SRCC CHILDREN'S HOSPITAL

Managed by Narayana Health

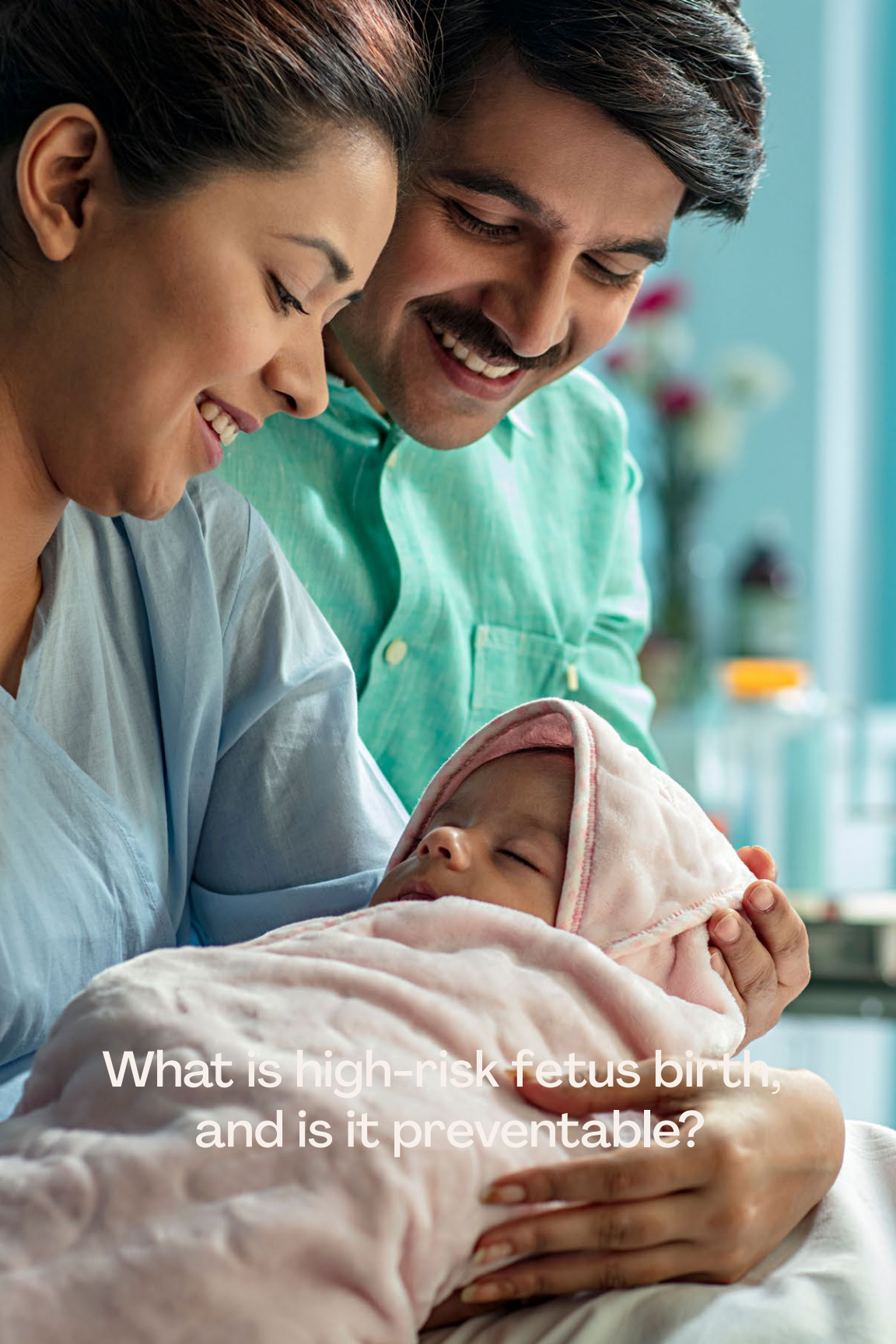
Haji Ali Park
Mumbai

CONTENTS

3.5 million babies are born prematurely in India.*

1.7 million of these babies are born with birth defects every year.*

*https://www.unicef.org/india/what-we-do/newborn-and-child-health



What is high-risk fetus birth, and is it preventable?

WHAT IS A CONGENITAL ANOMALY?

The word congenital means acquired in the womb. A child can have a range of mild to severe medical conditions right from birth. This phenomenon is referred to as Congenital Anomaly or Birth Defect.

