OFFICE HOURS:

Monday-Thursday	8:00 am – 5:00 pm	Lunch 12:30-1:15
Friday	8:00 am – 12:00 pm	

DAY OR NIGHT PHONE NUMBER: 281-446-4644

ANSWERING SERVICE 281-540-9629

After the office is closed, the answering service is available. They will take your phone number and the nature of the call and forward the information to the doctor. Always tell them if it is an emergency and your call will be answered immediately. The answering service number is also given on the message machine at the main number.

******In case of emergency and you are unable to reach the office or the answering service, you will need to go directly to Memorial Hermann Northeast (or nearest) emergency room. In case of life threatening emergency, call 911*******

Obstetrical Fees: $2700 for normal vaginal delivery
$2900 for cesarean delivery

NO INSURANCE:

Obstetrical fees must be paid by your 28th week of pregnancy. On your first visit, we ask that you bring a deposit of $1000. The amounts stated above DO NOT include your lab work and ultrasound. The fees for lab work are approximately $950 and will be added to your payment plan. Ultrasounds are $450. A letter will be sent to you with a payment schedule showing amounts that must be paid at each visit.

INSURANCE:

With verifiable insurance you will be responsible for your deductible plus the percentage that your insurance does not pay, by your 28th week. You will be receiving information in the mail that explains your plan's benefits, which will include a payment schedule.

Pre-certification: Some insurance companies require that all hospital stays be pre-certified. This means calling a number that is usually on your card and informing them of your pregnancy, due date, and name of hospital. Failure to do this will reduce your benefits. Our office will contact your insurance company for pre-certification; however, we do advise you to do this also.

IF YOUR INSURANCE CHANGES OR IS CANCELLED IT IS YOUR RESPONSIBILITY TO NOTIFY THE OFFICE IMMEDIATELY OTHERWISE YOU WILL BE RESPONSIBLE FOR THE ADDITIONAL EXPENSE

MEDICAID PATIENTS:

It is your responsibility to bring your Medicaid card to the office each month. Also, you must complete all information needed by Medicaid in order to remain eligible. If you lose your Medicaid eligibility you will be considered a cash-paying patient.

If you have insurance and Medicaid; you <u>MUST</u> provide your regular insurance information!! If you only show your Medicaid information and you have insurance with your job, spouse or parents and do not give this information to the office it is considered Medicaid fraud!

HOSPITAL PRE-REGISTRATION:
It is your responsibility to pre-register at the hospital on or before the 28th week of pregnancy. To pre-register go to the Main Admitting Office at Memorial Hermann Northeast; you will need to bring your insurance information and photo ID at this time. Dr. Schettler only has admitting privileges at Memorial Hermann Northeast Hospital.

ADDITIONAL CHARGES:
If a Cesarean Section is performed another doctor will be summoned to assist on the surgery. A separate bill will sent by his office.

High risk pregnancies (twins, vaginal birth after c-section, diabetes, smokers, obesity, hypertension, previous premature labor, etc.) will be charged an additional $500.

TUBAL LIGATIONS:
You must notify our staff if you wish to have a tubal ligation at the time of your delivery. Some insurance companies require special pre-certification for this procedure. Patients covered by Medicaid must have a signed tubal consent form dated **30 days prior** to delivery and you must be at least 21 years old. Tubal ligation done at the time of Cesarean Section is $500. If you have a vaginal birth, your tubal ligation will be scheduled as an outpatient surgery 4-6 weeks after delivery.

ON CALL SCHEDULE:
Dr. Schettler shares weekend call with Drs. Miles Mahan, David Bonilla, Esohe Faith Ohuoba and Larissa Colon-Rodriguez. They are also on call when Dr. Schettler is away from the office.

POLICY REGARDING CHILDREN:

Please make every effort to find childcare for your children during your appointments. If you do not have childcare, you may need to reschedule your appointment. Children are not allowed in the waiting room unaccompanied by an adult. When another adult is present, please have them stay with a sick or disruptive child in the common area near elevators.

Children should not accompany the mother during the last 4 weeks of pregnancy when pelvic exams are required; or you should bring another adult to stay with the child during the pelvic exam.

Food and/or drink in the waiting room is not allowed except for water bottles.

