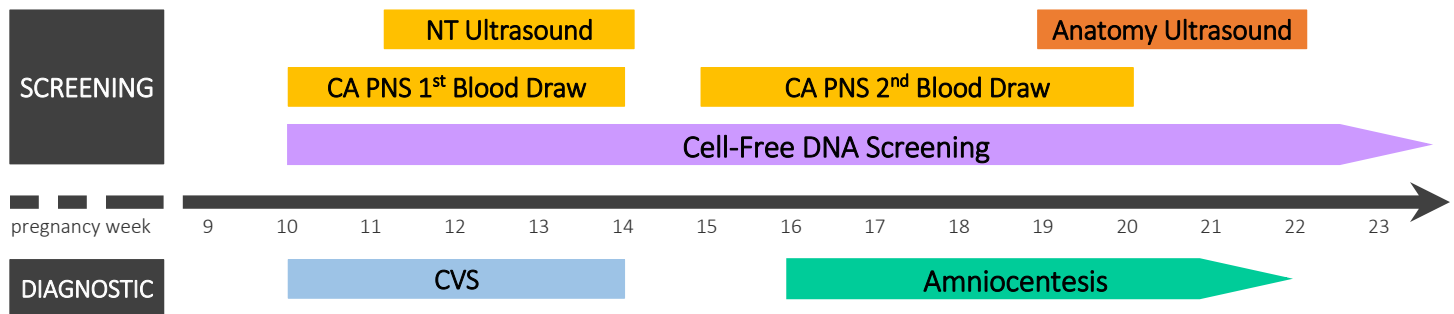


PRENATAL TESTING OPTIONS

There are many types of tests available during pregnancy that can provide more information. No test or combination of tests can detect every possible condition. Patients may elect or decline any or all of these tests as they are all optional.



SCREENING TESTS

Screening tests provide probabilities (odds) of having a baby with certain conditions, however screening tests cannot detect all cases of these conditions. Screening tests are performed through blood draw and ultrasound, so there is no increased risk for miscarriage. Patients will receive either “screen negative” or “screen positive” results from their screening tests. When results are screen negative, these are considered low risk for that condition, however it is not impossible that the baby has that condition. When results are screen positive, these are considered high risk for that condition, however these are not definitive and the baby may not have that condition. Follow up testing is available to confirm screening results. If electing screening blood tests, patients may elect to undergo one or both to screen for conditions in different ways.

■ California Prenatal Screening Program (CA PNS) (also called Sequential Integrated Screening)

- Combines blood draw(s) and the NT ultrasound with a person’s age-related risk to screen for Down syndrome, trisomy 18, open neural tube defects (example: spina bifida), and Smith-Lemli-Opitz syndrome
- The blood draw looks for natural chemicals made by the pregnancy which may show different patterns or levels if a baby has certain conditions; these patterns are used as part of the calculation of results
- The nuchal translucency (NT) ultrasound measures the fluid in the back of the baby’s neck, which may be larger if a baby has certain conditions; this measurement is used as part of the calculation of results
- A preliminary risk assessment result is available after the 1st blood draw plus the NT ultrasound
- A final, more accurate, result is available after the 2nd blood draw is combined with the preliminary results
- Other chromosome anomalies beyond Down syndrome and trisomy 18 can impact the blood chemical patterns or NT measurement, therefore these results can indicate a higher or lower risk for chromosome conditions in general

■ Cell-free DNA Screening (also called Non-invasive Prenatal Screening/Testing or NIPS/NIPT)