While 30-40% of anomalies are related to the parental genetic disorder, the rest have multiple reasons varying from the mother's health, the lifestyle of the would-be parents during the gestational period, or even the environmental factors.

These defects affect mental development, physical development, sensory organs, bodily functions, and a single organ or body part of the child in the womb.

If detected in time, it is possible to address, reduce, or avoid these birth defects. At the germinal or embryonic stage, one can leverage medical technology for the same.

RATE OF CONGENITAL ANOMALIES IN INDIA & AROUND THE WORLD

According to the WHO (World Health Organization) report, Birth defects are significant causes of newborn and childhood deaths, chronic illness, and disability.

An estimated 303000 newborns die yearly due to birth defects – out of which an estimated 90000 deaths occur in South-East Asia. In addition to mortality, birth defects cause long-term disability, significantly impacting individuals, families, healthcare systems, and societies.

According to a joint WHO and MOD (March of Dimes) meeting report in India, birth defects prevalence varies from 61 to 69.9 per 1000 live births.

TYPES OF BIRTH DEFECTS

There are two types of Birth Defects - Structural or Developmental.

An anomaly affecting a body part is known as a Structural anomaly. However, it is a developmental anomaly involving bodily functions or sensory organs' work.

Here are some of the most common birth defects in India:

STRUCTURAL ANOMALIES

HEART DEFECTS

Heart Anomalies are the most common birth defects. 8 to 10 per 1000 live babies have congenital heart disease*. If the heart doesn't form properly in the womb, it leads to a heart anomaly, and the affected area of the heart determines the type of defect. The most common heart anomaly is a ventricular septal defect, where a hole in the wall separates the two lower heart chambers. The child must undergo surgery soon after birth for a severe heart defect.

NEURAL TUBE DEFECTS

The neural tube forms the early brain and spine. If it does not close properly, then the deformity occurs which is known as Spina Bifida. Encephalocele, and Anencephaly are some of the Neural Tube Defects. Neural Tube Defects affect 4 to 11 babies per 1000 live births in India.*

LIMB DEFECTS

When a part of a limb is short or doesn't fully form in the womb, it is known as Limb Defects. These are all related to a missing finger, hand, arm, clubfoot, or an arm shorter than usual.

GUT ANOMALIES

Diaphragmatic Hernia, Omphalocele, and Gastroschisis are some of the common types of Gut and Stomach Anomalies. Most Gut and Stomach Anomalies occur in the case of the stomach not fully forming or forming by leaving intestines or other organs outside the body.

CLEFT LIP OR PALATE

A cleft lip or palate occurs when the tissues that form the roof of the mouth or lip do not join or close properly. Being one of the common birth defects, it causes a hindrance in speech, hearing, and eating. Cleft lip or palate can be easily fixed by surgical intervention.

Source - https://www.unicef.org/india/what-we-do/newborn-and-child-health)

DEVELOPMENTAL ISSUES

DOWN'S SYNDROME

An extra copy of Chromosome 21 causes Down's syndrome. Down's syndrome affects the body and brain development like heart defects, blood disorders, and the immunity system. It is one of the most common Chromosomal anomalies worldwide because it has a life expectancy of ~ 40 years.

VISUAL IMPAIRMENTS

An irregular eye shape or a dysfunctional connection between the brain and eyes can lead to Visual Impairment.

HEARING IMPAIRMENTS

One of the most common anomalies, hearing impairment in newborns, results from cytomegalovirus infection or genetic defects.

FETAL ALCOHOL SPECTRUM DISORDERS

A child's learning, growth, and development can be directly affected if the mother drinks alcohol during pregnancy. All the three trimesters of pregnancy are affected by heavy alcohol intake.

CEREBRAL PALSY

Cerebral Palsy is the most common motor disability affecting balance, movement, and posture. It is caused by abnormal brain development or damage to the developing brain. This usually happens before a child is born but can occur at birth or early infancy.

MUSCULAR DYSTROPHY

This anomaly makes muscles weaker over a period of time. They can impact any muscle group or type. They are mostly genetic in origin.

GENETIC DISORDERS

Genetic disorders include chronic diseases, developmental problems, and sensory deficits inherited from one or both parents.

LANGUAGE IMPAIRMENT

Congenital dysphasia is a developmental speech disorder present from birth and characterized by difficulty speaking or understanding spoken words. This is however detected at a later age in childhood once the child starts speaking.