APPOINTMENTS:

Early pregnancy to 28 weeks	Every 4 weeks
28 weeks to 36 weeks	Every 2 weeks
36 weeks to delivery	Every week

If deemed necessary by the doctor, you will be seen more frequently.

The length of pregnancy is 40 weeks.

First prenatal visit: On your first visit a medical; gynecological and obstetrical history will be taken. Dr. Schettler will then do a physical and pelvic examination to obtain information concerning your health and progress of your pregnancy. Prenatal lab work is required on every patient, which includes mandatory Acquired Immune Deficiency Syndrome (AIDS).

Each visit you will be weighed, have your blood pressure checked, urine checked for infection, protein and sugar and uterine growth measurements.

The doctor will listen for the baby's heart beat after 12 weeks. You should start to feel the baby move between 18 and 20 weeks.

If the doctor is called away from the office for an emergency, you may be seen by his nurse.

TESTING:

Q-Natal/MaterniT-21 non-invasive fetal testing: will be offered between 10-20 weeks of pregnancy. This is a blood test that screens for Downs Syndrome and other chromosome abnormalities (see pages 5-6). This is not covered by all insurance plans, you are responsible for checking with your plan to ensure coverage. If not covered by your insurance, there is special arrangements/ cash pricing. Special consent for this test will be discussed with you by your nurse at your appointment.

Maternal Serum Prenatal Screening (AFP): This is a blood test performed between 16-22 weeks of pregnancy. This test is recommended in order to assess the patient's risk for spina bifida, Down syndrome, and other chromosomal abnormalities. If you opt to have the Harmony/Panorama testing performed earlier in pregnancy, testing at this time would be for spina bifida risk only.

During the second trimester it is required that our patients undergo a Glucose Screening Test which requires drinking 50 grams of a glucose preparation and then having your blood drawn at the lab one hour later. A complete blood count for anemia will be done at the same time.

Rhogam: All patients whose blood type is RH negative and whose partner is RH positive must undergo a screening test to detect RH incompatibility. The patient will then receive an injection of Rhogam at 28 weeks.

Group Beta Strep Vaginal Culture: A vaginal culture will be done at initial visit and 34-36 weeks of pregnancy to determine the presence of bacteria that could be harmful to the baby during delivery. If previously have tested positive for GBS, you will not be retested as you will always be considered as positive (a carrier of this bacteria)

Ultrasounds: will be performed on the initial visit to determine your due date; follow up ultrasounds will be performed at the doctor's discretion. You will have an anatomy scan/detailed ultrasound at 24-26 weeks scheduled in the hospital imaging department.

NOTIFY THE OFFICE IF ANY OF THESE SYMPTOMS OCCUR:

- Bloody discharge from the vagina. If slight staining occurs during the night it can wait until morning. However, if bleeding is associated with pain, you saturate a pad, or it runs down your leg, contact the office immediately or go to Memorial Hermann Northeast or nearest emergency room.
- Persistent severe headaches.
- Severe or persistent nausea and/or vomiting (vomiting several times within a few hours).
- Chills and fever of over 101.
- Swelling of ankles, feet, hands, and face.
- Severe abdominal pain.
- Sudden gush of water from the vagina or leaking of fluid, notify the office immediately.
- Frequency or burning with urination.
- Blurring of vision or seeing spots.

DO NOT TAKE ANY MEDICATION WITHOUT CHECKING WITH THE OFFICE.

ACTIVITY: Continue a normal and active life. Work normal schedule. No lifting over 20 pounds. Use of hot tubs and tanning beds is not recommended.

TRAVEL: If taking a long car trip, stretch your legs and empty your bladder every 1 ½ to 2 hours. Check with the office about travel if you are over 26 weeks pregnant. We strongly discourage travel to any patient that has had problems during the current pregnancy.

POSTPARTUM: Your stay in the hospital is usually two to three days depending on the type of delivery you have (vaginal or cesarean).

You will need to schedule a follow up appointment in 10 days if you have a cesarean section so that we can check your incision; for a vaginal delivery your appointment will be at 3 weeks after delivery. Please call the office to schedule an appointment as soon as possible after leaving the hospital.

