

**NAME** \_\_\_\_\_ **AGE** \_\_\_\_\_ **DOB** \_\_\_\_\_  
**DATE OF VISIT** \_\_\_\_\_ **REASON FOR VISIT** \_\_\_\_\_  
**APPT. WITH** \_\_\_\_\_

MEDICAL HISTORY	Y	N	EXPLAIN	PROVIDER NOTES
1. Eye, ear, nose, throat problems				
2. Respiratory: asthma tuberculosis other				
3. Cardiac: heart disease Murmur, high cholesterol high blood pressure other				
4. Breasts: prior surgery or biopsy Breast pain Breast lump Mammography			when _____	
5. GI: ulcers, IBS, constipation Colitis, hemorrhoids other				
6. Neurologic: seizures migraines other				
7. Urinary: bladder infection kidney problems urine leakage other				
8. Musculoskeletal: arthritis other				
9. Endocrine: diabetes, osteoporosis thyroid disease other				
10. Bleeding disorder anemia varicose veins sickle cell blood clots other Have you ever had a transfusion? Y N				
11. Skin disease				
12. Psychiatric disorders: depression, counseling hospitalization medications			when _____	

13. Surgical procedures: \_\_\_\_\_ Date \_\_\_\_\_  
 \_\_\_\_\_

14. Medications (include dose and vitamins): \_\_\_\_\_  
 \_\_\_\_\_





Robert Wood Johnson  
Medical Group

ROBERT WOOD JOHNSON MEDICAL SCHOOL

Department of Obstetrics, Gynecology, and Reproductive Sciences

Phone: (732)235-6600

Fax: (732)235-6650

**Genetic Screening**

Date of Visit: \_\_\_\_\_

Patient Name: \_\_\_\_\_ DOB: \_\_\_\_\_

Our office strives to provide you with the most up-to-date information and technologies to determine if there are specific risks to your unborn child. To achieve this goal, we need you to provide us with detailed information about your family history, pregnancy history, medical history and ethnic background. Based on this information, we will offer you certain test(s) during your pregnancy. All of these tests are OPTIONAL. The information in the following pages will help you decide which tests you might want during your pregnancy. If you need additional information or if there are certain risk(s) identified for your unborn child, you may be referred to a genetic counselor here in our department.

**Medical History**

Yes	No	Questions	Who?
		Were you or the father of this baby born with any birth defect(s) or have a condition that you are concerned about passing to your unborn child?	
		Do you or the father of this baby have a child with a disability, birth defect, autism, learning disability or development problems?	
		Did you have a medical condition such as diabetes, thyroid disease, lupus, or epilepsy?	

**Pregnancy History**

Yes	No	Questions	Who?/What?
		Will you be 35 or older at the time your baby is due?	
		Will the father of this baby be older than 45 on your due date?	
		Have you or the father of this baby ever had a pregnancy during which a birth defect was diagnosed?	
		Do you have a history of infertility?	
		Have you had two or more miscarriages?	
		Have you taken or are you taking a medication during this pregnancy?	
		Have you or the father of this baby been exposed to chemicals, x-rays, radiation, or something in the workplace that you are worried about for the pregnancy?	
		During the time you've been pregnant, have you used drugs or alcohol?	
		Do you currently smoke cigarettes?	

Patient Name: \_\_\_\_\_

DOB: \_\_\_\_\_

**Family History**

Yes	No	Questions	Who?
		Do you or the father of this baby have any Jewish ancestors?	
		Do you or the father have French Canadian or Cajun ancestry?	
		Is there a chance you and the father are related by blood to each other?	
		Are there relatives in your family or the family of the father of this baby (mothers, sisters, sister-in-laws, aunts) who had multiple miscarriages, stillbirths, babies that died after birth or pregnancies diagnosed with a birth defect?	
		Do you or the father of this baby have any relatives in the family with the following problems:	
		Down syndrome?	
		Another chromosome or genetic problem?	
		Mental retardation, autism or problems with development?	
		Spina bifida?	
		Cystic fibrosis or someone with chronic lung disease from a young age?	
		Muscular dystrophy, spinal muscular atrophy, or someone who has a genetic muscle condition?	
		Hemophilia or someone who has a bleeding or clotting disorder?	
		Birth defect(s) or an uncommon physical trait such as cleft lip or palate, extra fingers, very short stature?	
		Deafness or blindness?	
		Sudden infant death or unexplained death in your family?	
		Neurologic conditions such as Alzheimer's disease, Huntington disease, Parkinson's disease or other unusual movement disorders?	
		Any other trait in the family that appears to be passed on that you are worried about?	

