

```

> se <- SummarizedExperiment(assays = SimpleList(cnv = t(cnv.obj$cnv.mat)),
  colData = DataFrame(id = rownames(cnv.obj$cnv.mat), row.names = rownames(cnv.obj$cnv.ma
  rowData = cnv.obj$cmp.gr)
> beaty.trios <- MinimumDistance::trios(beaty.pedigree)
> beaty.ped <- DataFrame(famid = do.call("rbind", strsplit(beaty.trios$O,
  "_"))[, 1], id = beaty.trios$O, fid = beaty.trios$F,
  mid = beaty.trios$M, sex = NA, dx = NA)
> ped <- PedClass(beaty.ped)
> fe <- FamilyExperiment(se, pedigree = ped)

```

The top 3 CNV components have the following trio-states. Where '1' indicates a deletion and order is F, M, O.

\$comp1994

```

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

```

\$comp1995

```

000 001 010 011 100 101 110 111
116  5  30  63  38  75  9 109

```

\$comp1996

```

000 001 010 011 100 101 110 111
113  5  30  64  37  74  11 111

```

The CNV components with significant 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]	*
comp2006	chr8	[39457082, 39460087]	*
comp2007	chr8	[39460088, 39469612]	*
comp2008	chr8	[39469613, 39476658]	*

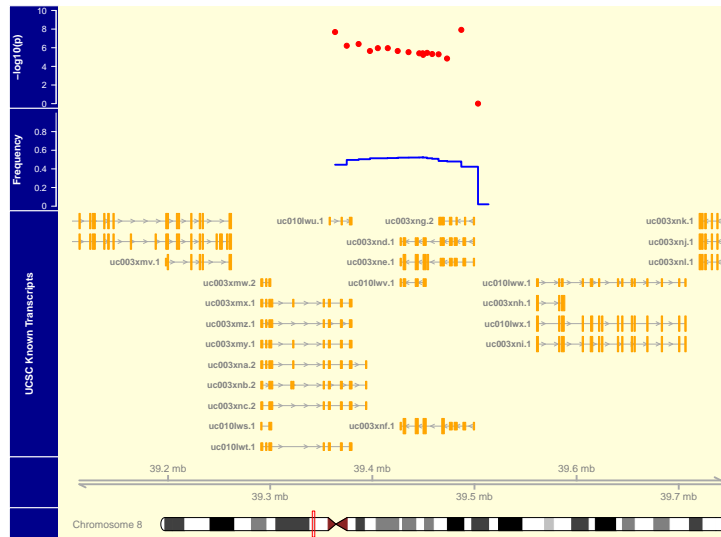


Figure 1: Red: $-\log_{10} p$ -values for each CNV component that has at least **five** “0/1” mating pairs. Null hypothesis is transmission rate of $\frac{1}{2}$, and alternative hypothesis is “one-sided.” Blue: The frequency of CNV component in the parents. Yellow: Gene tracks, labeled by transcript.

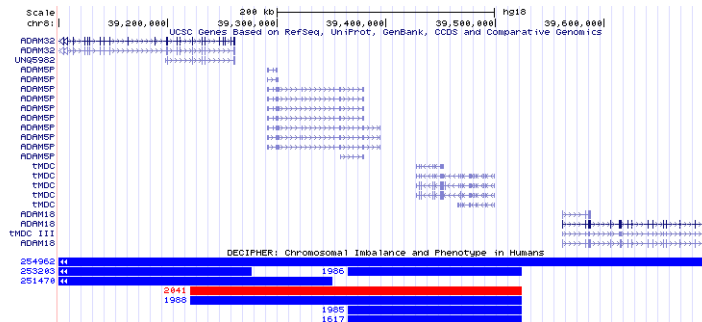


Figure 2: UCSC Genome Browser at chromosome 8 locus.

```

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

```