



Performance PrenatalSafe® KARYO : pre-clinical validation data*

	Trisomy 21 (n=1419)	Trisomy 18 (n=1419)	Trisomy 13 (n=1419)	SCA (n=1419)	CNV (n=1419)
True Positive	100	31	14	36	37
False Positive	0	0	0	0	0
True Negative	1319	1388	1405	1383	1382
False Negative	0	0	0	0	0
Sensitivity (95% CI)	100,00% (96.38% - 100.00%)	100,00% (88.78% - 100.00%)	100,00% (76.84% - 100.00%)	100,00% (90.26% to 100.00%)	100,00% (90.51% to 100.00%)
Specificity (95% CI)	100,00% (99.72% - 100.00%)	100,00% (99.73% - 100.00%)	100,00% (99.74% - 100.00%)	100,00% (99.73% to 100.00%)	100,00% (99.73% to 100.00%)
PPV (95% CI)	100,00% (96.38% - 100.00%)	100,00% (88.78% - 100.00%)	100,00% (76.84% - 100.00%)	100,00% (90.26% to 100.00%)	100,00% (90.51% to 100.00%)
NPV (95% CI)	100,00% (99.72% - 100.00%)	100,00% (99.73% - 100.00%)	100,00% (99.74% - 100.00%)	100,00% (99.73% to 100.00%)	100,00% (99.73% to 100.00%)

* Fiorentino et al., ESHG conference 2016; ISPD Conference 2016

Performance PrenatalSafe® KARYO : Clinical Cases with Follow-up (update March 2016)

	Trisomy 21 (n=11.932)	Trisomy 18 (n=11.932)	Trisomy 13 (n=11.932)	SCA (n=11.932)	Rare Trisomies (n=11.932)	CNV (n=11.932)
True Positive	88	15	12	36	10	8
False Positive	1	1	1	12	7	5
True Negative	11.843	11.916	11.919	11.884	11.915	11.919
False Negative	0	0	0	0	0	0
Sensitivity (95% CI)	100.00% (95.89% - 100.00%)	100.00% (78.20% - 100.00%)	100.00% (73.54% - 100.00%)	100.00% (90.26% - 100.00%)	100.00% (69.15% - 100.00%)	100.00% (63.06% - 100.00%)
Specificity (95% CI)	99.99% (99.95% - 100.00%)	99.99% (99.95% - 100.00%)	99.99% (99.95% - 100.00%)	99.90% (99.82% - 99.95%)	99.94% (99.88% - 99.98%)	99.96% (99.90% - 99.99%)
PPV (95% CI)	98.88% (92.54% - 99.84%)	93.75% (67.88% - 99.07%)	92.31% (62.83% - 98.84%)	75.00% (63.02% - 84.08%)	58.82% (40.52% - 74.97%)	61.54% (39.98% - 79.35%)
NPV (95% CI)	100.00% (99.95% - 100.00%)	100.00% (99.95% - 100.00%)	100.00% (99.95% to 100.00%)	100.00% (99.95% - 100.00%)	100.00% (99.95% - 100.00%)	100.00% (99.95% - 100.00%)

PPV: Positive Predictive Value NPV: Negative Predictive Value SCA: Sex Chromosomes Aneuploidy CNV: Copy Number Variation