### **Is your baby at risk for a chromosome problem, such as Down syndrome?**

The risk for a chromosome problem in a pregnancy is related to the age of the mother. As women get older, the risk for a chromosome problem increases. For example, a 25 year old woman has a 1 in 500 chance to have a baby with a chromosome problem. A woman who is 38 years old at delivery has a 1 in 100 chance. However, it is always important to remember, regardless of age, most babies are born healthy and do not have chromosome problems.

Chromosome problems occur by chance and have nothing to do with your family history. The most common chromosome problem that babies can be born with is called Down syndrome (trisomy 21). About 1 in 800 babies overall are born with Down syndrome. All healthy individuals have 46 chromosomes in every cell of their body. When a baby has a missing or extra chromosome this is called a chromosome problem.

Typically, women who are 35 years old or older have the option to have an invasive test, such as chorionic villus sampling or amniocentesis in their pregnancy to learn for sure if their baby has a chromosome problem. However, both these procedures are invasive and carry risks for miscarriage. In addition to these two procedures, there are other non-invasive options which will be reviewed below. Please remember that only amniocentesis and CVS testing can give accurate information about the baby's chromosomes. The other tests discussed below can give some information, but are never 100% accurate.

#### **First Trimester Screening**

This test has to be done between 11 weeks 0 days and 13 weeks 6 days. This is a blood test and ultrasound. The ultrasound is done to measure the amount of skin behind the baby's neck, called nuchal translucency. The results take 4 days to complete. Sometimes you will be asked to return at 16 weeks for a second blood test, called a **sequential screen**. The results of the second blood test and first trimester screen may be combined to give information. Overall, whether you have only the first trimester screen or if you are asked to have the sequential screen, this approach can find up to 90% of pregnancies at risk for Down syndrome and about 97% of pregnancies which are affected by two other chromosome problems called trisomy 13 and 18. These conditions are much more severe than Down syndrome, but are a lot less common than Down syndrome. Please note that sometimes this screen can suggest a risk, but there is nothing wrong with the pregnancy. All patients whose test comes back showing increased risk will be referred for genetic counseling and the option of further testing.

## **Level II ultrasound/Genetic Sonogram**

This is a special ultrasound that is done at 19-20 weeks of pregnancy. All the different body parts of the baby are evaluated including the arms, legs, brain, heart, kidneys, stomach, face, bladder, and spine. Many, but not all, birth defects can be seen. In addition to looking for birth defects, sometimes subtle features (called markers) can be seen that increase the suspicion that a baby has a chromosome problem. Approximately 50% of fetuses with Down syndrome will have some unusual features seen by ultrasound. More than 90% of fetuses with trisomy 13 and 18 will have unusual features seen at the time of this ultrasound. Therefore, a normal ultrasound, reduces, but does not eliminate the chance to give birth to a baby with a birth defect or a chromosome problem.

## **Non-invasive Prenatal Screening using cell free fetal DNA in the maternal blood**

Depending on the laboratory used, this test may be called the Harmony test, MaternT21 testing, or the Verifi test. This is a new test which has only been available since the beginning of 2012. This is a blood test that can be collected starting at 10 weeks of pregnancy. The test works by finding the fetal DNA (genetic material) in a pregnant woman's blood and testing that DNA for Down syndrome, trisomy 13 and trisomy 18. Some of the laboratories can also test that DNA for chromosome X and Y, thus also giving information about the baby's gender. In these cases, sometimes a baby may be found to have a sex chromosome abnormality (abnormal number of X or Y chromosomes). Please note that this test does not replace CVS or amniocentesis. However, this test has been shown to be over 99% accurate for Down syndrome and also accurate for trisomy 13 and 18. Currently, this test is offered to women who are 35 years old or women whose fetus has a slightly higher risk than typical to have a chromosome problem. If this test does suggest a chromosome problem, a CVS test or amniocentesis test would still be recommended to confirm whether the blood test is correct.

