CNV Mendelian Precision Report
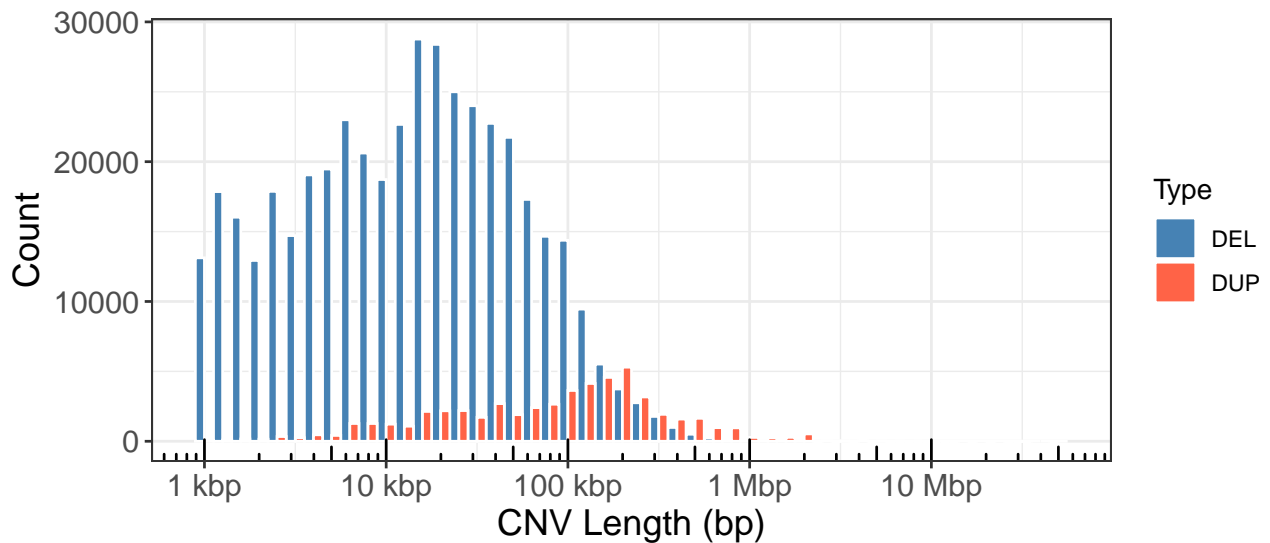
Jacquemont Lab

Dataset: SPARK_PennCNV_raw

Date: 04/08/2025

Number of Trios or Children analyzed: 16421

CNV Length Distribution Across Samples


```
##     Min.  1st Qu.   Median     Mean  3rd Qu.      Max.
##     1000     5026    16553    53641    47163  49397679
```

Distribution of CNVs by Copy Number State

```
##
##       0     0,1        1     1,3        2        3      3,4        4
##    1925       3   436550      25      315    53663        5       96
```

Summary of CNV Counts per Individual

```
##    Min. 1st Qu.  Median    Mean 3rd Qu.     Max.
##       1       8      13      30      35     1183
```

Mendelian precision across CNV length bins: assessing dataset quality (proportion of child CNVs observed in parents with $\geq 50\%$ reciprocal overlap) across multiple metrics.

Note: bins with fewer than 1000 CNVs were excluded from the analysis.