PLEASE INFORM THE NURSES OF THE FOLLOWING DECISIONS:
Breast or bottle feeding
Anesthesia choice
Pediatrician (baby's doctor)
Tubal ligation at the time of delivery (Cesarean Section)

NON INVASIVE PRENTAL TESTING (Q-Natal/MaterniT-21)

This is simple blood test offered to pregnant patients between 10-20 weeks of pregnancy. During pregnancy, some of the DNA from the baby crosses into mom's bloodstream. DNA is organized in structures known as chromosomes, which carry the baby's genetic information. This DNA can be taken and analyzed and with 99% accuracy can diagnose if your baby will have a chromosome abnormality for Downs Syndrome (Trisomy 21) and Trisomy 18/13. It can also, in some instances, determine the gender of the baby. As mentioned above, this test is not covered by all insurance plans and the cost is approximately $700.00. Your nurse will review this information and have special consent forms for you to sign.

MATERNAL SERUM PRENATAL SCREENING TEST (AFP)

This is a simple blood test that is offered between 16-22 weeks of pregnancy. The purpose of the test is to identify a group of people (in this case pregnant women) at increased risk for certain conditions. This screening test is not capable of diagnosing the conditions rather it can determine if you are at increased risk for the condition. The conditions screened for by this profile are birth defects called Down's Syndrome (Trisomy 21) and Trisomy 18/13 and neural tube defects. If you have previously had non-invasive prenatal testing (above), you will only be screened for neural tube defects at this time.

WHAT IS DOWN'S SYNDROME?

Down's Syndrome is caused by a change in the normal number of chromosomes. Chromosomes are units of inheritance. In a normal pregnancy each baby inherits a total of 46 chromosomes, 23 from the mother and 23 from the father. Sometimes an error in division will cause a baby to inherit too many or too few. In Down's Syndrome a baby inherits 3 of chromosome 21. Down's Syndrome is often called Trisomy 21. Individuals with Down's Syndrome have varying degrees of mental retardation and may also have heart and other defects. Down's Syndrome occurs in about 1 in 800 births.

WHAT IS TRISOMY 18 AND 13?

Trisomy 18 and 13 are other conditions in which the baby has extra genetic material (one extra copy of chromosome 18 or 13). This condition is associated with severe mental retardation. Trisomy 18 (Edwards Syndrome) occurs in only 1 in 6000 live births. Trisomy 13 (Patau Syndrome) occurs in 1 in 16,000 live births.

WHAT ARE NEURAL TUBE DEFECTS (SPINA BIFIDA)

Neural tube defects (NTDs) are defects in the normal formation of the spinal cord or brain. Spina bifida is a form of neural tube defect. In this condition, the neural tube does not close completely along the fetal spine; and the nerves in the spinal cord do not properly connect with all the nerves in the lower part of the body. Spina bifida can interfere with bowel and bladder control and may also be associated with an accumulation of fluid in the brain (hydrocephalus). In most cases, surgery is necessary (often more than one) during infancy and childhood. Neural tube defects occur in about 1-2 in 1,000 pregnancies.

WHAT DOES A NEGATIVE TEST MEAN?

A negative test results significantly reduces the likelihood that your baby has open neural tube defect, Down's Syndrome or Trisomy 18/13. A negative screen does not, however, guarantee that your baby will not have some form of birth defect.

WHAT DOES A POSITIVE TEST MEAN?

A positive NIPT result means that there is a 99% likelihood of your baby having a chromosome abnormality.

A positive AFP means there is an increased risk of your baby having an open neural tube defect, Down's Syndrome, Trisomy 18/13 or other problem.

A positive result on either test means further testing and consultation with a specialist will be recommended.

HOW WILL I LEARN THE RESULTS OF MY TEST?

All results are sent directly to our office and are generally available in 10-15 business days. Only **abnormal (positive)** test results are called to the patient. Normal results will be reported to you at your next prenatal visit.

WHAT HAPPENS NEXT IF I HAVE A POSITIVE RESULT?

One or all of the following will be recommended: genetic counseling, consultation with maternal fetal specialist, ultrasound and amniocentesis. Amniocentesis is a procedure in which amniotic fluid is withdrawn with a needle from the sac surrounding the fetus. Genetic counseling provides you with an opportunity to discuss the results and to make a decision whether further diagnostic testing such as amniocentesis would be helpful to you. The decision to do any further testing is yours.