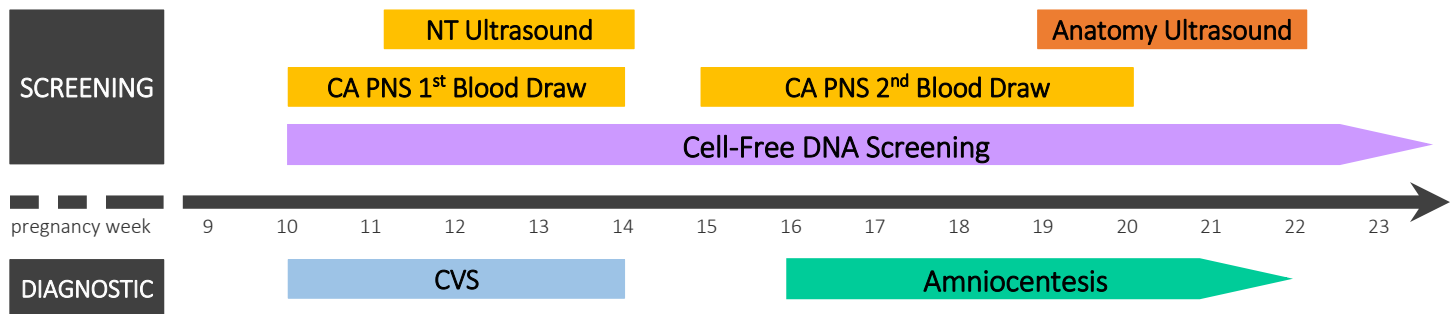
- A blood test which measures the amount of small pieces of DNA that have come from chromosomes 13, 18, 21, X and Y from both the pregnant person and the placenta
- Screens only for Down syndrome, trisomy 18, trisomy 13, and missing or extra sex chromosomes
- May not be covered by insurance for low-risk pregnancies
 - PAMF orders this screening through a company called Myriad, which is in-network with most insurance providers. Myriad can perform an insurance check to provide an estimate of the expected out of pocket cost. Patients can call Myriad directly to request this estimate at (888) 268-6795.

■ Anatomy Ultrasound (also called Complete Ultrasound, Detailed Ultrasound, or Level 2)

- Screens for major structural birth defects, including open neural tube defects
- Can increase suspicion for conditions like Down syndrome however cannot provide a definite yes or no

PRENATAL TESTING OPTIONS

There are many types of tests available during pregnancy that can provide more information. No test or combination of tests can detect every possible condition. Patients may elect or decline any or all of these tests as they are all optional.



DIAGNOSTIC TESTS

Diagnostic tests provide a definitive yes or no answer as to whether the baby has a known condition and are the most accurate prenatal testing for chromosome disorders. They can test for types of conditions that would be missed by screening tests. Procedure related pregnancy loss is a rare occurrence at 1 in 1000 (0.1%). If electing diagnostic testing, patients would typically undergo just one diagnostic procedure, not both.

■ CVS (chorionic villus sampling)

- Results obtained by analyzing a small sample of the placenta
- Performed either by using a catheter passing through the cervix, or by inserting a needle through the abdomen
- 1-2% risk for ambiguous (mosaic) result (amniocentesis is recommended if this is found)

■ Amniocentesis

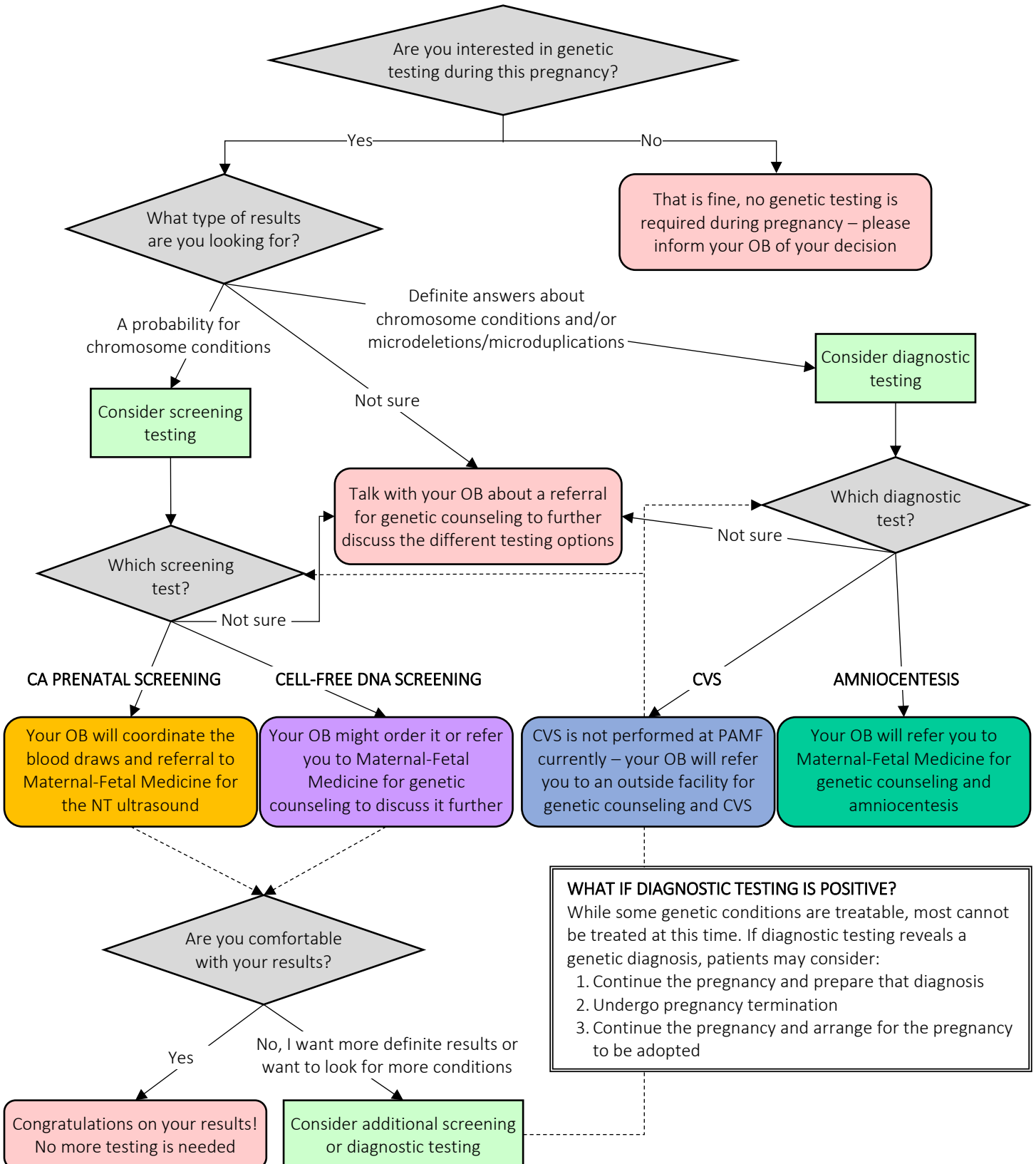
- Results obtained by analyzing a small sample of the amniotic fluid that surrounds the baby
- Performed by inserting a needle through the abdomen

