



PRENATAL GENETIC COUNSELING

WHO IS A GENETIC COUNSELOR?

A genetic counselor is a trained health care professional with a specialized graduate degree in the area of medical genetics. In the field of prenatal genetic counseling, a genetic counselor's role is to help a patient understand her risk for having a child affected with a genetic or chromosomal abnormality, educate the patient about the screening and/or diagnostic options available, and coordinate any prenatal or postnatal follow-up necessary.

DO I NEED TO BE PREGNANT TO SEE A GENETIC COUNSELOR?

No. Many people know they are at risk for a chromosome abnormality or a specific disease before they become pregnant. Genetic counselors will gladly help a patient understand a particular genetic condition, research the availability of testing, coordinate maternal and/or paternal testing if indicated, and coordinate prenatal testing if requested by the patient.

WHAT ARE THE MOST COMMON REASONS FOR A WOMAN TO SEE A GENETIC COUNSELOR?

While there are hundreds of reasons, the most common reasons include advanced maternal age (maternal age greater than 35), an abnormal screening test and/or ultrasound in a current pregnancy, a history of a genetic/chromosome condition in a prior pregnancy, a family history of a genetic disease, or a member of an ethnic group at risk for a specific disorder.

WHAT CAN I EXPECT IN A GENETIC COUNSELING SESSION?

Generally, genetic counselors will construct a three generation pedigree of your family asking specific questions about ethnic background, genetic diseases, age of onset of health conditions and prior pregnancy outcomes. They will review the risks, if any, to your current or future pregnancy, and assist you in understanding the risks, benefits and alternative to any available screening or diagnostic tests. Genetic counselors are trained to be objective — respecting and supporting the opinions and views of the patients they counsel. In the event of an abnormal test result, the genetic counselor becomes a valuable coordinator of the care between a patient's obstetrician and any subspecialty care that may be needed for or after the birth of the child.

ISN'T MOST OF THE INFORMATION AVAILABLE ON THE INTERNET?

The internet is a wonderful resource but can also provide misleading and inaccurate information. A genetic counselor can help guide you through the confusing array of information that is critical to know prior to making any decision about your pregnancy.



Diablo Valley Perinatal Associates

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WHY IS ULTRASOUND SO IMPORTANT IN PREGNANCY?

During the first trimester, a fetal ultrasound determines the health of an early pregnancy with first trimester screening.

During the second trimester, a fetal ultrasound rules out most birth defects with a comprehensive evaluation of the fetal anatomy.

During the third trimester, a fetal ultrasound evaluates the size of the developing baby and identifies pregnancies with abnormal growth.

FETAL ULTRASOUND

WHAT IS A FETAL ULTRASOUND?

An ultrasound examination or sonogram consists of an imaging technique that generates a high quality image of the developing fetus within the uterus.

WHY IS IT PERFORMED?

Many birth defects, such as spina bifida, heart defects, brain abnormalities, cleft lip, club foot, chest, abdominal and genitourinary abnormalities can be diagnosed while the fetus is between 18-20 weeks. If the abnormality exists, pediatric subspecialists including heart surgeons, orthopaedic surgeons, craniofacial surgeons, neurosurgeons or intensive care specialists can meet with the expecting parents to help them understand the course of medical care once the baby is born. In certain cases, the specific abnormality may require pediatric subspecialty care to be present at the time of delivery.

WHY DO I NEED AN ULTRASOUND IF MY CVS OR AMNIOCENTESIS WAS NORMAL?

Diagnostic tests such as chorionic villus sampling and amniocentesis identify abnormalities in the chromosomes of developing fetuses (such as Down syndrome). While birth defects are more common in fetuses with chromosome abnormalities, birth defects are also relatively common in fetuses with normal chromosomes. Therefore, a normal CVS or amniocentesis does not eliminate the need for comprehensive ultrasound.

DOES ULTRASOUND POSE ANY RISKS TO MY DEVELOPING BABY?