SUMMARY

It is important to remember that a normal test cannot ever guarantee a baby will be born "perfect". Prenatal screening and testing can only assess for specific conditions.

The decision to have prenatal screening is best made by you. Your health care provider and this packet can only offer information and guidelines. If you have other questions or concerns about testing, do not hesitate to ask our nursing staff so that you have all the information you need to make an informed decision.

PATIENT NAME_____ MR_____

MATERNAL SERUM PRENATAL SCREENING CONSENT

I have read the attached information concerning the maternal serum prenatal screening for neural tube defects/Down's Syndrome/Trisomy 18/13 and:

_____ I prefer to have the testing.

_____ I prefer NOT to have the testing.

_____ _____
Patient Signature Date

_____ _____
Witness Signature Date

PATIENT NAME _____ MR_____

PRENATAL EDUCATION ACKNOWLEDGEMENT

WE WOULD LIKE TO INFORM YOU OF THE FOLLOWING INFORMATION REGARDING YOUR PREGNANCY.

1. Tobacco smoke is a major source of carbon monoxide, which can interfere with oxygen supply to the baby. Studies have shown that smoking can result in miscarriages, smaller babies, prematurity and other serious complications. DO NOT SMOKE DURING THE DURATION OF YOUR PREGNANCY.

2. Recent studies indicate that even moderate drinking increases the risk of miscarriage and FAS (Fetal Alcohol Syndrome).
   DO NOT DRINK ALCOHOL DURING THE DURATION OF YOUR PREGNANCY.

3. Marijuana, cocaine and other mind altering drugs should be avoided during pregnancy. Recent studies have shown that women who use cocaine may have a higher rate of spontaneous abortion and fetuses exposed to cocaine may have a greater risk of birth defects and even death at or around the time of birth. DO NOT USE DRUGS DURING THE DURATION OF YOUR PREGNANCY.

I HAVE READ AND I UNDERSTAND THE INFORMATION ABOVE.

_____ _____
PATIENT SIGNATURE DATE

INSURANCE AND PRE-REGISTRATION

I HAVE READ AND I UNDERSTAND AND THE INSTRUCTIONS FOR PRE-REGISTRATION PROCEDURES AT THE HOSPITAL AND PRE-CERTIFICATION PROCEDURES FOR MY INSURANCE COMPANY.

_____ _____
PATIENT SIGNATURE DATE

MR_____

Patient's Name_____ Blood Type_____

Father of Baby_____Blood Type_____

	RACE	RELIGION	OCCUPATION	EDUCATION (YRS)
Patient	_____	_____	_____	_____
Father	_____	_____	_____	_____

Are you OR the baby's father of:

Greek, Italian or Asian ancestry?_____ If so, have you had Thalassemia testing?_____ When/where?_____

Jewish or French Canadian ancestry?_____ If so, have you had Tay-Sachs carrier testing?_____ When/where?_____

Black or East Indian ancestry?_____ If so, have you had Sickle Cell Disease carrier testing?_____ When/where?_____

Are you and the baby's father blood relatives?_____

Have you had any surgeries or chronic illnesses? Have you or the father been on medication for extended periods? If so, please describe:

_____

_____

Children (living or deceased):

Name	Age	Sex	Health	Father/Mother (If different)

Patient Name:_____ MR_____

Have you had any miscarriages?_____ If so, please describe:_____
_____

Have you had any stillborn infants?_____ If so, please give any available information:_____
_____

Has there been any x-ray, medication (prescription, over the counter, street drugs), or alcohol exposure in the current pregnancy?_____ If so, give dates of exposure, amount, and name of drugs:_____
_____

Is there any history in your OR the baby's father's family of the following disorders? If so, mark the blank and list details of the problem and relationship of affected individual to yourself at the bottom of the page. Include parents, grandparents, aunts/uncles, brothers/sisters, nieces/nephews, and cousins.

