Counsyl

THE COUNSYL PRELUDE™ PRENATAL SCREEN

Clarity in results reporting

Guidelines in action

Counsyl is the first non-invasive prenatal screen (NIPS) lab to implement ACOG/SMFM/ACMG's recommendation[1,2,3] to include **individualized PPV (positive predictive value) and residual risk estimates** on our reports.

Why individualized PPV?

Individualized PPV is a more accurate way of translating what a positive result actually means for each patient. It's the chance a screen positive is actually a true positive result for a particular patient — factoring in maternal and gestational age.

Clarifying true risk

Calculating and reporting patient-specific PPV and residual risk on each test result* helps you clarify a patient's true risk and guide next steps for your patient's care.

Prelude™ Prenatal Screen

POSITIVE: PREGNANCY AT INCREASED RISK

ABOUT THIS TEST

The **Counsyl Prelude Prenatal Screen** detects whether a pregnancy is at increased risk for certain chromosome conditions.

PANEL DETAILS

Chromosomes 13, 18, 21 + Sex Chromosome Analysis

RESULTS SUMMARY

Condition	Results	Patient-specific PPV or Residual Risk*
Trisomy 21 (Down Syndrome)	**POSITIVE: PREGNANCY AT INCREASED RISK** **Aneuploidy detected** Results consistent with trisomy for chromosome 21.	**97.94% (97.94 in 100) PPV**
Trisomy 13 (Patau Syndrome)	**NEGATIVE** Results consistent with two copies of chromosome 13.	< 0.01% (1 in 10,000) Residual Risk
Trisomy 18 (Edwards Syndrome)	**NEGATIVE** Results consistent with two copies of chromosome 18.	0.02% (1 in 5,600) Residual Risk

Predicted Fetal Sex: Male

Results consistent with two sex chromosomes (XY).

NEXT STEPS

Genetic counseling is recommended.

CLINICAL NOTES

* The positive predictive value (PPV) represents the risk for the pregnancy to be affected with the indicated chromosome anomaly in view of a positive result. The residual risks provided represent the remaining chance that the pregnancy is affected with the indicated chromosome anomaly in view of a negative result.

This is a screening test; therefore, false positive and false negative results can occur. Clinical correlation with ultrasound findings and history is indicated. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis is necessary.

About individualized PPV

In the case of cell-free DNA screening, individualized positive predictive value (PPV) tells a patient the likelihood that her specific pregnancy may be affected given her prior risk and test results. It is equivalent to the patient-specific risk estimates obtained from traditional maternal serum screening.

Patient-specific PPV depends not only on the accuracy of the test[4], but also on the prevalence of the condition, which is impacted by maternal and gestational age.[5,6]

Example of clarifying true risk

A **25 y/o patient at 16 weeks** with a positive NIPS result for Down syndrome has a **PPV of 70.1%**. This patient would have a 70.1% chance of having an affected pregnancy.

A **42 y/o patient at 12 weeks** with the same positive NIPS result for Down syndrome has a **PPV of 98.5%**. This patient would have a 98.5% chance of having an affected pregnancy.

"Given the importance of these data in providing accurate and understandable information to patients regarding screening test results, ACOG and SMFM encourage all laboratories to report results with PPV and residual risk values[1]..."

— *From the 2015 SMFM/ACOG statement on cell-free DNA screening for fetal aneuploidy*

Trisomy 21 positive predictive value (%)

Maternal age in years

Gestational age in weeks	20	25	30	31	32	33	34	35	36	37	38	39	40	41	42	43	44	45
10	71.7	74.1	81.3	83.3	85.5	87.7	89.7	91.6	93.3	94.7	95.9	96.9	97.6	98.2	98.7	99.0	99.2	99.4
12	70.0	72.5	80.0	82.1	84.4	86.7	88.9	91.0	92.7	94.3	95.6	96.6	97.4	98.0	98.5	98.9	99.2	99.4
14	68.6	71.2	78.9	81.1	83.5	85.9	88.2	90.4	92.3	93.9	95.3	96.4	97.2	97.9	98.4	98.8	99.1	99.4
16	67.5	70.1	78.0	80.4	82.8	85.3	87.7	89.9	91.9	93.6	95.0	96.2	97.1	97.8	98.3	98.8	99.1	99.3
20	65.8	68.5	76.7	79.1	81.7	84.3	86.9	89.2	91.3	93.1	94.6	95.9	96.9	97.6	98.2	98.7	99.0	99.3
40	62.0	64.8	73.6	76.3	79.1	82.0	84.9	87.5	89.9	92.0	93.8	95.2	96.3	97.2	97.9	98.4	98.8	99.1

Individualized PPV and residual risk estimates* provide a more accurate picture of a patient's chance to have an affected pregnancy. This is why they are recommended by ACOG/SMFM/ACMG in results reporting[1,2,3] — and why we include them on our reports.

For more information visit **counsyl.com/provider/prelude-prenatal-screen**

* Negative screen results receive a residual risk estimate, which is the chance a pregnancy could be affected given a negative test result.

1. Cell-free DNA screening for fetal aneuploidy. Committee Opinion No. 640. ACOG. Obstet Gynecol 2015;126:e31–7. **2**. Society for Maternal-Fetal Medicine (SMFM) Publications Committee. #36 Prenatal aneuploidy screening using cell-free DNA. AJOG 2015, 212(6):711-716. **3**. 2016 ACMG position statement on NIPS for fetal aneuploidy, Genetics in Medicine, doi:10.1038/gim.2016.97 **4.** Gil MM, et al. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. Ultrasound Obstet Gynecol 2017;50:302-14. **5**. Snijders RJM, et al. Maternal age and gestation-specific risk for trisomy 21. Ultrasound Obstet Gynecol 1999;13:167-70. **6**. Snijders RJM, et al. Maternal age and gestation age-specific risks for chromosomal defects. Fetal Diag Ther 1995;10:356-67.

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