No. Ultrasound technology is amazing. The probe that is used to generate the ultrasound image emits an ultrasound wave that reflects off of the developing fetus to help generate a high-resolution image. The amount of time the probe emits sounds waves is approximately 1/1000 of the time it spends listening. That means that if the ultrasound exam were to last 24 hours, the fetus would be exposed to roughly 1 minute of ultrasound energy.

WHY SHOULD I HAVE MY ULTRASOUND PERFORMED BY DIABLO VALLEY PERINATAL?

We are a specialized referral center that performs more than 10,000 fetal ultrasounds each year. Our sonographers and physicians are specifically trained to obtain and interpret fetal images. Furthermore, our physicians and genetic counselors are specifically trained to counsel and care for any abnormalities detected.

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WHO SHOULD CONSIDER CHORIONIC VILLUS SAMPLING

You should consider CVS if:

- *You are 35 years old or older*
- *You have had a chromosome abnormality in a previous pregnancy.*
- *You have a family history or are at risk for a specific genetic disorder.*
- *You have received abnormal results from an early screening test (first trimester screening).*

CHORIONIC VILLUS SAMPLING

WHAT IS CHORIONIC VILLUS SAMPLING?

Chorionic Villus Sampling (CVS) is a microscopic sampling of the early developing placenta performed in the first trimester. Since the placenta is made up of fetal tissue, a CVS tells you with nearly 100 percent accuracy the chromosomal makeup of the developing fetus.

WHAT TESTS CAN PERFORMED ON A CVS SAMPLE?

Most commonly, CVS samples are used to test for chromosome abnormalities such as Down syndrome (trisomy 21), trisomy 13, trisomy 18, sex chromosome abnormalities and in some cases, specific genetic disorders such as cystic fibrosis, fragile X or sickle cell disease.

HOW IS A CVS PERFORMED?

There are two possible ways to perform a CVS: transabdominal or transcervical. While some placental locations are accessible to both methods, usually one method is preferable due to the location of the early placenta.

In the transabdominal approach, a needle is passed through the skin (using local anesthesia) into the early developing placenta under ultrasound guidance.

In the transcervical approach, a speculum is placed in the vagina (the same as when a pap test is performed) and a small catheter is guided through the cervix into the placenta under ultrasound guidance.

ARE THERE ANY RISKS?

The risk of miscarriage following CVS is less than 1 in 350. This risk is less than the spontaneous pregnancy loss rate at the gestational age of the CVS for those people not undergoing a diagnostic procedure.

ARE THE RESULTS ACCURATE?

Approximately 1 percent of the time a phenomenon known as confined placental mosaicism may be seen. In this case, the results of a CVS are inconclusive and a follow up procedure such as an amniocentesis may be indicated.

WHAT IS THE ADVANTAGE OF A CVS?

CVS is typically performed between 10-14 weeks. This provides information about the health of the pregnancy earlier than an amniocentesis (which is usually performed between 15 and 20 weeks).



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REFERRAL FORM

You have been referred to Diablo Valley Perinatal Associates (previously known as John Muir Perinatal Medical Group). We look forward to meeting you at your visit. In order to make your time with us more efficient, we encourage you to visit our web site (www.diablovalleyperinatal.com) where you will find helpful information about our practice, your pregnancy, practice policies, directions to our office and registration forms you will need to complete prior to your visit.

PLEASE BRING THIS REFERRAL FORM TO YOUR APPOINTMENT.

