rare variant
- Snyder specific insertions:
  - ANPEP_chr13: freq. in ASN
  - PPIA_chr4: freq. south EUR
  - PRKRA_chr3: freq. in ASN



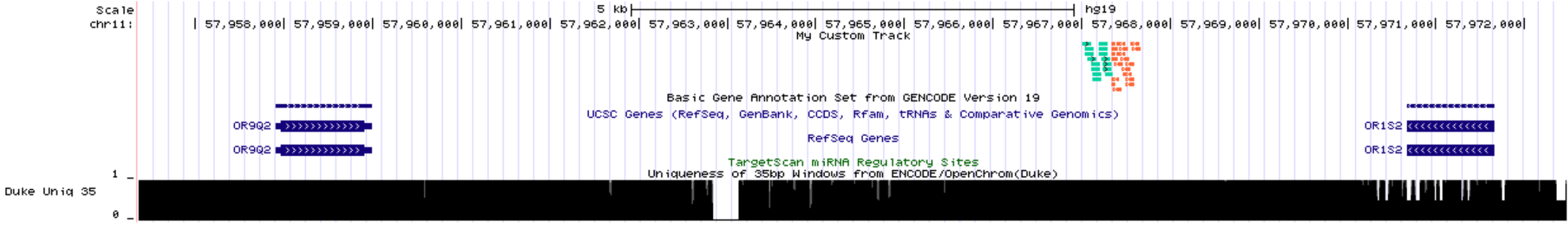
CBX3: Insertion Point



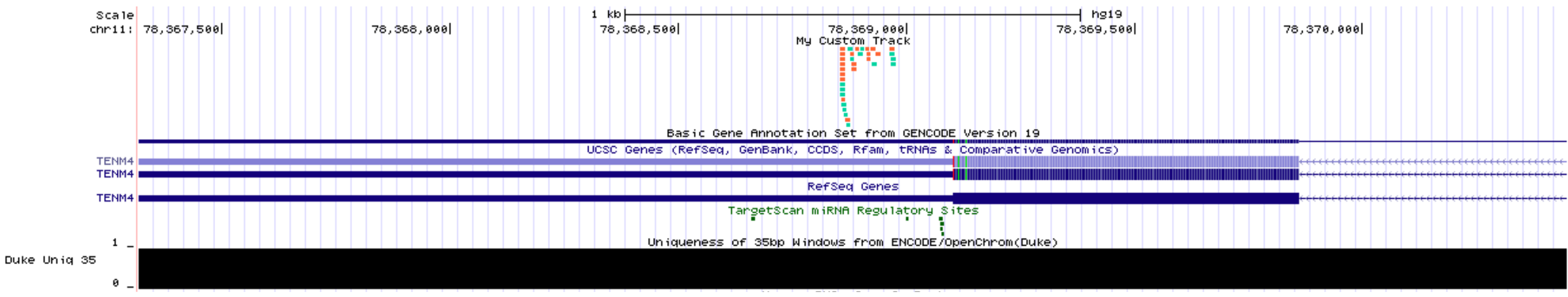
CBX3: Parental



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)