CAUSES OF CONGENITAL DISORDERS

NOISE POLLUTION

Noise pollution, including occupational and ambient noise, can affect the child at the fetal/ neonatal stage. Exposure to noise pollution may result in building up stress and related sleep disturbance. Stress has been hypothesized to affect fetal growth through the endocrine system by causing hormonal imbalances. Moreover, there is evidence for an increased risk of hypertension in women exposed to noise pollution, resulting in poor pregnancy outcomes.

AIR POLLUTION

Exposure to air pollution in the first trimester increases the risk of preeclampsia and high blood pressure. These complications can harm the mother and the baby, which may necessitate premature delivery. It can also cause adverse effects like higher infant mortality, lower birth weight, impaired lung development, increased later respiratory morbidity, and early alterations in immune development.

NUTRITIONAL DEFICIENCY

Nutritional deficiency due to poor and insufficient eating habits lead to extra strain on the body that causes more complications for both the mother and child.

CHROMOSOMAL DISORDER

Typically, humans have 46 chromosomes arranged in 23 pairs. Any variation from this pattern causes abnormalities resulting in chromosomal disorders or malfunction in the baby.

GENETIC DISORDER

Genetic disorders are caused due to genes passed down from parents to children. Sickle Cell Disease, Cystic Fibrosis, heart defects, Neural Tube Defects, blood disorders, etc., can be caused due to genetic changes or mutations.

PRE-EXISTING MEDICAL CONDITIONS

While every pregnancy has risks, some factors such as age, weight, and pre-existing medical conditions can lead to an increased risk of complications. Some pre-existing medical conditions that can make you more susceptible to complications during pregnancy are:

HIGH BLOOD PRESSURE

Pregnant women with chronic high blood pressure are at an increased risk for a low birth weight infant, preterm delivery, kidney damage, and preeclampsia during pregnancy.

AUTOIMMUNE DISEASE

An autoimmune disease is a condition in which the body attacks and damages its tissues, causing susceptibility to infections. The fetus may develop a slow heart rate, low platelet count, low white blood cell count, & anemia. Common autoimmune diseases include Lupus, Rheumatoid Arthritis, Immune Thrombocytopenia, etc.

THYROID

Uncontrolled hyperthyroidism (overactive thyroid) or hypothyroidism (underactive thyroid) may lead to preterm birth, respiratory distress, low birth weight, and even heart failure in the newborn baby.

ASTHMA

Asthma during pregnancy may be dangerous for the fetus because it can hinder blood flow and oxygen to the placenta. Preeclampsia, restricted growth, low birth weight, and premature birth are more likely.

POLYCYSTIC OVARY SYNDROME (PCOS)

It is a common hormonal condition in women where the ovaries produce higher than normal levels of male hormones (androgens). Women with PCOS are more prone to giving birth to a child with Autism. Besides this, PCOS increases the risk of complications, including preeclampsia (hypertension-related disorder) or Gestational Diabetes Mellitus - a dangerous condition for both mother and baby-to-be.

DIABETES

New born baby of a diabetic mother may develop Hypoglycemia (fluctuations in blood glucose level), respiratory distress (difficulty in breathing), Macrosomia (combination of high blood glucose levels from the mother and high insulin levels in the fetus that results in large deposits of fat causing the fetus to grow excessively large) following a birth injury.

UNDERWEIGHT

Babies born to underweight mothers are at higher risk for premature birth, and the newborn baby may also have impaired intellectual development.

OBESITY

If the expecting mother is obese, the fetus is at risk for stillbirth and congenital anomalies.

EPILEPSY

It is a central nervous system (neurological) disorder in which brain activity becomes abnormal, causing seizures or periods of unusual behavior, sensations, and sometimes loss of awareness. If the expecting mother has epilepsy, it leads to a slow heart rate, decreased oxygen, low birth weight of the fetus, preterm labor, and premature birth.

ABNORMALITIES ASSOCIATED WITH REPRODUCTION

There is a possibility of miscarriage, an abnormally positioned fetus, and difficult labor when there are structural problems in the uterus or cervix.

KIDNEY DISEASE

Expecting mothers with kidney disease may experience severe hypertension and anemia, affecting the blood flow to the fetus. The baby born is also at an increased risk of metabolic bone diseases and gestational diabetes.

