

Table 1. Performance relating to NIPT of common aneuploidies, SCAs and other abnormalities in 71883 pregnancies

Positive cases n= 1011 Total follow-up n=868	Trisomy 21	Trisomy 18	Trisomy 13	SCA	Other abnormalities*	Overall performance
True positives	437	93	37	156	58	781
False positives	3	1	8	17	54	83
True negatives	71392	71775	71828	65598	46577	70872
False negatives	2	0	0	1	1	4
Sensitivity (95% CI)	99.54% (98.36% - 99.94%)	100% (96.11% - 100.00%)	100% (90.51% - 100.00%)	99.36% (96.50% - 99.98%)	98.31% (90.91% - 99.96%)	99.49% (98.70% - 99.86%)
Specificity (95% CI)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	99.99% (99.98% - 100.00%)	99.97% (99.96% - 99.99%)	99.88% (99.98% - 99.99%)	99.88% (99.86% - 99.91%)
PPV (95% CI)	99.32% (97.92% - 99.78%)	98.94% (92.91% - 99.85%)	82.22% (69.82% - 90.24%)	90.17% (85.08% - 93.66%)	51.79% (45.08% - 58.42%)	90.39% (88.36% - 92.11%)
NPV (95% CI)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	99.99% (99.99% - 100.00%)

CI: Confidence Intervals; SCA: Sex chromosome aneuploidies.

Positive cases without follow-up excluded from the count of positives in Table 1 (No.): T21 (49); T18 (14); T13 (10); SCA (36); other abnormalities (34).

*Rare autosomal aneuploidies, segmental abnormalities and microdeletions are included.

Table 2. Performance relating to NIPT of sex chromosome aneuploidies

Sex chromosome aneuploidies	X0	XXX	XXY	XYY
True positives	52	27	51	26
False positives	13	0	3	1
True negatives	65724	65775	65747	65776
False negatives	1	0	0	0
Sensitivity (95% CI)	98.11% (89.93% - 99.95%)	100% (87.23% - 100.00%)	100% (93.02% - 100.00%)	100% (86.77% - 100.00%)
Specificity (95% CI)	99.98% (99.97% - 99.99%)	100% (99.99% - 100.00%)	99.99% (99.99% - 100.00%)	99.99% (99.99% - 100.00%)
PPV (95% CI)	80% (69.88% - 87.34%)	100% (99.99% - 100.00%)	94.44% (84.57% - 98.14%)	96.3% (78.55% - 99.46%)
NPV (95% CI)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)

CI: Confidence intervals; SCA: Sex chromosome aneuploidies. Positive cases without follow-up excluded from the count of positives in Table 2 (No.): X0 (18); XXX (6); XXY (7); XYY (5).

ROMA

Laboratori e Studi Medici
Sede Legale e Laboratorio
di Ricerca e Sviluppo
in Genetica Molecolare
Via Castel Giubileo, 11
00138

Laboratorio Genetica
Medica e Diagnostica
Molecolare
Prelievi e Consulenze
Via Castel Giubileo, 62
00138

MILANO

Laboratorio Genetica
Molecolare e Studi Medici
Via Enrico Cialdini, 16
(Affori Centre)
20161

FIRENZE

Laboratorio e studi medici
Via Cavour, 168r
50121



Table 3. Performance relating to NIPT of rare autosomal aneuploidies, segmental abnormalities and microdeletions

Other abnormalities	RAA	Segmental abnormalities (>7 Mb)	Microdeletions* (segmental abnormalities <7 Mb)
True positives	33	20	5
False positives	36	16	2
True negatives	46630	46681	28743
False negatives	0	0	1
Sensitivity (95%CI)	100% (89.42% - 100.00%)	100% (83.16% - 100.00%)	83.33% (35.88% - 99.58%)
Specificity (95%CI)	99.92% (99.89% - 99.95%)	99.97% (99.96% - 99.99%)	99.99% (99.99% - 100.00%)
PPV (95%CI)	47.83% (39.81% - 55.96%)	55.56% (43.37% - 67.11%)	71.43% (37.40% - 91.27%)
NPV (95%CI)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)	100% (99.99% - 100.00%)

CI: Confidence intervals; RAA: Rare chromosomal abnormalities.

Positive cases without follow-up excluded from the count of positives in Table 3 (No.): RAA (25); Segmental abnormalities >7Mb (7); Microdeletions (2).

*Investigated microdeletion syndromes: DiGeorge syndrome, Cri-du-chat syndrome, Prader-Willi syndrome, Angelman Syndrome, 1p36 deletion syndrome, Wolf-Hirschhorn syndrome, Jacobsen syndrome, Langer-Jeidion syndrome and Smith-Magenis syndrome.

ROMA

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Sede Legale e Laboratorio
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00138

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Medica e Diagnostica
Molecolare**
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00138

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50121