Patient Name _____ DOB ___/___/___ LMP/EDC ___/___/___

ULTRASOUND

- Early U/S - Missed or Threatened AB - r/o Ectopic
- Nuchal Translucency (*first trimester screening - between 11-14 weeks*)
- Comprehensive Anatomic Survey
- Follow-up Ultrasound (reason)
- Cervical Length
- Second Opinion for Prior Abnormal Ultrasound (*Please provide report*)
- Other, please specify _____

PERINATAL SERVICES REQUESTED

(Please check all that apply)

- Consultation
- Co-Manage
- Transfer Care
- Other, please specify _____

ANTENATAL TESTING

- NST/AFI
- Biophysical Profile (BPP)
- Amniocentesis-Lung Maturity
Reason _____

PRENATAL DIAGNOSIS/SCREENING/COUNSELING

- Amniocentesis
- Chorionic Villus Sampling (CVS)
- Genetic Counseling
- Other, please specify _____

If patient is having amniocentesis or CVS, we require:

Blood type _____ Anti-body screen _____

Please fax all prenatal records and previous ultrasounds for any consultation with Perinatologist.

Please provide: Blood type and Rh status for any patient undergoing an amniocentesis or CVS, reports for any previous ultrasounds done at another facility and any relevant labs and MCV for every genetic counseling appointment.

Perinatal consultation and follow-up ultrasound requested for any ultrasound abnormality identified.

Physician signature (*required*) _____ Date _____



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THE CALIFORNIA PRENATAL SCREENING PROGRAM

WHAT IS SERUM INTEGRATED SCREENING?

Serum integrated screening is a non-invasive test that provides a pregnant woman with information about her risk of having a child with Down syndrome (trisomy 21), Edward syndrome (trisomy 18), Smith-Lemli-Opitz syndrome (a metabolic defect of cholesterol metabolism) and spina bifida (open neural tube defect).

HOW IS THE CALIFORNIA PRENATAL SCREENING TEST PERFORMED?

There are two components to the California Prenatal Screening Program. The first component (performed between 11-14 weeks) combines an early ultrasound measurement of the fetal neck (the nuchal translucency) with a maternal blood test. These two tests will provide a preliminary risk assessment that a woman's pregnancy is affected with Down Syndrome or Edward Syndrome.

The second component (performed between 15-20 weeks) involves a second trimester maternal blood test. This test results in the final risk assessment for not only Down Syndrome and Edward Syndrome but also for Smith Lemli Opitz Syndrome and spina bifida.

HOW ACCURATE IS THE CALIFORNIA PRENATAL SCREENING TEST?

If the first and second components of the screening test are performed – the screening test is approximately 90 sensitive with a 5% false positive rate for Down Syndrome. That means that approximately 90 of 100 affected pregnancies will be identified as "at risk" while 5% of normal pregnancies will be identified as at risk when they are not. The detection rates for trisomy 18, Smith Lemli Opitz and spina bifida are lower than the detection rate for Down Syndrome.

WHAT IF I SCREEN POSITIVE FOR AN ABNORMALITY?

You will be contacted with your results and offered an appointment with our genetic counselor to discuss your results and options for additional screening or diagnostic testing such as chorionic villus sampling or amniocentesis. It is important to understand that a "screen positive" result does not diagnose an abnormality. Rather, it means you are at an increased risk for an abnormality and you should be informed about the risks and options for additional diagnostic testing.

WHAT IS THE DIFFERENCE BETWEEN A SCREENING TEST AND A DIAGNOSTIC TEST SUCH AS AN AMNIOCENTESIS OR CVS?

A screening test, such as the California Prenatal Screening Program will tell you the probability your pregnancy is affected with one of several birth defects while a diagnostic test such as an amniocentesis or chorionic villus sampling will tell you with nearly 100 percent accuracy whether your pregnancy is affected with a chromosome disorder.



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AMNIOCENTESIS

WHAT IS AMNIOCENTESIS?

An amniocentesis or “amnio” is a diagnostic procedure that samples the amniotic fluid surrounding the developing fetus. Within this fluid (which happens to be fetal urine) are fetal cells that have been shed from the fetal skin, genitourinary system and lungs that can be tested to determine the health of the developing fetus.

WHAT IS THE DIFFERENCE BETWEEN A SCREENING AND DIAGNOSTIC TEST?

Unlike screening tests such as the first trimester screen, or the second trimester expanded AFP screen which generate a risk assessment, amniocentesis is diagnostic — with nearly 100 percent accuracy — in determining whether the pregnancy is affected with a chromosome disorder.