UTERINE FIBROIDS

Fibroids are benign tumors that grow in or on the uterus or womb. Due to spatial shortage, large fibroids may prevent a fetus from growing fully inside a mother's womb. Cramps due to fibroids lead to uterine contractions resulting in early premature delivery.

OTHER FACTORS

TERATOGENIC DRUGS

Teratogenic drugs are drugs that can cause birth defects. A teratogen is a substance that interferes with the normal development of a fetus.

AGE

The risk of pregnancy complications is higher for women over 35.

INFECTION

Infections in expecting mothers can cause complications such as hearing loss, visual impairment, or blindness, as well as learning difficulties and epilepsy in the newborn baby.

ALCOHOL

Prenatal alcohol exposure can cause ailments for the child such as heart, kidney, bone-related problems, and other malformations, such as sight and auditory issues, along with reduced immune function. Fathers who drink alcohol regularly before conception have greater chances of having a child with birth defects like congenital heart disease, limb anomalies, clefts, and digestive tract anomalies.

PROLONGED HYPER THERMIA

The term hyperthermia refers to an abnormally high body temperature. Humans generally have a body temperature of about 98.6°F (37°C). The body's temperature exceeding 101° F (38.3°C) during pregnancy is detrimental to the fetus. Furthermore, studies have shown an increased risk of neural tube defects (NTD) in babies of women who had prolonged high temperatures during pregnancy. It can also lead to premature labour pains and preterm delivery.

MULTIPLE PREGNANCIES

A pregnant woman is more vulnerable to abnormally premature labor and uncontrollable blood loss if she has had five or more pregnancies.

MULTIPLE-BIRTH PREGNANCIES

The womb becomes crowded when multiple babies are growing in the womb, resulting in complications. Premature births are more likely to occur with multiple fetuses because of limited space and extra burden on the mother. Multiple pregnancies are more likely to result in pregnancy complications, such as high blood pressure and diabetes. Most important risk is premature labour pains and preterm delivery and risk increases with every increase in number of fetuses.

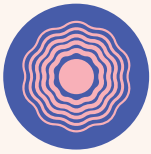
COMPLICATIONS ASSOCIATED WITH PREVIOUS PREGNANCIES

Previous pregnancy complications may increase one's chances of having the same difficulty in the following pregnancy. The possibility of preterm birth or stillbirth and genetic or chromosomal defects is relatively high.

SEXUALLY TRANSMITTED INFECTIONS (STIS)

There is a potential risk that STIs will be transmitted to a baby if a woman has one. A baby born to a woman with an STI is at higher risk for:
low birth weight, conjunctivitis, pneumonia, neonatal sepsis (infection in the baby's bloodstream), neurologic damage, blindness, deafness, acute hepatitis, meningitis, chronic liver disease, cirrhosis.

SYMPTOMS OF HIGH-RISK FETUS

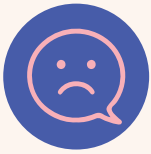


PAINS OR CRAMPS
IN LOWER ABDOMEN

VAGINAL BLEEDING
OR WATERY DISCHARGE

CHANGES IN VISION,
BLURRED VISION

SEVERE
HEADACHES



THOUGHTS OF
HARMING YOURSELF
OR YOUR BABY

SUDDEN OR SEVERE
SWELLING IN THE FACE,
HANDS OR FINGERS

VOMITING OR
PERSISTENT NAUSEA

FEVER OR
CHILLS

DECREASED
FETAL ACTIVITY

DIZZINESS

WHAT IS PRE & PERI CONCEPTIONAL COUNSELING?

Preconception is the period before pregnancy. Preconception, on the other hand, is the period from when oocyte growth, fertilization, conceptus formation, and development start taking place until week 10 of pregnancy. Maternal and neonatal health outcomes have been seen to be impacted majorly by these two significant periods.

Regarding pregnancy readiness, the appropriateness of family planning and having a healthy baby cannot be overstated. An unplanned or unintended pregnancy results in delayed prenatal care, which may lead to adverse pregnancy outcomes.

OBJECTIVES O PRECONCEPTIONAL COUNSELLING

1. To reduce the chances of disorders such as Down's Syndrome and Autosomal Dominant Disorders due to new dominant mutations related to advanced parental age.

2. To minimize the occurrence of Congenital Abnormalities such as Neural Tube Defects related to folate deficiency and Mental Defects because of iodine deficiency in would-be mothers.

3. To curtail Congenital Abnormalities and Stillbirths by more robust management of maternal diabetes before and during pregnancy.