There are different options for which tests to perform on the diagnostic testing sample: A fetal **karyotype** looks for chromosome conditions and looks for large structural rearrangements of the chromosomes. Examples of chromosome conditions include Down syndrome, trisomy 18, trisomy 13, Turner syndrome, and Klinefelter syndrome. A fetal **chromosomal microarray** looks for chromosome conditions and also looks for small missing or extra pieces of DNA within a chromosome, called microdeletions or microduplications. Symptoms of microdeletions and microduplications are variable but often cause significant birth defects, lifelong physical issues, intellectual disabilities, developmental delays or limitations, and/or shortened lifespan. **Other testing** can also be performed on the diagnostic testing sample as indicated (for example: testing for a specific genetic condition when both parents are known carriers for that condition).

CONDITION	CHANCE FOR CONDITION		CHANCE TO <u>NOT</u> HAVE CONDITION	
Down syndrome (chance changes with age)	AGE 25 0.10%	AGE 35 0.34%	AGE 25 99.90%	AGE 35 99.66%
	AGE 30 0.14%	AGE 40 1.15%	AGE 30 99.86%	AGE 40 98.85%
Trisomy 18 (chance changes with age)	AGE 25 0.02%	AGE 35 0.09%	AGE 25 99.98%	AGE 35 99.91%
	AGE 30 0.04%	AGE 40 0.30%	AGE 30 99.96%	AGE 40 99.70%
Microdeletions or microduplications	1-2%		98-99%	
Open neural tube defects (example: spina bifida)	0.1%		99.9%	
Overall chance for a birth defect or genetic condition	3-4%		96-97%	

PRENATAL TESTING OPTIONS

Use this guide to navigate through different possible pathways for prenatal genetic testing. Please reference the other two pages titled “prenatal testing options” for descriptions of the different tests and to review the chances for different conditions detectable by the testing. Patients may elect or decline any or all of these tests as they are all optional.



BILLING CODES AND COST ESTIMATES

Please find the billing (CPT) codes for the different tests covered in this handout, as well as common billing reasons for testing (ICD-10 codes) and ways to obtain a cost estimate for various tests. Please note that patients are responsible for confirming their insurance coverage for tests and for any charges not covered by their insurance.

