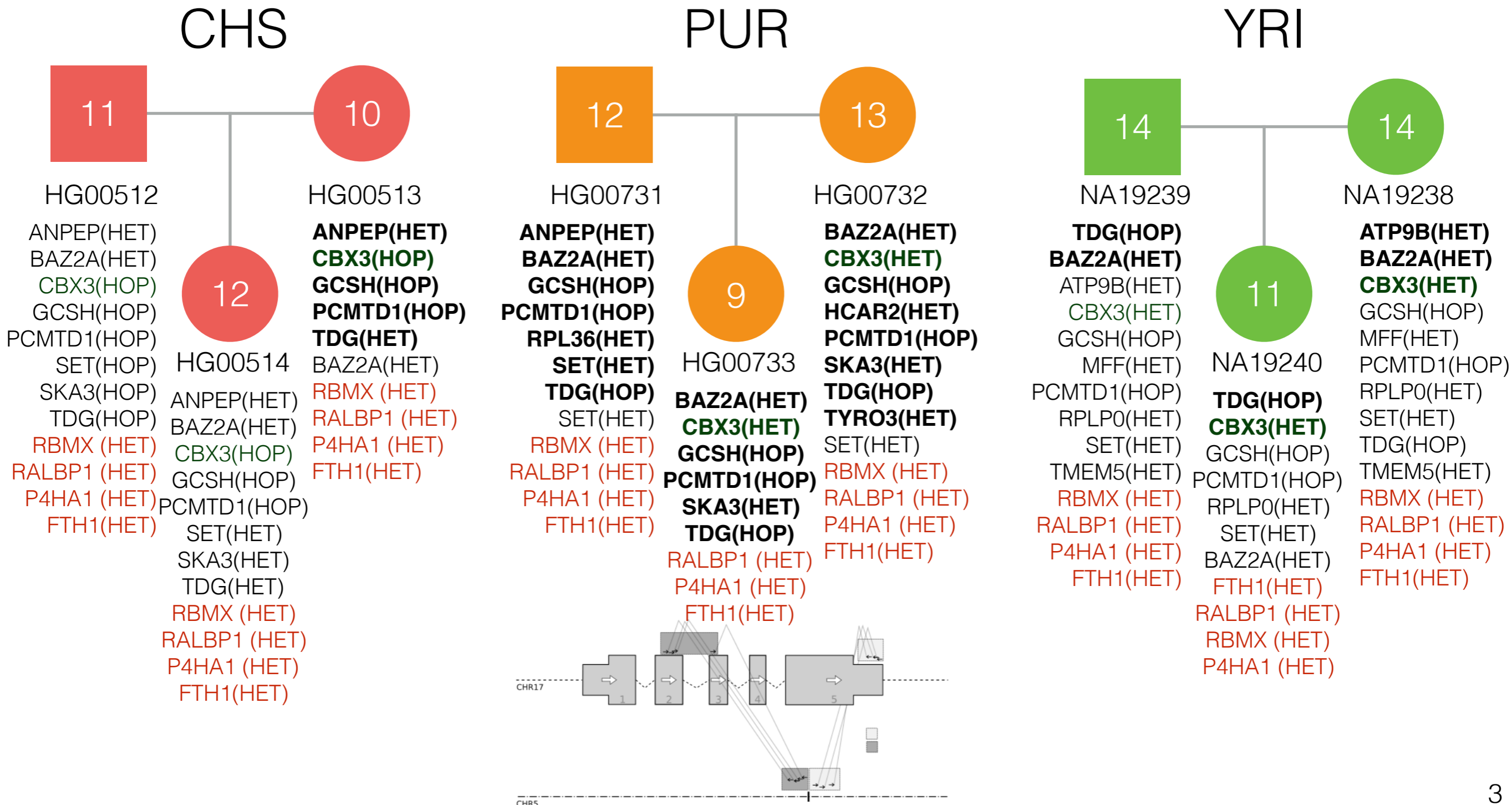


retroCNV (retrodup)  
calling pipeline  
1000g-sv-Trio

# “Available data”

- **Low coverage Illumina (phase3)**
- **High Coverage Illumina**
- **PacBio**
- Bionano 90x  
No sequence info
- Jumping library  
Very short reads
- Strand-Seq  
CHS trio
- 10x Genomics  
Unaccessible
- Moleculo  
Not uploaded
- ACGH (array)
- RNA-Seq (PacBio)
- RNA-Seq (Illumina)

