UNITY

1035 O'Brien Drive
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POWERED BY Billion To One

PATIENT		SAMPLE		PROVIDER	
First Name	Jane	**Sample Type**	BLOOD	**Name**	Dr. Jane Smith
Last Name	Doe	**Date Collected**	05/03/2021	**Address**	1234 Street Name
DOB	01/20/1986	**Date Received**	05/04/2021		San Francisco, CA 94102
Ethnicity		**Accession ID**	123-45-6789	**Phone**	555-555-5555
Gender	Female	**Requisition ID**	987-65-4321	**Fax**	555-555-5555
Gestational Age	14w2d	**Date Reported**	05/11/2021		
Medical Record #	321-15-5321				

UNITY™ Complete: **Aneuploidy Screen**

Twin Gestation

SUMMARY OF RESULTS

H **HIGH RISK** Trisomy 21

♀ **FEMALE** FETAL SEX

♀ **FEMALE** FETAL SEX

9.9% FETAL FRACTION

CONDITIONS SCREENED	FETAL RISK *by* NIPT	RISK *Before* NIPT	RISK *After* NIPT
Trisomy 21	**HIGH RISK**	**1 in 91**	**9 in 10**
Trisomy 18	**Low Risk**	**1 in 288**	**<1 in 10,000**
Trisomy 13	**Low Risk**	**1 in 804**	**<1 in 10,000**

RECOMMENDED FOLLOW-UP

PRENATAL DIAGNOSIS via chorionic villus sampling or amniocentesis is **RECOMMENDED**.

GENETIC COUNSELING is recommended to review the implications of this result.

The patient may contact BillionToOne at (650) 460-2551 to schedule an appointment for a complimentary telephone genetic consultation to review these results. A genetic counselor can also be found at www.nsgc.org.

Interpretation *next page* ›

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Patient Name Jane Doe	**DOB** 01/20/1986	**Gestational Age** 14w2d	**Medical Record #** 321-15-5321

INTERPRETATION

UNITY™ Complete: **Aneuploidy Screen**
Twin Gestation

The pregnancy is HIGH RISK for one or both fetuses to be affected with TRISOMY 21. The estimated fetal fraction was 9.9%.

NIPT was performed to evaluate for fetal copy number of chromosomes 13, 18, & 21.

This NIPT result is valid only for a twin gestation achieved without egg donation or gestational carrier.

Prenatal diagnosis via chorionic villus sampling or amniocentesis is recommended. UNITY™ NIPT is not diagnostic. No irreversible decisions regarding the pregnancy should be made without confirmatory invasive prenatal testing. Genetic testing can also be performed postnatally.

Genetic counseling is recommended for this patient to review the implications of this result. The patient may contact BillionToOne at (650) 460-2551 to schedule an appointment for a complimentary telephone genetic consultation to review these results. Comprehensive genetic counseling is recommended for a patient with a family history of a chromosome abnormality or other genetic disorder so that risks can be accurately discussed, as well as additional testing options that may be available. A genetic counselor can be found at www.nsgc.org.

METHODS AND LIMITATIONS

UNITY™ Complete: **Aneuploidy Screen**

Cell-free DNA (cfDNA) was isolated from 2-4 mL of plasma from whole blood collected in a Streck cell-free DNA tube. A paternal inheritance NIPT was performed as a multiplex PCR on common single nucleotide variants (SNVs) to measure the fraction of cell-free DNA of fetal origin. A modified fetal fraction calculation is performed for twin pregnancies and pregnancies conceived with a gestational carrier. The fetal risk for aneuploidy of chromosomes 13, 18, 21, X, & Y for singleton pregnancies or chromosomes 13, 18, & 21 for twin pregnancies, was determined by a separate multiplex PCR on cfDNA using no more than 99 amplicons per chromosome to perform relative chromosomal dosage analysis. When multiple blood tubes are analyzed for NIPT (e.g. for redraws), we report the combined reported fetal fraction by taking the arithmetic mean of fetal fractions across different tubes. Due to the tube-to-tube assay variability, the reported fetal fraction for the same patient can differ between single-gene NIPT and aneuploidy NIPT. Post-test residual risks ('Risk After NIPT') for high-risk results are calculated based on the test sensitivity, test specificity, and prior risks as determined by maternal age and are therefore personalized. Post-test residual risks for low-risk results are calculated based on the test sensitivity, test specificity, and prior risk in the general population, and are therefore not personalized.

Test Limitations: Results may not be reported when the amount of cell-free fetal DNA in the blood sample is too low. Results from this test are highly accurate; however, discordant results may occur. Potential causes of discordant results include: maternal, fetal, or placental mosaicism, low fetal fraction, vanishing twin, maternal malignancy, maternal organ or bone marrow transplant, or other reasons. Findings of unknown significance will not be reported. UNITY™ does not screen for neural tube defects or abdominal wall defects. This test does not screen for microdeletions. UNITY™ NIPT is not diagnostic. No irreversible decisions regarding the pregnancy should be made without confirmatory invasive prenatal testing.