COMMON BILLING CODES

Test/Procedure	CPT Code(s)
Cell-free DNA screening	81420, 81507
Expanded carrier screening	81220
CA Prenatal Screening	81508 (1 st trimester) 81511 (2 nd trimester)
Amniocentesis procedure	59000, 76946, 76811, 76942
CVS procedure	76945, 96040, 59015, 76801, 99199, 76813
Karyotype (on amniotic fluid or CVS)	88235, 88269, 88280, 82106
Microarray (on amniotic fluid or CVS)	81229
AF-AFP (on amniotic fluid for neural tube defects)	82106
NT ultrasound	76813
Detailed ultrasound	76811
Genetic counseling	96040

COMMON BILLING INDICATIONS

Indication	ICD-10 Code(s)
Advanced maternal age, 35+ at delivery (first pregnancy)	O09.519
Advanced maternal age, 35+ at delivery (not first pregnancy)	O09.529
Low risk/maternal concern (first pregnancy)	Z34.02
Low risk/maternal concern (not first pregnancy)	Z34.82
Previous pregnancy/child with a chromosome abnormality	O09.291, O09.292, O09.293, O35.2XX0
Family history of a hereditary disease, birth defects, or a chromosome abnormality	Z84.89, Z82.79
Abnormal maternal serum screening or cell-free DNA screening suggestive of a chromosome issue	O28.5
Abnormal maternal serum screening suggestive of a neural tube defect	O28.1
Abnormality on ultrasound	O28.3, O35.1XX0
Carrier screening for a female	Z31.430
Carrier screening for a male	Z31.440
Carrier screening for a high risk ethnicity	Z15.89
Couple is related by blood (Consanguinity)	Z84.3

COST ESTIMATES

Cell-Free DNA Screening and/or Expanded Carrier Screening

PAMF typically uses a company called Myriad Women's Health for cell-free DNA screening and expanded carrier screening. Myriad is in-network with most insurance providers. Myriad can perform an insurance check to provide an estimate of a patient's out of pocket cost. Patients can call Myriad directly to request this estimate at (888) 268-6795. Myriad's website is myriadwomenshealth.com.

Microarray

PAMF uses a company called Invitae for the prenatal microarray. Invitae can perform a Benefits Investigation before an order for testing is submitted. To request this, send an email to bi@invitae.com and provide the following information:

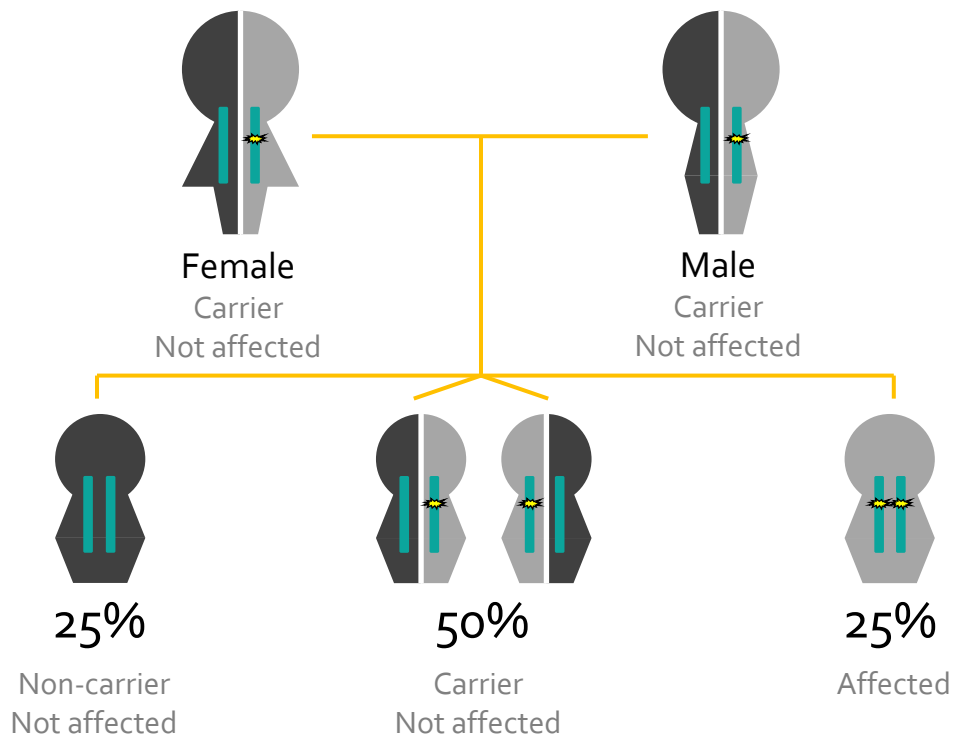
- Patient's full name
- Patient's date of birth
- Patient's insurance information (insurance company name, insurance company address, member ID, group number)
- The test name, which is "Targeted Prenatal Microarray"
- The ICD-10 code(s) indication(s) for the testing

Benefits Investigations can take as long as 5 business days to come back, but often come back sooner.

