Performance of VERACITY: Clinical Cases with Follow-up

| | Trisomy 21 (n=10446) | Trisomy 18 (n=10446) | Trisomy 13 (n=10446) | Monosomy X (n=6200) | Trisomy X (n=6200) | 47, XXY (n=6200) | 47, XYY (n=6200) | 48, XXYY (n=6200) |
|---|---|---|---|---|---|---|---|---|
| **True Positive** | 44 | 10 | 5 | 4 | 2 | 4 | 0 | 1 |
| **False Positive** | 0 | 0 | 2 | 3 | 0 | 0 | 0 | 0 |
| **True Negative** | 10402 | 10436 | 10439 | 6193 | 6198 | 6196 | 6200 | 6199 |
| **False Negative** | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| **Sensitivity (95% CI)** | **100% (95% CI, 92–100%)** | **100% (95% CI, 69–100%)** | **100% (95% CI, 48–100%)** | **100% (95% CI, 40–100%)** | - | - | - | - |
| **Specificity (95% CI)** | **99.98% (95% CI, 99.93–99.998%)** | **99.98% (95% CI, 99.93–99.998%)** | **99.98% (95% CI, 99.93–99.998%)** | **99.95% (95% CI, 99.86–99.99%)** | **99.95% (95% CI, 99.86–99.99%)** | **99.95% (95% CI, 99.86–99.99%)** | **99.95% (95% CI, 99.86–99.99%)** | **99.95% (95% CI, 99.86–99.99%)** |
| **PPV** | **100% (95% CI, 92–100%)** | **100% (95% CI, 69–100%)** | **71% (95% CI, 29–96%)** | **57% (95% CI, 18–90%)** | - | - | - | - |
| **NPV** | **100% (95% CI, 99.96–100%)** | **100% (95% CI, 99.96–100%)** | **100% (95% CI, 99.96–100%)** | **100% (95% CI, 99.94–100%)** | **100% (95% CI, 99.94–100%)** | **100% (95% CI, 99.94–100%)** | **100% (95% CI, 99.94–100%)** | **100% (95% CI, 99.94–100%)** |

CI: Confidence Interval; PPV: Positive Predictive Value; NPV: Negative Predictive Value

*The overall clinical performance of Veracity for the detection of trisomies 13, 18, 21 and SCAs during routine NIPT testing conditions was assessed in a cohort of 10564 mixed-risk samples accessioned by our laboratories until February 2018. The median gestational age in this testing cohort was 13 weeks. The median maternal age was 35 years. The median fetal fraction of reported samples was 9.6%. In this cohort twin pregnancy samples represented 3% of all referrals. The median turn-around time for reporting was 5 business days.*

*Follow-up confirmation results were available for 44 samples of trisomy 21, 10 trisomy 18, 7 trisomy 13. Based on the confirmation feedback the PPV for trisomies 21, 18, 13 was estimated at 100% (95% CI, 92–100%), 100% (95% CI, 69–100%) and 71% (95% CI, 29–96%) respectively (Table 3). Follow-up results for sex chromosome aneuploidies was limited. Estimated PPV for Monosomy X was estimated at 57% (95% CI, 18–90%) while the NPV for SCA detection was estimated at 100% (95% CI, 99.94–100%)*

*Kypri et al. Molecular Cytogenetics (2019) 12:34*