CONDITIONS SCREENED	SENSITIVITY	SPECIFICITY	POSITIVE PREDICTIVE VALUE	NEGATIVE PREDICTIVE VALUE
Trisomy 21	99.8% [98.9% - 100%]	99.9% [99.8% - 99.9%]	84.2%	> 99.9%
Trisomy 18	99.9% [99.0% - 100%]	> 99.9% [99.9% - 100%]	94.2%	> 99.9%
Trisomy 13	99.1% [97.4% - 100%]	> 99.9% [99.9% - 100%]	52.5%	> 99.9%
Monosomy X	97.7% [93.7% - 100%]	> 99.9% [99.9% - 100%]	85.5%	> 99.9%
Sex Chromosome Aneuploidy (XXX / XXY / XYY)	Reported when identified	> 99.9%	n/a	> 99.9%
Presence of Y Chromosome	> 99.9%	> 99.9%	n/a	> 99.9%

** Internal analytical validation data for singleton pregnancies. Performance metrics are estimated based on analytical data from contrived aneuploidy samples at 9% fetal fraction. Representative coefficients of variance from human plasma samples were applied to this analytical data to perform Monte Carlo simulations to calculate these metrics, assuming prior risks for a patient population of advanced maternal age. Mosaicism is not reflected in the validation.*

This NIPT was developed and its performance characteristics determined by the BillionToOne laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration. The BillionToOne laboratory is regulated under CLIA. This test is used for clinical purposes. It should not be regarded as investigational or for research.

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UNITY™ ANEUPLOIDY SCREEN – HIGH RISK TRISOMY 21

UNITY™ ANEUPLOIDY SCREEN

Non-invasive prenatal testing (NIPT) through UNITY™ screens for specific chromosome changes called aneuploidy. The average person has 23 pairs of chromosomes, or 46 chromosomes total, with one of each chromosome inherited from each parent. A person with aneuploidy has an extra or missing copy of a chromosome, which may cause them to have health problems, physical birth defects, and/or developmental delays. The type and severity of symptoms vary depending on which chromosome is extra or missing. Any woman can have a baby born with a chromosome change. Most aneuploidy is not inherited.

UNITY™ screens for some of the most common aneuploidies present at birth

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)

YOUR PREGNANCY'S RISK: HIGH RISK FOR TRISOMY 21

The testing performed by UNITY™ shows a **significantly increased** chance for one or both of your babies to have trisomy 21. Please reference your report to review the risk for your pregnancy. **No irreversible pregnancy decisions, such as pregnancy termination, should be considered based on UNITY™ results alone.**

Follow-up testing and genetic counseling are recommended. You may contact BillionToOne at (650) 460-2551 to schedule an appointment for a complimentary telephone genetic consultation to review these results. A local genetic counselor can also be found at www.nsgc.org

WHAT IS TRISOMY 21?

People with trisomy 21 (Down syndrome) have three copies of chromosome 21. Children with Down syndrome typically look like their parents, but also have some unique features such as a flat facial profile, small nose, and upward slanted eyes. In addition, they are at risk for various health problems, including a 50% chance of having a heart defect. Children with Down syndrome may also have problems with their vision, hearing, or digestion. Children with Down syndrome usually learn to walk and talk but at a slower rate than their siblings or peers. Almost all individuals with Down syndrome have some degree of intellectual disability.

Pregnancies with trisomy 21 may have physical birth defects or other differences identified on first or second trimester ultrasound. These pregnancies may have a higher risk for miscarriage or stillbirth.

Many health problems are treatable, but some may require surgery after birth. The average life expectancy of a person with Down syndrome is approximately 60 years. Most adults require support; however, some adults may work or live on their own in a supervised setting. The type and severity of symptoms vary from one person to another. Genetic testing cannot predict the type or severity of symptoms.

WHAT CAUSES TRISOMY 21?

People with trisomy 21 have an extra copy of chromosome 21. Trisomy 21 is not usually inherited. This is typically a chromosome change that happens randomly at conception. There is nothing you could have done to cause or prevent trisomy 21.

CONFIRMATORY TESTING

UNITY™ uses an advanced technology to determine whether your pregnancy is at high or low risk of having trisomy 21. Although UNITY™ is a highly accurate screening test, diagnostic testing is recommended to determine whether your babies have trisomy 21.

Before Birth

Testing during pregnancy can be performed by obtaining a sample of placenta (chorionic villus sampling or CVS) or fluid around your babies (amniocentesis) which both contain DNA that is representative of your babies. A CVS is ideally performed between 10 to 13 weeks pregnancy while an amniocentesis is typically performed between 15 to 24 weeks pregnancy. Both procedures are generally safe, but have a small risk for pregnancy complications, including miscarriage.

After Birth

Infants with trisomy 21 typically have physical features of the condition. However, if confirmatory testing during pregnancy is not done, your children's pediatrician should be informed of this result. Chromosome analysis can be performed after birth. Physical exam by a medical geneticist can also be considered.

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UNITY™ ANEUPLOIDY SCREEN – HIGH RISK TRISOMY 21

RESOURCES

- National Down Syndrome Society - https://www.ndss.org/
- Down Syndrome Pregnancy - https://www.downsyndromepregnancy.org
- Global Down Syndrome Foundation - https://www.globaldownsyndrome.org/
- Lettercase, Understanding Down Syndrome - https://understandingdownsyndrome.org/
- National Society of Genetic Counselors - https://www.nsgc.org
- American College of Obstetricians and Gynecologists Guide to Prenatal Diagnosis - https://www.acog.org/Patients/FAQs/Prenatal-Genetic-Diagnostic-Tests