_____ birth defects
_____ down syndrome (mongolism)
_____ mental retardation
_____ infant or childhood deaths
_____ spina bifida (open spine defects)
_____ hydrocephalus (water on the brain)
_____ hemophilia or bleeding disorders
_____ cystic fibrosis
_____ muscle disease (muscular dystrophy)
_____ cleft lip or palate
_____ multiple family members with the same trait/disease ("runs" in the family)
_____ stillbirths in relatives
_____ individuals much taller or shorter than rest of family
_____ individuals who look unusual or very different from rest of family
_____ born with heart defect
_____ blindness or deafness (not age or accident related)
_____ kidney disease
_____ early onset of cancer (under 35 years)
_____ early onset of heart disease (under 35 years)
_____ early onset of emphysema (under 35 years)
_____ multiple miscarriages in relatives (2 or more)

Details of above:_____
_____

Do you have any additional concerns not covered above?_____
_____

PATIENT NAME_____ **MR**_____

IF YOU ARE PREGNANT OR THINK YOU MAY BE PREGNANT,
YOU NEED TO KNOW ABOUT HIV

You need to take care of yourself and get regular checkups for your health and your baby's health. Your healthcare provider will ask you questions and check you for conditions that can harm you and your baby. As part of your routine care, you should have a confidential HIV test

PLEASE INITIAL EACH LINE

WHAT IS HIV?

_____Human Immodeficiency Virus (HIV) is a disease that weakens the immune system, making it hard for the body to fight infections.

HAVE YOU RECENTLY HAD AN HIV TEST?

_____For your health and your baby's, you should know if you're infected with HIV – the virus that causes AIDS, If you are infected, there are things you can do to protect your baby.

_____A confidential HIV test will be performed on every pregnant woman in Texas at the first prenatal visit and at delivery. You may refuse the test, but there are benefits to knowing your HIV status.

_____If you refuse testing, your healthcare provider will let you know about where to get an anonymous test done.

_____A "confidential test" means information about the test results will be written in your medical record. An "anonymous test" means your real name won't be used and the test results won't be written in your medical record.

HOW WILL A TEST HELP ME?

_____If you are infected with HIV; there are medicines that may prevent your baby from becoming infected and help you stay healthier. You will need to start taking the medicine early in your pregnancy.

HOW WILL A TEST HELP MY BABY?

_____The test will help your baby by alerting you to the need for treatment. If you have HIV, you might give it to your baby during pregnancy, at delivery, or by breastfeeding. Without treatment, about one out of every four babies born to HIV-infected mothers are born with HIV.

_____Doctors have learned that the drug AZT can greatly reduce your chances of giving HIV to your baby. You may want to discuss this treatment with your healthcare provider.

HOW CAN I AVOID HIV INFECTION?

_____Abstain from sexual activity. This is the 100% sure way to remain uninfected.

_____Stay in a relationship with one person you know does not inject drugs and is not infected with HIV or other Sexually Transmitted Diseases.

_____Use latex condoms every time you have sex unless you are sure your sex partner is not infected. Condoms are not just for preventing pregnancy. When used correctly, condoms can help prevent diseases like HIV and other Sexually Transmitted Diseases.

_____Do not use drugs! Drugs can hurt you and your unborn baby. If you use drugs, ask about treatment programs to help you stop. If you can't stop, do not share needles or syringes. Be sure to clean needles with water and bleach between uses.

WHERE CAN I GET MEDICAL HELP?

Private doctor's office, Local Health department, Texas Department of Health Regional Clinics;
Call the TEXAS AIDSLINE at 1-800-299-AIDS, to find out about HIV testing and medical services in your area.
Call BABY LOVE HOTLINE at 1-800-422-2956 to receive a referral for medical care for you and your baby.

_____ _____

Signature Date

PATIENT NAME_____ **MR**_____

CONSENT FOR TESTING FOR THE ACQUIRED IMMUNE DEFICIENCY SYNDROME (AIDS)

1. The Acquired Immune Deficiency Syndrome (AIDS) is a viral illness that is spread by contact with the blood or body fluids of an infected person.

2. As part of your obstetrical care, it will be MANDATORY that you be tested to determine if you have had previous contact with this virus. The test for AIDS will be drawn at the time we do your prenatal labwork.

3. The AIDS test sometimes gives a false positive result (the test is positive without the AIDS virus being present). Therefore, a confirmatory test is done on all positive results. It is possible in the early stage of illness (the first few weeks after contact with an infected person) that the test could be negative, even though active infection is present. Especially for individuals in high-risk groups or for their intimate contacts a single negative test cannot establish with certainty that infection is not present.