4. To avoid the danger of hereditary disorders in high-risk families.

5. To abet the possibilities of miscarriages and Fetal Growth Retardation.

6. To promote the importance of a healthy lifestyle and work-life balance during pregnancy.

7. To spread awareness on carriers of hemoglobinopathies and G6PD deficiencies and the impact and availability of carrier testing.

8. To put a stop to Congenital Rubella Syndrome through immunization.

9. To promote the importance of a healthy lifestyle and work-life balance during pregnancy.

FIRST TRIMESTER

The first-trimester screening includes pregnancy tests completed between Weeks 11 and 13. Certain birth defects related to the baby's heart or chromosomal disorders, such as Down's syndrome, can be detected with this test. A maternal blood test and ultrasound are included in this screening.

MATERNAL BLOOD SCREEN

It is a simple blood test to measure the levels of two proteins, human chorionic gonadotropin (hCG) and pregnancy-associated plasma protein A (PAPP-A). The abnormally high or low protein level is indicative of a chromosomal disorder.

ULTRASOUND

During pregnancy, an ultrasound forms images of the baby for the physician. The baby's neck is majorly examined in the first-trimester screen look. If the ultrasound detects an excess of fluid, the baby could have a chromosomal disorder or heart defect.

SECOND TRIMESTER

In the second trimester, screening tests are conducted between Weeks 15 and 20 of pregnancy to examine specific birth defects in babies. This ultrasound consists of a maternal serum screen and a comprehensive ultrasound evaluation of the baby, looking for the presence of structural anomalies (also known as an anomaly ultrasound).

MATERNAL SERUM SCREEN

The maternal serum screen is a simple blood test used to identify if a woman is at increased risk for having a baby with certain birth defects, such as Neural Tube Defects or Chromosomal Disorders like Down's syndrome. It is also known as a "triple screen" or "quad screen," depending on the number of proteins measured in the mother's blood. For example, a quadruple screen tests 4 proteins AFP (alpha-fetoprotein) levels, hCG, estriol, and inhibin-A. Generally, the maternal serum screen is completed during the second trimester.

ANOMALY ULTRASOUND

An anomaly ultrasound is a test conducted around weeks 18 to 20 of pregnancy. It provides an image of the baby in the womb and is used to look for birth defects and other problems.

FETAL ECHO CARDIOGRAM

A fetal echocardiogram test uses sound waves to scan for heart defects. The echocardiogram provides detailed images of the baby's heart, better than regular pregnancy ultrasounds. But some heart defects cannot be detected before birth, even with a fetal echocardiogram. A detailed ultrasound may be done to look for other problems with the developing baby if your healthcare provider finds a problem in the structure of the baby's heart.

HIGH RESOLUTION ULTRASOUND

More detailed than an anomaly ultrasound, high-resolution ultrasound provides clear images of the baby in the womb to identify problems related to possible birth defects. It is usually done between 18 and 22 weeks of pregnancy.

DIAGNOSTIC TESTS

If the screening test results are abnormal, the treating physician recommends further diagnostic tests to determine if any birth defect or other possible anomaly with the baby is present. Women who might be facing riskier pregnancies are prescribed further diagnostic tests. Women older than 35, or women who have had previous pregnancies, one suffering from chronic diseases like high blood pressure, diabetes, or women on specific medication can also be advised to go for diagnostic tests.

CHORIONIC VILLUS SAMPLING (CVS)

To test for chromosomal or genetic disorders, a Chorionic Villus Sampling is done. A sample of the placenta is tested to check for disorders. This is prescribed to women who have had an abnormal first-trimester screening test. It is done between 11 and 13 weeks of pregnancy, usually before amniocentesis.

AMNIOCENTESIS

It is a prenatal test that diagnoses genetic disorders (such as Down's syndrome and Spina bifida) and other health issues in the fetus. A provider uses a needle to remove a small amount of amniotic fluid inside one's uterus. Generally, amniocentesis is prescribed in higher risk pregnancies or for women who have received an abnormal screening test and is usually conducted between 15 and 24 weeks.

Below are some of the biochemical proteins for which amniocentesis tests are conducted:

ALPHA-FETOPROTEIN

AFP stands for alpha-fetoprotein, a protein the fetus produces. A high level of AFP in the amniotic fluid might mean that the baby has a defect indicating an opening in the tissue, such as a neural tube defect (anencephaly or spina bifida), or a body wall defect, such as omphalocele or gastroschisis.

