

Patient		Sample		Provider	
Name	N/A	Sample Type	N/A	Provider	N/A
Date of Birth	N/A	Date Collected	N/A	Clinic Address	N/A
Sex Assigned at Birth	N/A	Date Received	N/A	Phone Number	N/A
Gestational Age	N/A	Requisition ID	N/A	Fax Number	N/A
Medical Record #	N/A	Date Reported	N/A		

SUMMARY OF RESULTS



LOW RISK FETUS



Male fetal sex

9.9% fetal fraction

UNITY Aneuploidy™ NIPT

Singleton Gestation

ANEUPLOIDIES SCREENED	FETAL RISK <i>by</i> NIPT	RISK <i>Before</i> NIPT	RISK <i>After</i> NIPT
Trisomy 21	Low Risk	1 in 415	<1 in 10,000
Trisomy 18	Low Risk	1 in 1306	<1 in 10,000
Trisomy 13	Low Risk	1 in 3646	<1 in 10,000
Monosomy X	Low Risk	1 in 250	<1 in 10,000
Sex Chromosome Aneuploidy (XXX / XXY / XYY)	Not Detected		

UNITY 22q11.2 Microdeletion™ NIPT

Singleton Gestation

MICRODELETIONS SCREENED	FETAL RISK <i>by</i> NIPT	RISK <i>Before</i> NIPT	RISK <i>After</i> NIPT
22q11.2 Microdeletion	Low Risk	1 in 2000	<1 in 10,000

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Patient Name **N/A**

DOB **N/A**

Gestational Age **N/A**

Medical Record # **N/A**

Interpretation

UNITY Aneuploidy™ NIPT Singleton Gestation

The fetus is LOW RISK to be affected with aneuploidy of chromosomes 13, 18, 21, X & Y.

A low risk NIPT result significantly reduces the risk of the screened aneuploidies; it does not eliminate the risk. This result does not guarantee a normal pregnancy outcome.

UNITY 22q11.2 Microdeletion™ NIPT Singleton Gestation

The fetus is LOW RISK to be affected with 22q11.2 deletion syndrome.

A low risk NIPT result significantly reduces the risk of a fetal 22q11.2 microdeletion; it does not eliminate the risk.

Genetic counseling is available 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. NIPT does not exclude abnormalities of other chromosomes and microdeletions not evaluated by the screen, triploidy, other genetic syndromes, or birth defects.

Patient Name **N/A**

DOB **N/A**

Gestational Age **N/A**

Medical Record # **N/A**

Interpretation

UNITY Aneuploidy™ NIPT Singleton Gestation

The fetus is LOW RISK to be affected with aneuploidy of chromosomes 13, 18, 21, X & Y.

A low risk NIPT result significantly reduces the risk of the screened aneuploidies; it does not eliminate the risk. This result does not guarantee a normal pregnancy outcome.

UNITY Fetal RhD™ NIPT Singleton Gestation

The *RHD* gene was NOT DETECTED in the cell-free DNA (RhD negative).

NIPT was performed to determine the presence or absence of the *RHD* gene. A functioning *RHD* gene was NOT DETECTED in the cell-free DNA (RhD negative). This result indicates fetal RhD negative blood type.

Anti-D prophylaxis is not indicated for RhD negative patients carrying an RhD negative fetus.

Genetic counseling is available 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. NIPT does not exclude abnormalities of other chromosomes and microdeletions not evaluated by the screen, triploidy, other genetic syndromes, or birth defects.

PATIENT		SAMPLE		PROVIDER	
First Name	Jane	Sample Type	Blood	Name	Dr. Jane Smith
Last Name	Doe	Date Collected	05/01/2021	Address 1	1234 Street Name
DOB	10/20/1990	Date Received	05/02/2021	Address 2	Suite 120
Gender	Female	Sample ID	123-123-123	City	San Francisco
Ethnicity	Caucasian	Requisition ID	11223344	State Zip	CA, 94102
Gestational Age	12W	Date Reported	05/16/2021	Phone	555-555-5555
Medical Record #	12344321			Fax	555-555-5555

