PrenatalSafe® KARYO

Performance PrenatalSafe® KARYO: dati di validazione pre-clinica*

	Trisomia 21 (n=1419)	Trisomia 18 (n=1419)	Trisomia 13 (n=1419)	SCA (n=1419)	CNV (n=1419)
Veri positivi	100	31	14	36	37
Falsi positivi	0	0	0	0	0
Veri negativi	1319	1388	1405	1383	1382
Falsi negativi	0	0	0	0	0
Sensibilità (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%)
Specificità (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: Casistica Clinica con Follow-up (aggiornamento Marzo 2016)

	Trisomia 21 (n=7048)	Trisomia 18 (n=7048)	Trisomia 13 (n=7048)	SCA (n=7048)	Trisomie Rare (n=7048)	CNV (n=7048)
Veri positivi	58	9	6	20	8	6
Falsi positivi	1	0	1	10	4	5
Veri negativi	6989	7039	7041	7018	7036	7037
Falsi negativi	0	0	0	0	0	0
Sensibilità (95% CI)	100.00% (93.84% - 100.00%)	100.00% (66.37% - 100.00%)	100.00% (54.07% - 100.00%)	100.00% (83.16% - 100.00%)	100.00% (63.06% - 100.00%)	100.00% (54.07% - 100.00%)
Specificità (95% CI)	99.99% (99.92% - 100.00%)	100.00% (99.95% - 100.00%)	99.99% (99.92% - 100.00%)	99.86% (99.74% - 99.93%)	99.94% (99.85% - 99.98%)	99.93% (99.83% - 99.98%)
PPV (95% CI)	98.31% (90.91% - 99.96%)	100.00% (66.37% - 100.00%)	85.71% (42.13% - 99.64%)	66.67% (47.19% - 82.71%)	66.67% (34.89% - 90.08%)	54.55% (23.38% - 83.25%)
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: Valore Predittivo Positivo **NPV**: Valore Predittivo Negativo **SCA**: aneuploidie dei cromosomi sessuali **CNV**: Copy Number Variation (alterazioni cromosomiche strutturali)

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