## **Amniocentesis**

This is a highly accurate diagnostic test. It is usually performed between 16 and 22 weeks of pregnancy. The amniocentesis is performed by inserting a small needle through your abdomen into the uterus under ultrasound guidance to collect about 2 tablespoons of amniotic fluid. The needle does not touch the baby. However, the test does carry a risk for a complication which includes miscarriage of pregnancy. The risk of this procedure is up to 1 in 200 (0.5%). By obtaining the amniotic fluid, the baby's cells, which are floating in that fluid, are collected. In addition to testing for Down syndrome and other chromosome problems, additional genetic testing can be done, depending on your specific situation. All patients undergoing an amniocentesis test are encouraged to meet with a genetic counselor prior to the procedure.

## **Chorionic Villus sampling (CVS):**

This is another accurate diagnostic test that gives similar information to amniocentesis. However, this test is done between 10-13 weeks of pregnancy. It is done under ultrasound guidance by a high risk pregnancy doctor. This test is done by either inserting a needle through the women's abdomen or through the cervix to collect some cells from the placenta. How the test is done is dependent on where the placenta is located. The risk of this procedure for complications including miscarriage is up to 1 in 100 (1%). By obtaining the placental cells, you can get accurate information about the baby's chromosomes. In addition, other genetic testing can be done, depending on your specific situation. All patients undergoing an amniocentesis test are encouraged to meet with a genetic counselor prior to the procedure.

**Fetal Microarray testing**

If a patient undergoes the CVS or amniocentesis test, an additional test that can be run on these cells is called the microarray test. Microarray testing is a special way to look at the baby’s chromosomes, but at a much higher resolution. A typical chromosome test counts the number of chromosomes to make sure all 46 are present. A microarray test looks at all the 46 chromosomes with much higher scrutiny to make sure that no small pieces are missing or extra. Missing or extra small segments (also known as microdeletions or microduplications) are associated with hundreds of different and unique genetic conditions. Most of these conditions happen by chance and are not related to the age of the mother. One study has shown that about 1 in 60 fetuses have an important piece missing or extra in their chromosome which can only be picked up by the microarray test. If this is a test in which you are interested in obtaining more information about, please consider meeting with one of our genetic counselors.

I have read and understand this information. I have decided the following:

- I do not want to have any of the above tests.
- I want to have First Trimester Screening.

Patient name (printed): \_\_\_\_\_

DOB: \_\_\_\_\_

Patient signature: \_\_\_\_\_

Date: \_\_\_\_\_

Witness: \_\_\_\_\_

Date: \_\_\_\_\_

### **Genetic Carrier Screening (available to all women)**

**All humans have about 30,000 pairs of genes. Within each pair, one gene is inherited from each parent. It is believed that all people have at least 2-3 genes that “do not work properly.” This causes a person to be a carrier for a genetic condition. Typically, carriers for a genetic condition do not have symptoms of the disease, they are healthy. However, if the father and mother are carriers for the same condition, their child has a 25% chance of having this condition. This is called autosomal recessive inheritance. Below are two conditions, for which carrier testing is readily available. Please note that carriers usually do not have a family history of the condition, as the condition stays “silent’ until two carriers have a child together. Our practice currently recommends testing for both of these conditions.**

#### **Cystic Fibrosis disease carrier testing:**

This is the most common autosomal recessive genetic disorder affecting about 1 in 3,300 people in the United States. The chance for a person to be a carrier for this condition depends on their ethnic background. Approximately 1 in 25 Caucasians, 1 in 46 Hispanic individuals, 1 in 65 African Americans and 1 in 90 Asians are carriers for this condition. Cystic Fibrosis causes the body to produce abnormally thick mucous, leading to life-threatening lung disease and digestive problems. Symptoms can be variable ranging from mild to severe. However, the average lifespan for a person with this condition is into their later 30s. A blood test can determine if you are a carrier for this condition. If you are negative, this REDUCES your chance to be a carrier, but does not eliminate it. If you are found to be a carrier, testing the father of this baby will be necessary. If you are both found to be carriers, there will be a 25% chance for your baby to have this condition. If you are both found to be carriers, it is possible to test the baby during the pregnancy by either chorionic villus sampling or amniocentesis. Genetic counseling is recommended for all couples determined to be at risk.

