

	Gene panel data	Whole exome data
Gold Standard	comprehensive QMPSF / 4 genes	aCGH data
Number of samples	465	147
Comparison to	GPS-CANOES calls (from panel 1)	WES-CANOES calls
Number of Gold Standard CNVs	14	102†
True positives	14	89
False negatives	0	13
Sensitivity	100% (95%CI:[73.24-100])	87.25% (95%CI:[78.84-82.77])
Number of CANOES calls	14	223†
True positives	14	190
False positives	0	33
Positive Predictive Value	100% (95%CI:[73.24-100])	85.2% (CI:[95%79.70-89.46])

**Table 1. Summary of step 1 evaluation of the CANOES-centered workflow.** Step 1 consisted in the comparison of a gold standard dataset to the results of a CANOES-centered workflow from exome or gene panel sequencing. aCGH: array Comparative Genomic Hybridization. CNV: Copy Number Variation. GPS: Gene Panel Sequencing. QMPSF: Quantitative Multiplex PCR of Short Fluorescent. WES: Whole Exome Sequencing. CI: confidence Interval.

†The difference in the number of CNVs compared from WES comparison is due to the selection process: we selected all CNVs theoretically detectable in regards to the comparison approach.