WHICH TESTS CAN BE PERFORMED ON AN AMNIOCENTESIS SAMPLE?

Most commonly, an amniocentesis sample is used to test for chromosome abnormalities such as Down syndrome (trisomy 21), trisomy 13, trisomy 18, sex chromosome abnormalities and in some cases, specific genetic disorders such as cystic fibrosis, fragile X or sickle cell disease.

HOW IS AN AMNIOCENTESIS PERFORMED?

Using ultrasound guidance, a very thin needle is guided through the lower abdomen into the amniotic fluid. A small sample of fluid is withdrawn and sent for chromosome/genetic testing. The fetus is kept in view at all times to assure the procedure is accomplished safely. Despite most women’s fears that the procedure is painful, approximately 97 percent of our patients report the procedure to be significantly less painful than what they expected.

WHAT ARE THE RISKS?

The risk of miscarriage following an amniocentesis is less than 1 in 1,000. Our specialized center with qualified perinatologists and state-of-the-art ultrasound equipment minimizes the risks associated with amniocentesis.

I WOULDN'T TERMINATE MY PREGNANCY REGARDLESS, SO WHY SHOULD I DO AN AMNIOCENTESIS?

Pregnancies affected with Down syndrome are managed very differently than those that are not. Performing an amniocentesis may improve the outcome of your pregnancy, provide peace of mind or help prepare for the delivery of your baby.

WHO SHOULD CONSIDER AMNIOCENTESIS?

You should consider Amniocentesis if:

- *You are 35 years old or older*
- *You have had a chromosome abnormality in a previous pregnancy.*
- *You have a family history or are at risk for a specific genetic disorder.*
- *You have received abnormal results from a screening test (first or second trimester maternal serum screening).*



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FIRST TRIMESTER SCREENING

HOW IS FIRST TRIMESTER SCREENING PERFORMED?

First trimester screening combines a maternal blood sample with an ultrasound evaluation of the early developing fetus. By combining the results of the blood test, the ultrasound evaluation and the maternal age, the risk of having a pregnancy affected with Down syndrome or Edward syndrome can be determined.

WHY AM I BEING REFERRED FOR THIS TEST?

First trimester screening requires specialized training, certification, and state-of-the-art ultrasound equipment usually only available at specialized prenatal diagnostic centers.

WHAT IS THE FIRST TRIMESTER SCREENING?

First trimester screening is a non-invasive test that provides a pregnant woman with information about her risk of having a baby with Down syndrome (trisomy 21) or Edward syndrome (trisomy 18).

WHO IS AT RISK FOR HAVING A CHILD WITH DOWN SYNDROME OR EDWARD SYNDROME?

Every pregnancy is at some risk of being affected with Down syndrome or Edward syndrome. While most people think that only women who are 35 years of age or older are at risk for having a pregnancy affected with Down syndrome, the majority of Down syndrome cases occur in women less than 35 years of age (because that is the age group that has the most pregnancies). Still, per pregnancy, advancing maternal age equates to increased risk of having a pregnancy affected with trisomy 21 or trisomy 18.

HOW ACCURATE IS FIRST TRIMESTER SCREENING?

The accuracy of first trimester screening is approximately 85 percent with a false positive rate of approximately 5 percent. That means that approximately 85 of 100 affected pregnancies will be identified as "screen positive" and approximately 5 percent of normal pregnancies will be identified as being at risk. It is important to know that a negative screening test does not completely rule out the chances that a pregnancy will be affected with Down syndrome — it merely gives a risk assessment.

ARE THERE ALTERNATIVE SCREENING TESTS?

The only alternative screening is a maternal serum test performed between 15 and 20 weeks, otherwise known as "maternal serum screening test" or "AFP test". The accuracy is approximately that of the first trimester screen but results are provided at least 4 weeks later in a pregnancy than the first trimester screening test.

