Figure 1: Cleft Trios — Red: $-\log_{10} p$-values for each CNV component that has at least **five** "0/1" mating pairs. Null hypothesis is transmission rate of $\frac{1}{2}$, and alternative hypothesis is "one-sided." Blue: The frequency of CNV component in the parents. Yellow: Gene tracks, labeled by transcript.

The top CNV component has the followinfg trio-states. Where '1' indicates a deletion and order is F, M, O.

```
000 001 010 011 100 101 110 111
149   7  26  57  28  71   5 102
```

The CNV components with signiifcant p-values (Bonferroni) are given below.

```
GRanges with 16 ranges and 0 metadata columns:
            seqnames                  ranges strand
               <Rle>               <IRanges>  <Rle>
  comp1994      chr8 [39356825, 39370186]       *
  comp1995      chr8 [39370187, 39379683]       *
  comp1996      chr8 [39379684, 39393417]       *
  comp1997      chr8 [39393418, 39401865]       *
  comp1998      chr8 [39401866, 39408899]       *
       ...       ...                  ...      ...
  comp2005      chr8 [39450168, 39457081]       *
```
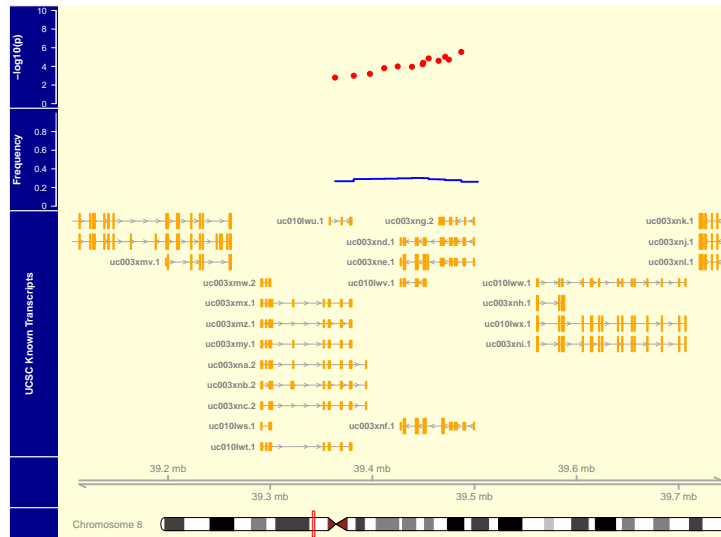
1

Figure 2: Control Trios — Red: $-\log_{10} p$-values for each CNV component that has at least **five** "0/1" mating pairs. Null hypothesis is transmission rate of $\frac{1}{2}$, and alternative hypothesis is "one-sided." Blue: The frequency of CNV component in the parents. Yellow: Gene tracks, labeled by transcript.
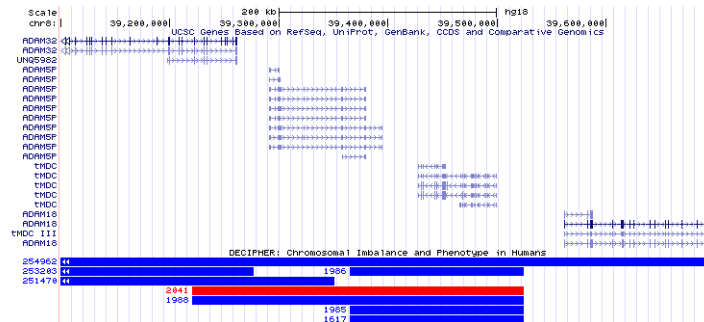

Figure 3: UCSC Genome Browser at chromosome 8 locus.

```
comp2006      chr8 [39457082, 39460087]      *
comp2007      chr8 [39460088, 39469612]      *
comp2008      chr8 [39469613, 39476658]      *
comp2009      chr8 [39476659, 39497557]      *
---
seqlengths:
          chr1   chr1_random          chr2 ...          chrY          chrM
     247249719       1663265     242951149 ...      57772954         16571
```

All CNV components are contiguous and the total width is 140.7 kB.

```
function (object)
{
    if (sum(object[c("101", "011", "100", "010")], na.rm = TRUE) >=
        5) {
        T <- sum(object[c("101", "011")], na.rm = TRUE)
        U <- sum(object[c("100", "010")], na.rm = TRUE)
        return(binom.test(x = T, n = T + U, p = 0.5, alternative = "greater")$p.value)
    }
    else {
        return(NA)
    }
}
<environment: namespace:trioClasses>

> gr.deletion.pitt <- gr.pitt[values(gr.pitt)$numsnp >= 10 &
     values(gr.pitt)$cn %in% 0:1]
> sum(countOverlaps(gr.deletion.pitt, reduce(locus)))

[1] 417
```