4. If my blood is found to be positive, I will be notified and provided with information regarding follow-up care.

5. I have had the opportunity to ask questions concerning this blood test and understand that I will be given counseling concerning the meaning of the test result and its implications.

6. I understand that my test result will be kept confidential to the full extent required by law. I understand that particular care is being taken to maintain my records in a secure manner.

7. In consenting to this test, I have read and I understand this information.

I DO CONSENT:

_____ _____

PATIENT WITNESS

_____ _____

DATE PARENT OR GUARDIAN IF MINOR OR OTHER LEGAL IMPEDIMENT TO SIGNING THIS STATEMENT.

HEINRICH SCHETTLER, M.D.
18955 MEMORIAL NORTH, STE 350
HUMBLE, TEXAS 77338
281-446-4644

BILLING CONSENT FOR HIV TESTING

PATIENT NAME:_____

ADDRESS: _____

_____

PLEASE SELECT ONE OPTION BELOW BY INITIALING SPACE PROVIDED:

1._____ I authorize the forwarding of my name, address, birth date, name of test and charges to my insurance company,_____.

2._____ I consent to allow the disclosure of my name, address, birth date, name of test and the charges to Medicaid or other medical assistance programs. Medicaid #_____,

3._____ I do not give my consent to release the name of the test to my insurance company, or medical assistance program. I agree to pay the charges of $30.00 for each HIV test performed.

PATIENT SIGNATURE:_____ DATE:_____

WITNESS SIGNATURE:_____ DATE:_____

PATIENT NAME_____ MR_____

HEINRICH G. SCHETTLER, M.D.
18955 MEMORIAL NORTH STE 350
HUMBLE, TX 77338
281-446-4644

HERPES SIMPLEX VIRUS – HSV 2

Herpes or HSV 2 is an available and optional blood test to see if you have ever been exposed to herpes in your life.

If you are positive for this virus, results will be discussed with you along with precautionary measures taken during pregnancy.

_____ I prefer to have this test

_____ I do not prefer to have this test.

_____ _____
Signature of Patient Date

_____ _____
Witness Date

PATIENT NAME_____ **MR**_____

HEINRICH G. SCHETTLER, M.D.
18955 MEMORIAL NORTH, STE 350
HUMBLE, TEXAS 77338

What should I know about Cystic Fibrosis (CF)?
Cystic fibrosis (CF) is a life-long illness that is usually diagnosed in the first few years of life. The disorder causes problems with breathing and digestion. Cystic fibrosis does not affect intelligence.

A brief review of the genetics of Cystic Fibrosis (CF):
Cystic fibrosis is a genetic disorder. It is caused by changes in a pair of CF genes. All genes come in pairs. One set comes from the mother and the other from the father. Some genes do not function properly because there is a mistake in them. If a gene has a mistake, it is said to be altered or changed.

Everyone has two copies of each gene. For CF to occur, a person has to inherit one altered gene from each parent. If a person inherits one changed copy of a CF gene, that person is a carrier for CF. A carrier does not have CF. There are no known health problems associated with being a carrier. However, when couples are both carriers, any child they have may inherit one changed copy of the gene from each parent. A child with two changed copies of the CF gene will develop CF.

CONSENT FOR CYSTIC FIBROSIS (CF) TESTING

You should be certain you understand the six items listed below. If you are not certain about any of them, please ask Dr. Schettler or one of the nurses to explain them further before signing this form accepting or declining CFcarrier testing.

1. I understand that the decision to be tested for CF carrier status is completely mine.
2. I understand that the test does not detect all CFcarriers.
3. I understand that if I am a carrier, testing the baby's father will help learn more about the chance that my baby could have CF.
4. I understand that if one parent is a carrier and the other is not, it is still possible that the baby will have CF, but that the chance is very small.
5. I understand that if both parents are carriers, additional testing can be done in order to know whether or not the baby will have CF.
6. I understand that if the baby has cystic fibrosis, the only way to avoid the birth of a baby with CF is by terminating the pregnancy.

I have read and I understand the information in this document and:

_____ **I WANT CF carrier testing**

_____ **I DO NOT want CF carrier testing**

_____ _____
Patient Signature Date

PATIENT NAME_____ MR_____

HEINRICH G. SCHETTLER M.D., P.A.
18955 MEMORIAL NORTH, STE 350
HUMBLE, TEXAS 77338

What is spinal muscular atrophy?