Invitae's website is www.invitae.com and their phone number is (800) 436-3037.

EXPANDED CARRIER SCREENING

We offer expanded carrier screening to patients who are considering pregnancy or who are currently pregnant. Expanded carrier screening is a blood test which looks to see if someone is a “carrier” for a genetic condition. We typically have two copies of each of our genes. A person who is a carrier has a change in one copy of a gene which makes it not work, but that person is not affected because he or she still has the other working copy of the gene. If both members of a couple are carriers for the same genetic condition, they have a 1 in 4 (25%) chance that they will both pass on the non-working gene in each pregnancy they have together. When that happens, that child has no working copies of the gene, and therefore is affected with that condition. People who are carriers usually have no family history, so most people do not know they are carriers until they have a child born with the condition.



The expanded carrier screening test currently offered by PAMF looks for approximately 175 genetic conditions, including Cystic Fibrosis, Tay-Sachs disease, common types of Thalassemia, Spinal Muscular Atrophy, and Fragile X syndrome. Some genetic conditions are very severe and can significantly impair an affected individual’s normal development or even lead to death in childhood. Some require lifelong management. Other conditions are milder and may be treatable. For some conditions, treatment during pregnancy may be available to improve pregnancy outcome.

Like any carrier screening test, some carriers will not be detected. This test also does not look for every possible genetic condition. Therefore this test can reduce, but not eliminate, the chance for a genetic condition.

EXPANDED CARRIER SCREENING

Frequently Asked Questions

Where can I see a list of the genetic disorders screened by expanded carrier screening?

PAMF orders expanded carrier screening through a company called Myriad. The full list of genetic disorders including descriptions of the conditions is available at myriadwomenshealth.com.

What type of sample is needed for this testing?

Typically expanded carrier screening is done on a single blood draw, however it is also possible to do this testing on saliva.

When is the test performed?

The test can be performed at any time. To have the biggest benefit to you and your partner, it is recommended to have the test done before pregnancy or very early in a pregnancy.

How common is it to be identified as a carrier?

Approximately 60% of individuals who undergo the current expanded carrier screening panel will be identified as a carrier for at least one condition. If one person is identified as a carrier, additional testing will likely be recommended for the other partner. Approximately 2.4% of couples who undergo the current expanded carrier screening panel will be identified as high risk, meaning they have a 25% chance to have an affected child in each pregnancy.

Do the chances to be a carrier change with age?

No, your chance to be a carrier for a genetic condition is not age-dependent.

Do the chances to be a carrier depend on ethnic background?

Some genetic conditions are more common in certain ethnic backgrounds, however people of all ethnicities have a chance to be a carrier of a genetic condition.

What if it is identified that my partner and I are both carriers for the same condition?

If it is identified that you both are carriers for the same condition, you have options. If you are not currently pregnant, you may decide to achieve pregnancy through in vitro fertilization (IVF) with pre-implantation genetic testing (PGT-M), a pre-pregnancy process that significantly reduces the risk that a child will inherit the genetic disease. During pregnancy, you may decide to undergo diagnostic testing for that condition to make informed reproductive decisions. Some individuals consider adoption, egg or sperm donation, or opt to not have children. Even if you would not choose any of these options, you can use the information to prepare for the birth of a child with a genetic condition. If you are both identified as carriers, you will have the opportunity to speak with your physician or a genetic counselor about the options available to you.

What is the accuracy for expanded carrier screening?

There are many factors which determine the accuracy of carrier screening including your ethnic background and how common a condition is. Expanded carrier screening will not be able to detect everyone who is a carrier, however for most of the conditions it will find over 99% of the carriers. After the test is completed, the expanded carrier screen will provide a residual risk that you could still be a carrier even after testing negative (low-risk) on the screen.

Will my insurance cover the cost for expanded carrier screening?

Typically this testing is covered by insurance, however insurance coverage can be variable. The cost for the test may vary for different insurance companies and plans. PAMF orders expanded carrier screening through a company called Myriad, which is in-network with most insurance providers. Myriad can perform an insurance check to provide an estimate of your out of pocket cost. You can call Myriad directly to request this estimate at (888) 268-6795.

Who do I talk to if I'm interested in learning more about expanded carrier screening?

Speak with your healthcare provider or request a referral for you to meet with a prenatal genetic counselor to discuss carrier screening in more detail.