

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)	<b>POSITIVE</b> c.1521_1523delCTT (p.Phe508delPhe)	<b>HIGH RISK</b> <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	<b>HIGH</b>	<b>1 in 96 – 1 in 376</b>	<b>9 in 10</b>	<b>6.2%</b>
		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.

<b>Patient Name</b> Jane Doe	<b>DOB</b> 10/20/1990	<b>Gestational Age</b> 12W	<b>Medical Record #</b> 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](http://www.nsgc.org).



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

Interpretation *next page* 

<b>Patient Name</b> Jane Doe	<b>DOB</b> 10/20/1990	<b>Gestational Age</b> 12W	<b>Medical Record #</b> 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](http://www.nsgc.org).

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

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.