

CNV Mendelian Precision Report

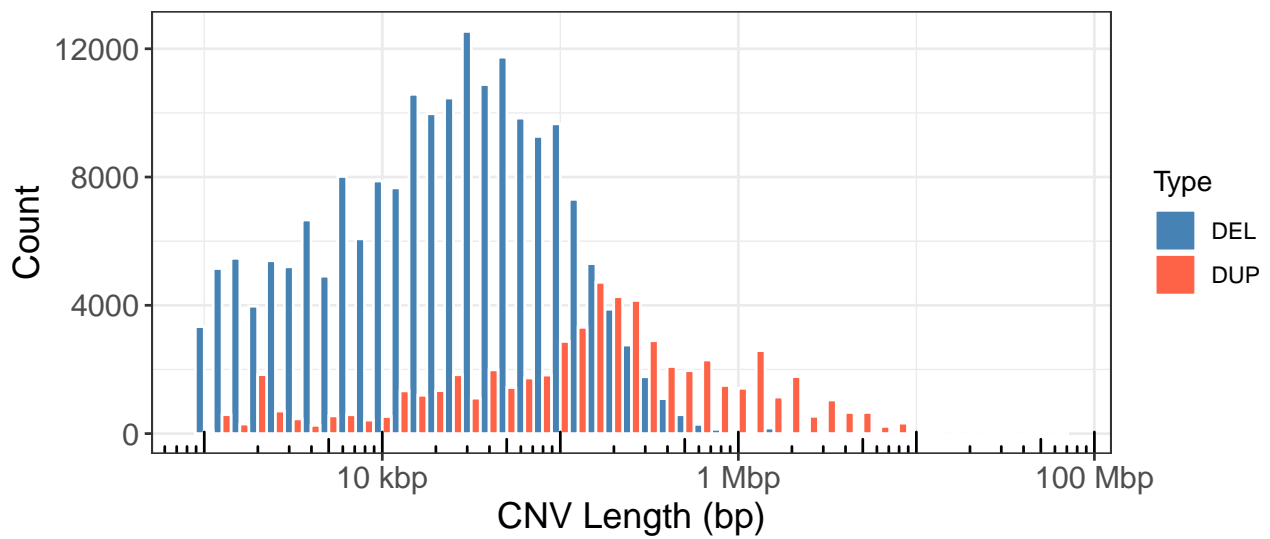
Jacquemont Lab

Dataset: SPARK CNV dataset

Date: 04/08/2025

Number of Trios or Children analyzed: 16001

CNV Length Distribution Across Samples



##	Min.	1st Qu.	Median	Mean	3rd Qu.	Max.
##	1000	9411	34269	190638	110590	70336896

Distribution of CNVs by Copy Number State

##					
##	0	0,0,1	0,1	0,1,1	1
##	1769	1	291	4	186178
##	1,1,3	1,3	1,3,1,4,3,4,4	1,3,3	1,3,4
##	3	61	1	11	2
##	1,4	2	2,3	3	3,3,4
##	14	758	2	51669	21
##	3,4	4			
##	190	8064			

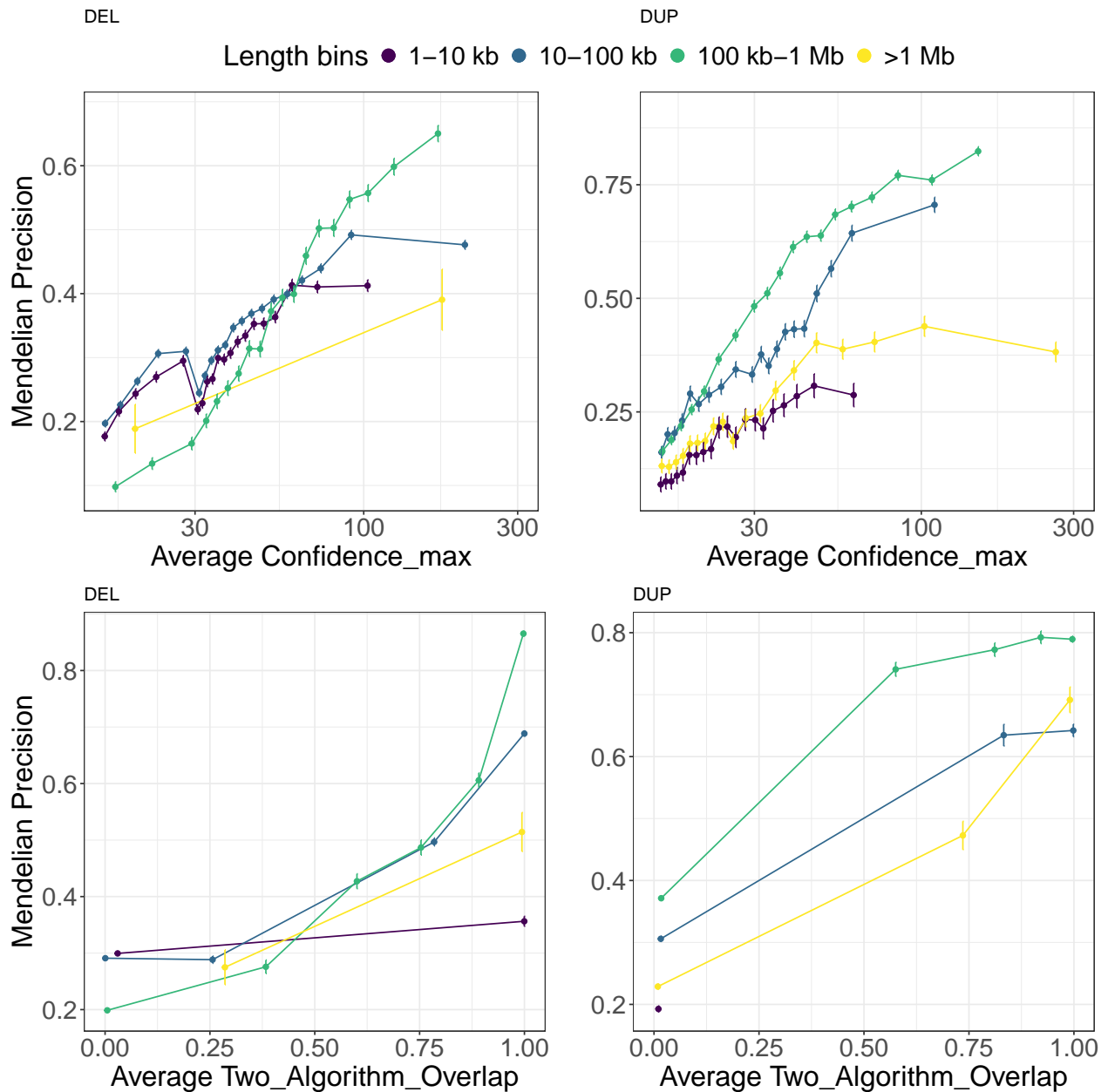
Summary of CNV Counts per Individual

##	Min.	1st Qu.	Median	Mean	3rd Qu.	Max.
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1.00 5.00 8.00 15.56 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 confidence score), with error estimates and minimum count filtering.