Spinal muscular atrophy (SMA) is a genetic disease that results in progressive muscle weakness and paralysis. The condition occurs in 1 in 10,000 live births and affects both males and females.

There are three types of SMA. The most severe type is usually diagnosed within the first few months of life. Affected children have severe muscle weakness and typically do not survive past the age of 2.

The other two types of SMA, which are less common than the severe type, involve a lesser degree of muscle weakness. Most affected individuals need to use wheelchairs or need assistance with walking. Life expectancy for the less severe types ranges from the teenage years to adulthood. Those with the mildest form of SMA are expected to have a normal lifespan.

Is there treatment for spinal muscular atrophy?

There is no cure for SMA. Medical treatments for the milder forms of SMA may help extend the expected lifespan in some cases. The great majority of babies with the most severe form of SMA do not survive past 2 years of age.

How is spinal muscular atrophy inherited?

SMA is a genetic condition. Our genes come in pairs, with one copy inherited from each parent. Some genes have mutations in them, and do not function properly. Someone with one non-functional copy of the gene is a carrier.

Carriers for SMA have no symptoms, but can pass the non-functioning gene on to their children. An individual must inherit two non-functioning SMA genes — one from each parent — to have symptoms of SMA.

If both parents are carriers there is a one in four (25 percent) chance that both will pass on the non-functioning gene, which would result in a pregnancy affected with spinal muscular atrophy.

Who should consider carrier testing for spinal muscular atrophy?

Those who have a family member with SMA or a family member known to be a carrier are at increased risk to be a carrier themselves. Genetic counseling is recommended in these cases to determine the likelihood of having a pregnancy or child affected with SMA.

An individual without a family history of SMA can be a carrier for this condition as well. About 1 in 50 people, regardless of ethnic background, are carriers of the abnormal gene that causes SMA.

How do I know if I am a carrier for spinal muscular atrophy?

Carrier testing is available through a simple blood test. The test can detect the most common mutation that is associated with SMA and will detect approximately 90 percent of carriers. A negative carrier test will greatly reduce the likelihood of having a child affected with this condition.

There are gene changes (mutations) associated with SMA that are not included in the carrier test. In addition, in a small number of cases where a child is found to be affected, only one

parent is a carrier. For both of these reasons, a negative carrier test cannot eliminate the possibility of having an affected child.

What does it mean if I am a carrier for spinal muscular atrophy?

If you are found to be a carrier for SMA, your partner should undergo carrier testing as well. If your partner's test is negative, the chance to have an affected child is low. If your partner is also found to be a carrier for SMA, there is a 1 in 4 (25 percent) chance of an affected pregnancy.

If two parents with no family history of SMA are found to be carriers with routine screening, it is not possible to predict whether they are at risk of having children with the severe form of SMA or one of the less severe forms of SMA.

Is fetal diagnostic testing available for spinal muscular atrophy?

Yes. If both partners are carriers of SMA, prenatal testing is available. You will be referred to a maternal fetal medicine specialist who can perform Chorionic villus sampling (CVS) at 10 to 14 weeks or amniocentesis at 16 to 20 weeks to determine if the fetus has inherited two copies of the SMA gene mutation. If both partners are carriers, you will meet with a genetic counselor to discuss the test results.

CONSENT FOR SPINAL MUSCULAR ATROPHY (SMA) TESTING

You should be certain you understand the five items listed below. If you are not certain about any of them, please ask Dr. Schettler or one of the nurses to explain them further before signing this form accepting or declining SMA carrier testing.

1. I understand that the decision to be tested for SMA carrier status is completely mine.
2. I understand that if I am a carrier, testing the baby's father will help learn more about the chance that my baby could have SMA
3. I understand that if one parent is a carrier and the other is not, it is still possible that the baby will have SMA, but that the chance is very small.
4. I understand that if both parents are carriers, additional testing can be done in order to know whether or not the baby will have SMA
5. I understand that if the baby has spinal muscular atrophy, the only way to avoid the birth of a baby with SMA is by terminating the pregnancy.

I have read and I understand the information in this document and:

_____ I WANT SMA carrier testing

_____ I DO NOT want SMA carrier testing

_____ _____

Patient Signature Date