UNITY™ Five Gene Carrier Screen with Reflex NIPT



POSITIVE CARRIER



HIGH RISK FETUS

CONDITIONS SCREENED	MATERNAL CARRIER STATUS	FETAL RISK BY NIPT
Alpha-Thalassemia (HBA1, HBA2)	Negative	
Sickle Cell Disease / Beta-Thalassemia / Hemoglobinopathies (HBB)	Negative	
Cystic Fibrosis (CFTR)	POSITIVE c.1521_1523delCTT (p.Phe508delPhe)	HIGH RISK <i>See results below</i> ▼
Spinal Muscular Atrophy (SMN1)	Negative 2 SMN1 copies, SNP not present	

NIPT RESULT DETAILS

CONDITIONS SCREENED	FETAL RISK	Risk Before NIPT	Risk After NIPT	Fetal Fraction
Cystic Fibrosis	HIGH	1 in 96 – 1 in 376	9 in 10	6.2%
		Fetal Risk Before NIPT is dependent on paternal ethnicity and assumes paternal carrier status is unknown. See disease carrier frequencies based on ethnicity on the last page of the report.		

Recommended Follow-Up *next page* ➤

The ACOG Committee on Genetics (co486 and co691) recommends cystic fibrosis, hemoglobinopathy, and spinal muscular atrophy carrier screening for all patients who are planning a pregnancy or seeking prenatal care. UNITY™ carrier screening evaluates for cystic fibrosis (CFTR), hemoglobinopathies (HBB, HBA1 and HBA2), and spinal muscular atrophy (SMN1). Reflex NIPT is performed to evaluate fetal risk when a pregnant patient is identified as a carrier.

Patient Name Jane Doe	DOB 10/20/1990	Gestational Age 12W	Medical Record # 12341234
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RECOMMENDED FOLLOW-UP



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



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. A genetic counselor can also be found at www.nsgc.org.



CARRIER SCREENING for cystic fibrosis for the patient's reproductive partner is recommended prior to a future pregnancy.

Interpretation *next page*

Patient Name Jane Doe	DOB 10/20/1990	Gestational Age 12W	Medical Record # 12341234
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INTERPRETATION

UNITY™ Five Gene Carrier Screen

This patient has the c.1521_1523delCTT (p.Phe508delPhe) pathogenic variant in the *CFTR* gene (NM_000492.3) and is a CARRIER for cystic fibrosis.

If this patient's reproductive partner is a carrier for cystic fibrosis, there is a 25% risk for an affected child with each pregnancy. Carrier screening for cystic fibrosis for the patient's reproductive partner is recommended prior to a future pregnancy to clarify the risks for an affected child.

This patient's first-degree relatives each have a 50% chance to be a carrier for cystic fibrosis as well. We recommend these results be shared with blood relatives, especially those of reproductive age.

UNITY™ NIPT for Cystic Fibrosis

The fetus is HIGH RISK to be affected with cystic fibrosis. The estimated fetal fraction was 6.2%.

NIPT was performed to evaluate for fetal *CFTR* variants and concluded the fetus is high risk to be homozygous for the c.1521_1523delCTT (p.Phe508delPhe) pathogenic variant in the *CFTR* gene. Therefore, the fetus is HIGH RISK to be affected with cystic fibrosis.

This NIPT result is valid only for a singleton pregnancy 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. A genetic counselor can also be found at www.nsgc.org.

Patient Name Jane Doe	DOB 10/20/1990	Gestational Age 12W	Medical Record # 12341234
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INTERPRETATION

UNITY™ Five Gene Carrier Screen

No other reportable gene variants were found.

Alpha-Thalassemia <i>HBA1</i> (NM_000558.5), <i>HBA2</i> (NM_000517.6)	Negative
Sickle Cell Disease/Beta-Thalassemia/Hemoglobinopathy <i>HBB</i> (NM_000518.5)	Negative
Spinal Muscular Atrophy <i>SMN1</i> (NM_000344.3) <ul style="list-style-type: none"> • <i>SMN1</i> Copy Number • SMA Region Informative SNP (rs143838139) 	Negative <ul style="list-style-type: none"> • 2 copies (most common) • Not present (most common haplotype)

Carrier frequencies both before and after screening vary by ethnicity and assume no personal or family history of the condition. See Pre- and Post-Test Carrier Frequencies tables on the last page of the report.

Comprehensive genetic counseling is recommended for a patient with a family history of a genetic disorder so that carrier risks can be accurately discussed, as well as potential reproductive risks and additional testing options that may be available.

Carrier screening does not evaluate for all genetic conditions. In addition, carrier screening is not able to identify all possible variants in the genes analyzed. As a result, a negative result significantly reduces the probability of being a carrier; it does not eliminate the risk.