#### **Spinal Muscular Atrophy carrier testing:**

This is the second most common genetic disorder in the U.S. affecting about 1 in 6,000-10,000 individuals of all racial and ethnic groups. Approximately 1 in 41 people carry the gene for this disorder. There are several forms of this disorder causing variable age of onset and severity of symptoms. In general, progressive muscle degeneration occurs affecting crawling, walking, swallowing and head and neck control. In the most severe form, children have difficulty with breathing by age two. A blood test can determine if you are a carrier for this condition. If you are negative, this REDUCES your chance to be a carrier, but does not eliminate it. If you are found to be a carrier, testing the father of this baby will be necessary. If you are both found to be carriers, there will be a 25% chance for your baby to have this condition. If you are both found to be carriers, you will have the option to test the fetus for this condition either through chorionic villus sampling or amniocentesis. Genetic counseling is recommended for all couples determined to be at risk.

There are some conditions that are caused by genes on the X chromosomes. Therefore, if a woman is a carrier for a gene problem on the X-chromosome, she will have up to a 50% chance to have an affected child (mostly boys would be affected). This is different from the two above conditions because having a carrier mother is what increases a child's chance to be affected. Below is an X-lined condition for which carrier testing for the mother is readily available.

Our practice currently recommends testing for this condition.

### **Fragile X carrier testing:**

This is the most common cause of inherited mental retardation affecting about 1 in 4,000 males and 1 in 8,000 females. It has been estimated that about 1 in 260 women are carriers for this condition. People with this condition have delayed speech and language, a degree of mental retardation, behavioral characteristics such as hyperactivity, autism and poor eye contact. You can have a blood test to see if you are a carrier for this condition. If you are found to be a carrier, genetic counseling is recommended to help you understand the specific risk to your unborn child. A woman who chooses to have this blood test can end up having one of the four following results: negative (children not at risk), intermediate (children most likely not at risk), premutation (children at risk), full mutation (children at risk). All women who are determined to be at risk for a child with Fragile X syndrome can choose to undergo chorionic villus sampling or amniocentesis to determine if their fetus has Fragile X syndrome.

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### **NEW TECHNOLOGY- Expanded Carrier Testing**

This is a new panel which includes all of the above conditions (Jewish disorders, cystic fibrosis, Spinal Muscular Atrophy, Fragile X syndrome) plus an additional number of conditions. This panel allows you to be tested for over 100 genetic conditions simultaneously to see if you are a carrier. For most of these conditions, it requires both the mother and father to be carriers for the same condition in order for there to be a risk to their unborn child. This panel contains conditions that are mostly severe and may cause significant health problems. However, some of the conditions are milder and may only affect one body system. For some of the conditions on the panel, a negative result is highly accurate. For other conditions, a negative result only slightly reduces the chance of being a carrier. **It is expected that at least 25% of people who have this panel will be found to be a carrier for at least one condition.**

Please remember this does not mean that you are sick or have this condition. If you are found to be a carrier, we will refer you and the father of the baby to our genetic counseling department so that he may be tested. All carriers may be asked to see a genetic counselor to discuss the result and the accuracy of the test for your partner. In rare cases, being a carrier may provide some information about your health. In this case you will be asked to meet with a genetic counselor to discuss the results. It is important for you to understand that this is the most comprehensive panel currently available but it still does not test for every possible genetic condition.

**Our practice currently offers Expanded Carrier Screening via the company, Counsyl. Many insurance companies will cover at least part of the cost of genetic carrier screening, including Expanded Carrier Screening, however our office cannot determine your exact coverage. It is likely that if you do not have complete coverage, the out of pocket expense to you would not exceed \$99.**

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I have read and understand the information on this page. I have decided the following:

- I do not want to have any of the above tests.
- I only want to be tested for Cystic Fibrosis, SMA and Fragile X.
- I want to have Expanded Carrier Screening (which includes Cystic Fibrosis, SMA and Fragile X).