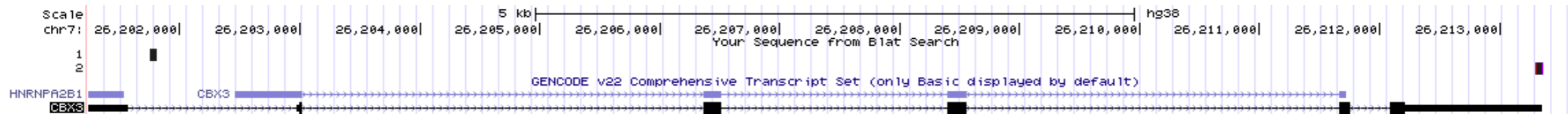
# High coverage PCR free



# Breakpoint analysis Illumina High Coverage

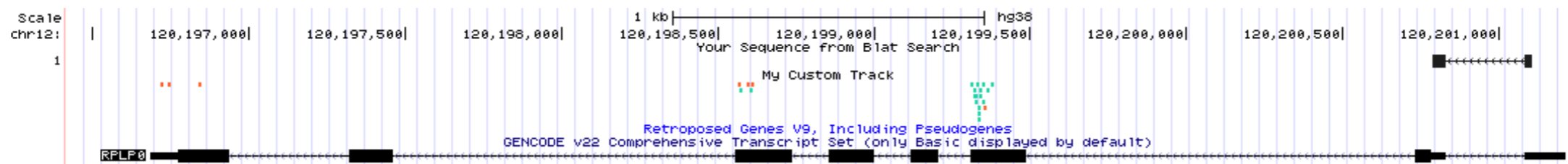
## CBX3 inserted into chr15

```
chr15 40561981 0 313 CTTCCGGATGTGGCTTGAGCTGTAGGCGCGGAGGGCCGGAGACGCTGCAGACCCGCGACCCGGAG
chr15 40561992 1 46 ATTTTTTTTTTTAAAGAAATATAACTATTTATTAACCACTGTTTCAGTATTTACAATAAAGTAAAC
```



## RPLP0 inserted into chr11

```
chr11 60274156 0 6 TTTTTTTTTTTTTTTTTTAAGAATTAAGCCTTTTTTTCTTTTTTTTTTAATTAATCTGGCATAGTTGGTTATTTTTTGTGT
chr11 60274167 0 55 CTCTCGCCAGGCGTCCTCGTGGAAGTGACATCGTCTTTAAACCCTGCGTGGCAATCCCTGACGCACCGCC
```

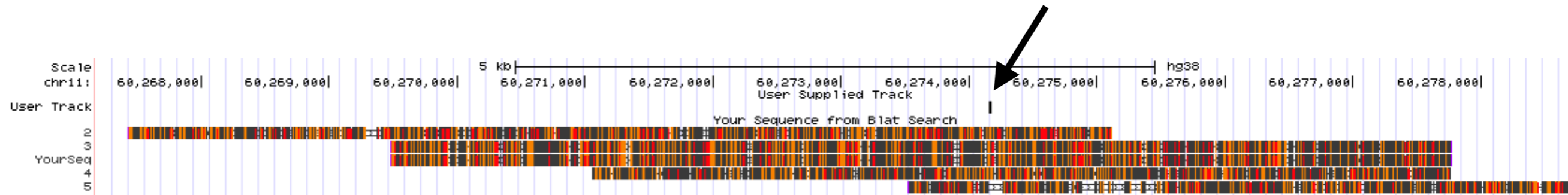


**Breakpoints analysis is able to identify all insertions extremities,  
as well as Target Site Duplications and poly(A)s**