ACETYLCHOLINESTERASE

AChE stands for acetylcholinesterase, an enzyme that the fetus produces. This enzyme can pass from the fetus to the fluid surrounding it if there is an opening in the neural tube.

GENETIC COUNSELLING

WHY GENETIC COUNSELING?

Genetic Counselling is critical for the would-be parents to have some information on how genetic illness or defect in either or both of them and their families holds the potential risk to their offspring's health.

A genetic counselor examines the family health history to learn more about genes, chromosomes, and other maternal and paternal health aspects that can affect the fetus. A genetic counselor thus helps you identify the causes of genetic disorders, would-be mothers' chances of delivering a baby with a genetic condition/s, and the available forms of testing to identify one.

WHAT TESTS CAN BE DONE BEFORE PREGNANCY TO DETERMINE THE GENETIC CONDITIONS THAT MAY AFFECT THE FETUS?

An expecting mother can undergo carrier screening tests that check her blood, saliva, or tissue. This helps to determine if she is a carrier of certain genetic conditions. As a carrier, even if you may not have the condition yourself, you may still have a gene change for it that may potentially be carried forward to the offspring.

The partner can get tested for this as well. If both parents are carriers of the same condition, then the risk of it being passed on to the baby intensifies. Thus, getting tested before conceiving can help the parents look at the probable risks if any, so that the right decisions can be taken to avoid any future mishaps.

WOMEN WHO ARE CONSIDERING CONCEIVING CAN UNDERGO CARRIER SCREENING FOR THE FOLLOWING:

Cystic Fibrosis: In this condition, one's breath and digestion get affected.

Spinal Muscular Atrophy: This is a group of conditions that causes muscle damage and weakness.

Hemoglobinopathies: This condition directly affects the red blood cells in one's body.

There are two types of hemoglobinopathies:

Thalassemias: In this blood condition, the body makes fewer healthy red blood cells and less hemoglobin than average.

Sickle Cell Disease: In this blood condition, red blood cells are caused to be shaped like a "C."

TARGETED CARRIER SCREENING

Carrier screening can be done for several reasons, but Targeted Carrier Screening is done for conditions based on one's family history.

THE SCREENING INCLUDES:

FRAGILE X SYNDROME

If Fragile X syndrome runs in your family, this carrier testing is recommended. Fragile X Syndrome is a genetic disorder causing changes in a gene called Fragile X Messenger Ribonucleoprotein 1 (FMR1), responsible for making a protein called FMRP needed for brain development.

PREIMPLANTATION GENETIC TESTING

If testing shows that the parents are carriers of a genetic condition and have had fertility treatments (IVF), then preimplantation genetic testing is recommended. This test checks the embryo for gene changes before they are placed into the uterus. In such a situation, the IVF provider can choose embryos that do not test positive for gene changes. A counselor can rightly guide you based on test results and make you understand the chances of affecting the offspring.

WHAT ARE THE BENEFITS OF TESTING?

The normal result assures you that your baby is not at risk for any genetic deformity conditions.

If it is found out that your baby has a condition, you can initiate an early treatment which is available during pregnancy or post-birth.

If your baby is identified with any such condition, it will make you take the right decision regarding an ideal hospital with the best providers and equipment for good care.

WHAT ARE THE RISKS OF TESTING?

Since many tests just use saliva or blood samples, the physical risks of testing are small. Diagnostic tests such as amino and CVS have slight chances of miscarriage since they require a sample of fluid / tissue around the baby.

WHAT PROBLEMS CAN GENETIC CONDITIONS CAUSE DURING AND AFTER PREGNANCY?

Genetic conditions may result in stillbirth or miscarriage. This is when a baby dies in the mother's womb before 20 weeks of pregnancy. Chromosomal disorders cause more than 50% of these miscarriages, and sometimes babies with these genetic conditions barely survive after a long birth.

Every child who is born with a genetic condition is unique. The child's problems depend on which genes or chromosomes are affected. However, some children do not have serious issues. Some children may have intellectual or developmental disabilities or birth defects or all.

SRCC CHILDREN'S HOSPITAL

Managed by Narayana Health

Haji Ali Park
Mumbai

Appointments
1800-309-0309 (Toll Free)

Emergencies
022-71222333

1A, Haji Ali Park, Mahalaxmi, Mumbai
www.narayanahealth.org