Patient name (printed): \_\_\_\_\_

DOB: \_\_\_\_\_

Patient signature: \_\_\_\_\_

Date: \_\_\_\_\_

Witness: \_\_\_\_\_

Date: \_\_\_\_\_

**Jewish Carrier Screening Consent Form**

You have indicated that you and/or the father of this baby have Jewish ancestry. Please understand that people of Jewish descent are not more likely to have a child with a genetic disorder. Rather, certain disorders are more common in the Jewish population and blood tests for these disorders are readily available. All of the conditions described below are inherited in an autosomal recessive pattern. This means that both the mother AND the father need to be a carrier (for the same condition) for a baby to be at risk. We can start by testing you first, and then if you are a carrier, we will recommend that the father of this baby be tested. There are a few pieces of information that are important to understand:

- 1) Carriers will ALWAYS be healthy.
- 2) Carriers should tell their family members about their results because the gene is passed in their family.
- 3) If both the mother and father are carriers for the same condition, they have a 25% chance to have a child with the disease. In these cases, you will have the choice to test the fetus during pregnancy.
- 4) Carrier testing will not diagnose 100% of carriers. However, if you are negative by the carrier test, your chance to be a carrier is very small.

The following is a list of sixteen conditions for which a blood test is readily available

<b>Disorder</b>	<b>Chance for a Jewish person to be a carrier</b>	<b>What the condition causes</b>
Tay Sachs disease	1 in 30	Progressive brain disease that causes death by age five
Canavan	1 in 40	Progressive brain disease that causes death in early childhood.
Cystic Fibrosis	1 in 26-29	Chronic condition that causes varying lung disease, digestive disease, sometimes infertility. Does NOT cause mental retardation.
Familial Dysautonomia	1 in 32	Present from birth and causes the body to have a difficult time regulating blood pressure, temperature, swallowing, and ability to respond to stress.
Gaucher Disease	1 in 15	Can be very mild to severe and can start in childhood or adulthood. There is treatment for this condition. May cause enlargement of the liver/spleen, neurologic problems, nasal bleeding, and bone fractures.
Niemann-Pick disease (type A)	1 in 90	Progressive disease that causes mental retardation and physical deterioration with death typically by age four.

Fanconi anemia (Group C)	1 in 89	Variable and usually diagnosed in early childhood. Findings include a combination of aplastic anemia, short stature and malformations of the heart, kidneys, or limbs. These individuals are also at risk for mental retardation and increased predisposition to cancer.
Bloom syndrome	1 in 100	Causes poor growth, immune system, and high rate of cancer. Affected individuals usually die of cancer before the age of 30. The intelligence is normal.
Mucopolysaccharidosis type IV	1 in 100	Starts in first year of life causing mental retardation and eye disease
Maple Syrup Urine Disease		Symptoms progress very quickly after birth and include poor feeding, vomiting, seizures, mental retardation and developmental delay
Glycogen Storage Disease type IA	1 in 130	Starts in infancy with delayed growth, seizures, enlarged liver, renal problems, slow growth and short stature.
Familial Hyperinsulinism	1 in 68	Starts with low blood sugar in the newborn and can result in seizures, poor muscle tone, feeding and breathing difficulties. This condition is fatal, if left untreated.
Joubert syndrome 2	1 in 110	Neurologic disorder associated with a spectrum of brain abnormalities, renal and eye problems.
Nemaline Myopathy	1 in 168	A progressive disorder that affects the brain and the muscles leading to difficulty breathing
Usher syndrome I	1 in 147	Causes congenital deafness and progressive blindness
Usher syndrome III	1 in 120	Causes later onset hearing loss and variable onset of blindness

Jewish Carrier testing

I have read and understand the information. I have decided the following:

- I do not want to have any of the above tests
- I want my blood drawn and tested for all the above tests.
- I want to only have testing for the following conditions:

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Patient name (printed): \_\_\_\_\_ DOB: \_\_\_\_\_

Patient signature: \_\_\_\_\_ Date: \_\_\_\_\_

Witness: \_\_\_\_\_ Date: \_\_\_\_\_

# **IMPORTANT**

**If you are a pregnant patient, and have a question, problem, concern, and/or are in labor, please call our Obstetrical Coordinator at one of the following numbers:**