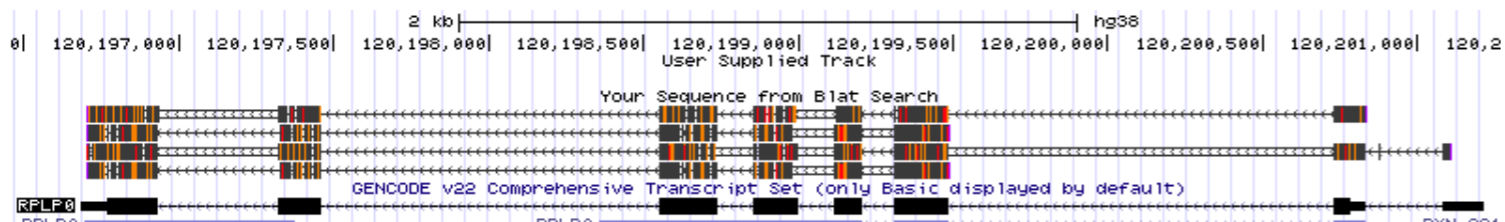
# RPLP0

chr11:60274156-60274179

## Insertion Point



## Parental Gene



|            |            |            |            |            |      |
|------------|------------|------------|------------|------------|------|
| CTCTGCCA   | GGggCgTCCT | CaGTGGAAGT | GACATCGcTC | TTTAAACCCT | 5050 |
| GCGTGCAATC | CCTGaACGCA | CCGCCGTGAT | GCCAGGGAA  | GACAaGGGCG | 5100 |
| ACCTGGAAGT | CCAACTACTT | CTTAAGaATC | ATCCAACTAa | TGGATGATTA | 5150 |
| TCCgAAATGt | TTTCATtGTG | GGAGCAGACA | ATGTGGGCTC | CAAGCAGATG | 5200 |
| gCAGCAGATC | CGaATGTCCC | tTTCGCGGGA | AGGctGTGGT | GCTGATGGGC | 5250 |
| AAGAACACCA | TGATGCGCAA | GGCCATCCGA | GGCACCTGGA | AAACAACCCA | 5300 |
| GCTCTGagaA | AACTGCTGCC | TCgttTATCc | GGGGGAATGT | GGGCTTTGTG | 5350 |
| TTCACCAAGG | AGGACCgtTC | ACTGAaGATC | AGGacCATGT | TGCctggcCC | 5400 |
| ...        |            |            |            |            |      |
| ATgcagaCCC | ATTCTAaTCA | TCAACaGGGT | AgCAAACGAG | TCCTCCTTGT | 5850 |
| CTGTGGAGAC | GGATTACACC | TTCCAaTTGC | TGAAGtCAA  | GGCCTTCTTG | 5900 |
| GCTaCCATCT | gGCCTTTGTG | GCTGCcTGCC | CCTGTGGCTG | CTGCACCACA | 5950 |
| GCTCTCCaTG | CTGCTGCaTG | CAGcCCCCAG | CTAAGGTTGA | AGCCAAGGAA | 6000 |
| GAGTaGGAGG | AGTCGGcacy | aAGGATATGG | GATTTGTCTC | TTGACTAAat | 6050 |
| ACCAAagcaa | AcCCAACtct | agGCCAGgtT | TTATTTGCAA | ACAAGAAATA | 6100 |
| AAGGCTTACT | TCTTTAAAAA | Aaaaaaaaaa | aaaaaacttc | cgaaactgaa | 6150 |
| gagcaaaagg | gaaaaaaatg | gaaaaaaaga | agcagaacaa | cccaaagtc  | 6200 |