

	Gene panel data	Whole exome data
Data source	GPS-CANOES calls	WES-CANOES calls
Number of samples	3311	1056
Comparison to	Targeted QPMSF or MLPA	Targeted QPMSF / ddPCR
CANOES calls	101	122
True positives	87	108
False positives	14	14
Positive Predictive Value	86.13% (95% CI:[77.50-91.94])	88.52% (95% CI:[81.18-93.35])

**Table 2. Summary of step 2 evaluation of the CANOES-centered workflow.** Step 2 consisted in the validation by independent targeted techniques of candidate CNVs from gene panel or exome sequencing. aCGH: array Comparative Genomic Hybridization. CNV: Copy Number Variation. GPS: Gene Panel Sequencing. QPMSF: Quantitative Multiplex PCR of Short Fluorescent. WES: Whole Exome Sequencing. CI: confidence Interval.