CNV Mendelian Precision Report
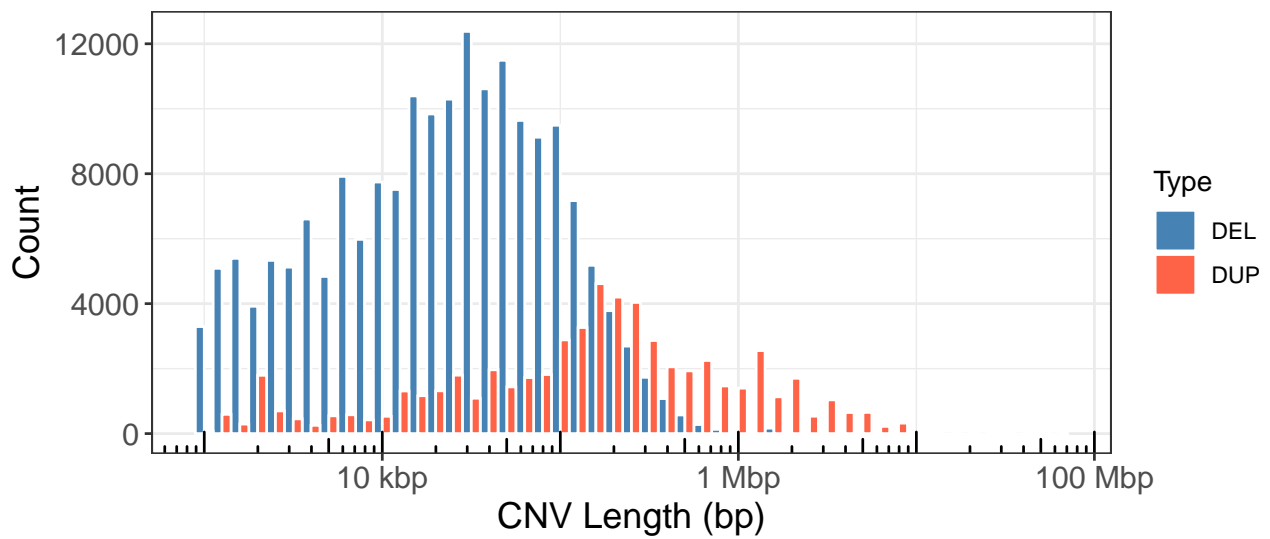
Jacquemont Lab

Dataset: SPARK_Array_GRCh37 CNV merged dataset

Date: 08/09/2025

Number of Trios or Children analyzed: 16000

CNV Length Distribution Across Samples


```
##      Min.  1st Qu.    Median     Mean  3rd Qu.      Max.
##      1000     9341     34269   190186   110586  70336896
```

Distribution of CNVs by Type

```
##
##    DEL    DUP    MIX
## 185101  59779     36
```
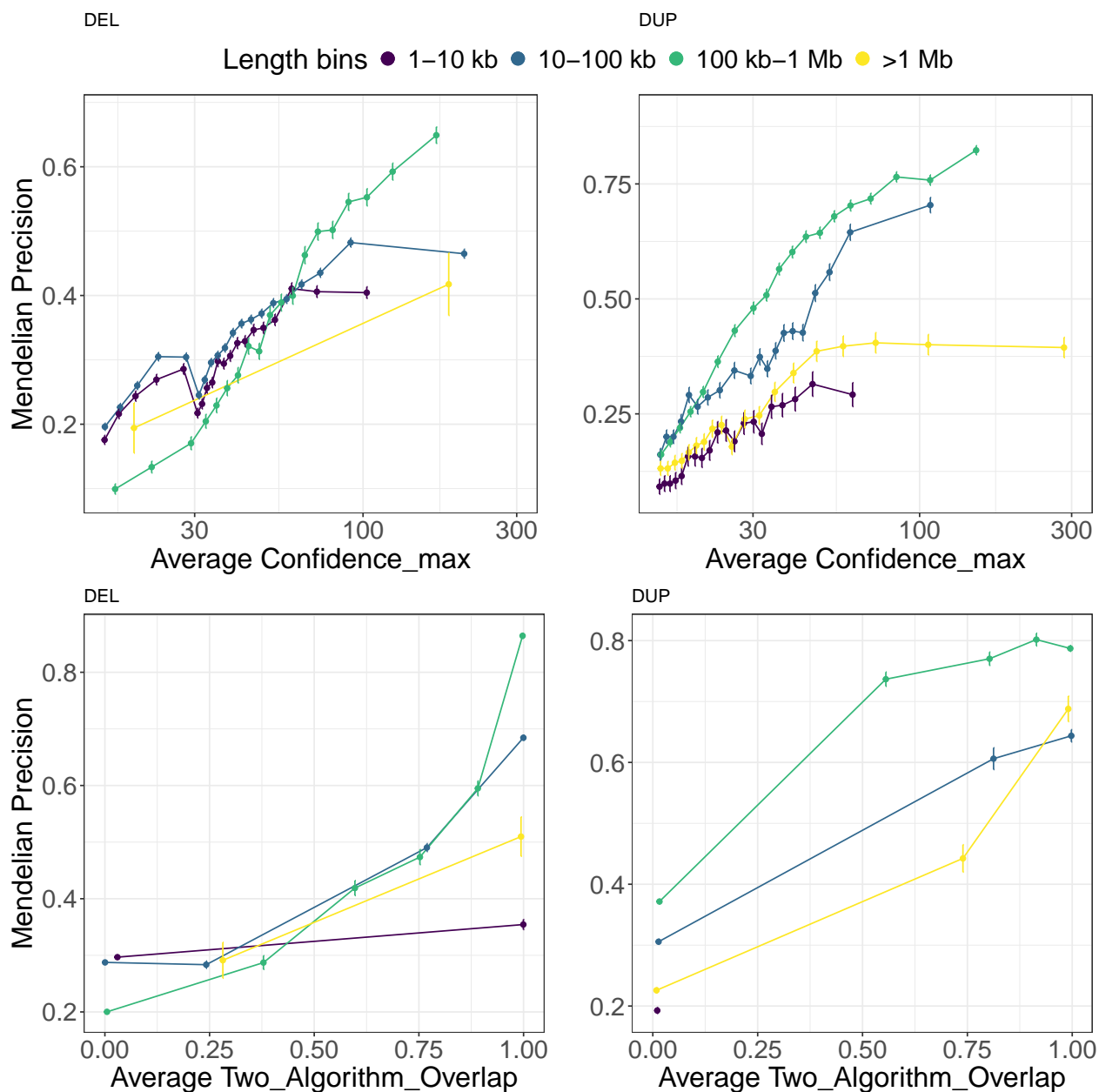
Summary of CNV Counts per Individual

```
##    Min. 1st Qu.  Median    Mean 3rd Qu.    Max.
##    1.00    5.00    8.00   15.31   17.00 1482.00
```

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 100 CNVs were excluded from the analysis.

Mendelian precision across CNV length bins stratified by deletion and duplication types, two algorithm overlap thresholds, and filtering criteria (including segmental duplication exclusion and maximum confidence score), with error estimates and minimum count filtering.