WHAT IS THE DIFFERENCE BETWEEN A SCREENING TEST AND A DIAGNOSTIC TEST?

A screening test provides a risk assessment while a diagnostic test provides a definitive result. For example, a first trimester screening test will provide a result that identifies the odds a pregnancy will be affected with Down syndrome with an amniocentesis or chorionic villus sample will provide a definitive "yes" or "no" answer.

DOES FIRST TRIMESTER SCREENING DETECT OTHER ABNORMALITIES?

Yes. Although not part of the exam, numerous other abnormalities are detected at the time of the first trimester screen. This is largely due to the high resolution ultrasound equipment used to perform the first trimester screen.

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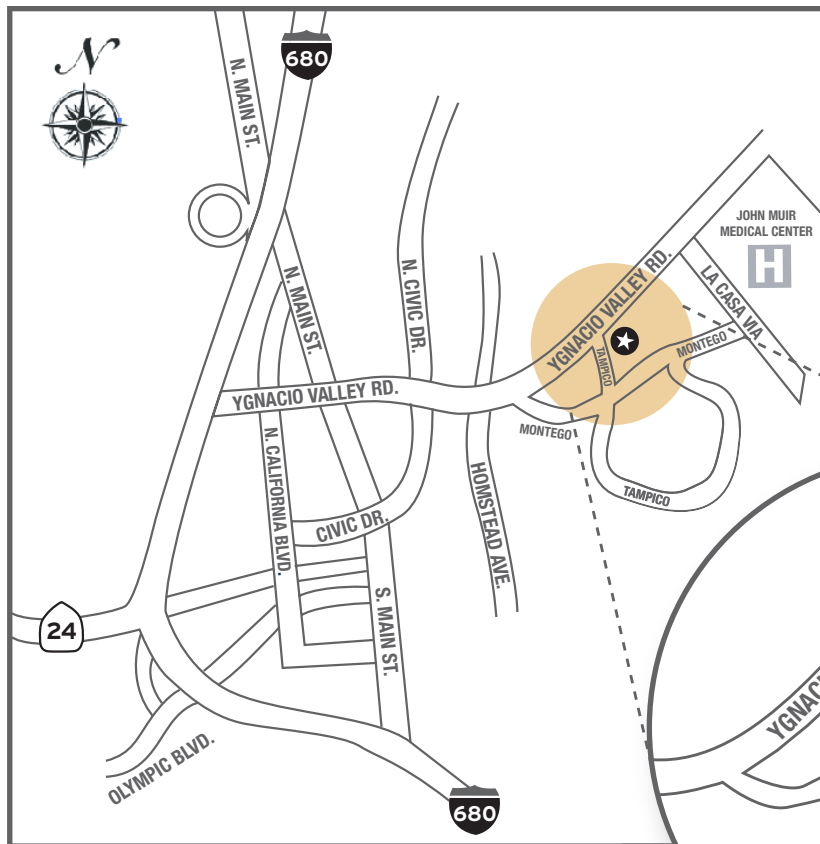
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DIRECTIONS



**110 TAMPICO DRIVE
SUITE 100
WALNUT CREEK, CA 94598**

FREE Parking available.

FROM THE SOUTH BAY

Follow I-680 North to Walnut Creek.
Take the Ygnacio Valley Rd exit.
Turn right onto Ygnacio Valley Rd.
Turn right onto Tampico.
Turn left into first driveway.

FROM THE WEST

Follow CA-24 East through the Caldecott Tunnel.
Keep left, follow signs for I-680 N/Sacramento/Concord
Take Ygnacio Valley Rd exit.
Turn right onto Ygnacio Valley Rd.
Turn right onto Tampico.
Turn left into first driveway.

FROM THE NORTH BAY

From Interstate 80, merge onto I-680 South.
Exit N. Main Street (southbound) in Walnut Creek.
Turn left onto Ygnacio Valley Rd.
Turn right onto Tampico.
Turn left into first driveway.



**DIABLO VALLEY
PERINATAL ASSOCIATES**

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