**Monday through Friday – 9am to 5pm  
(732) 235-6143**

**\*Monday through Friday after 5pm,  
Saturday, Sunday, or a Legal  
Holiday  
(732) 235-6600**

**\*The service will contact the on-call physician**

**DIRECTIONS – HOW TO GET TO THE**  
**RUTGERS ROBERT WOOD JOHNSON MEDICAL GROUP**  
**CLINICAL ACADEMIC BUILDING (CAB)**  
**125 Paterson Street Suite 4200**  
**New Brunswick, New Jersey 08901**  
**(732)235-6600**

**FROM NEW JERSEY TURNPIKE:**

Exit 9 (New Brunswick). Take Route 18 North about two miles to the Route 27 South (Princeton) exit. Follow Route 27 for six traffic lights. At the 6<sup>th</sup> traffic light, make a left turn onto Paterson Street. Entrance to the CAB parking deck is on your right.

**FROM ROUTE #1:**

Take Route 18 North exit and follow directions as above under N.J. Turnpike.

**FROM ROUTE #287:**

Take Exit #10 (Route 527, New Brunswick). Follow Route 527 (Easton Ave.) for about six miles to the end, and make a right turn onto Route 27 South. At the 2<sup>nd</sup> traffic light make a left turn onto Paterson Street. Entrance to the CAB parking deck is on your right.

**FROM GARDEN STATE PARKWAY NORTH:**

Exit #130 (Route 1 South). Go about nine miles to Route 18 North. Take Route 18 North and follow directions above under New Jersey Turnpike.

**FROM GARDEN STATE PARKWAY SOUTH:**

Exit #105 (Route 18 North). Stay on Route 18 North for approximately a half-hour drive until you pass the New Jersey Turnpike entrance. Then follow directions on Route 18 North above under N.J. Turnpike.

**NOTE: Entrance to the Clinical Academic Building is across the street from the entrance to Paterson Street CAB parking deck.**

**Please arrive 20 Minutes prior to your scheduled appointment time to allow for parking and registration activities. You have several options for parking:**

- Valet parking at the entrance of the Clinical Academic Building at 125 Paterson Street
- Self-Parking directly across the street from the Clinical Academic Building at the 124 Paterson Street Parking Deck
- Self-Parking one block from the Clinical Academic Building at the 20 Plum Street Parking Deck
- Self-Parking one block from the Clinical Academic Building at the 95 Peterson Street Parking Deck

\* **Please note that we DO NOT validate for parking**

**DIRECTIONS TO:**  
**The Rutgers Robert Wood Johnson Medical Group Monroe Office**  
**18 Centre Drive**  
**Monroe Township, New Jersey 08831**  
**Phone: (609)655-2411**  
[www.rwjma.com](http://www.rwjma.com)

**Directions from points north via the Garden State Parkway/NJ Turnpike:**

- Take the Garden State Parkway South
- Take exit 129 off the Parkway (I-95/NJ Turnpike)
- Follow the signs toward the NJ Turnpike South
- Take Exit 8A towards Jamesburg
- Merge onto CR-612 E/Rt. 32 East
- Turn Right onto Applegarth Road
- Make the first left on Centre Drive
- Make the first right into 18 Centre Drive

**Directions from points east via Route 18:**

- Take Route 18 North
- Take exit 29 towards CR-520/Tennent Road
- Merge onto CR-3/Tennent Road
- Turn right at Bridge Road/CR-520
- Turn left at CR-520/Texas Road
- Continue to follow Texas Road (turns into Matchaponix Avenue)
- Continue on Matchaponix Avenue (turns into Pergola Avenue)
- Continue on CR-612/Pergola Avenue
- Turn right at Buckelew Avenue/CR-522/CR-612
- Turn left at CR-612 Forsgate Drive
- Turn left onto Applegarth Road
- Make first left onto Centre Drive
- Make first right into 18 Centre Drive

**Directions from points east via I-95:**

- Take I-195 west
- Follow the signs toward the NJ Turnpike
- Take the NJ Turnpike North
- Take Exit 8A towards Jamesburg
- Merge onto CR-612 E/Rt. 32 E
- Turn right onto Applegarth Road
- Make the first left onto Centre Drive
- Make the first right into 18 Centre Drive

**Directions from points south via the Garden State Parkway:**

- Take the Garden State Parkway North to I-195 West
- Follow **Directions from points east via I-195**