## About BabyShield

BabyShield is India's comprehensive provider of preventive healthcare services with a focus on care for the health and well being of babies and mothers, right from the time of pregnancy. BabyShield presents a complete panel of pre & post-natal diagnostic tests including maternal infectious diseases test, prenatal screening, first milk test and newborn screening test. Comprehensive panels, expeditious reporting and post diagnostic guidance make BabyShield a trusted partner for mother and child care during and after pregnancy.

## Stop Guessing Your Baby's Health!
## Get Answers With BabyShield Prenatal Testing


### References

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Disclaimer : All the information provided in this document is purely intended to help you have a stress free pregnancy and prepare you for parenthood. The information contained herein cannot be considered as a medical opinion. Please seek a direct opinion of your doctor in connection with any queries or issues that you may have regarding your baby's health.

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# Prenatal Screening

## Ensure The Health Of Your Baby Even Before Birth

## PREGNANCY IS A PERIOD OF BLISSFUL MOMENTS AND PREPARING FOR PARENTHOOD RESPONSIBILITIES.

During pregnancy, it is important to know the baby's health inside the womb. Hence various tests are recommended for you to ensure that your baby is born healthy. Prenatal chromosomal screening gives valuable information about your unborn baby's health and helps you understand the risk of any birth abnormalities.

## cfDNA Vs QF-PCR

| | cfDNA | QF-PCR |
|---|---|---|
| Safety | Non-invasive sample collection and hence it is safe | Invasive sample collection. Poses a small risk of miscarriage. |
| Efficiency | Detection rate - 99.44% & False positive rate – 0.03% [7] (For Trisomy 21) | Detection rate – 100% & False positive rate – 0% [8] (For Trisomy 21) Better accuracy than cfDNA results |
| Conditions Detected | • Down Syndrome<br>• Edwards Syndrome<br>• Patau Syndrome<br>• Turner Syndrome<br>• Triple X Syndrome<br>• Klinefelter Syndrome<br>• XYY Syndrome | • Down Syndrome<br>• Edwards Syndrome<br>• Patau Syndrome<br>• Turner Syndrome<br>• Triple X Syndrome<br>• Klinefelter Syndrome<br>• XYY Syndrome<br>• DiGeorge Syndrome<br>• Williams-Beuren Syndrome<br>• Smith-Magenis Syndrome<br>• Langer-Giedion Syndrome<br>• Miller-Dieker Syndrome<br>• Potocki Lupski Syndrome |

**High Risk:** If the first trimester chromosomal screening test indicates high-risk, your doctor may recommend you to take non-targeted Karyotyping test that does not limit to a particular set of conditions but checks for all microscopically visible chromosomal conditions that may or may not have serious health consequences for the baby.

Karyotyping is the gold standard methodology for detection of chromosomal conditions in unborn babies. It is performed by observing the number, size, and structural changes of chromosomes present in the sample (amniotic fluid or placental fluid) collected from the mother.

*It is important to understand that a high-risk test result does not equate to the presence of an abnormality.*

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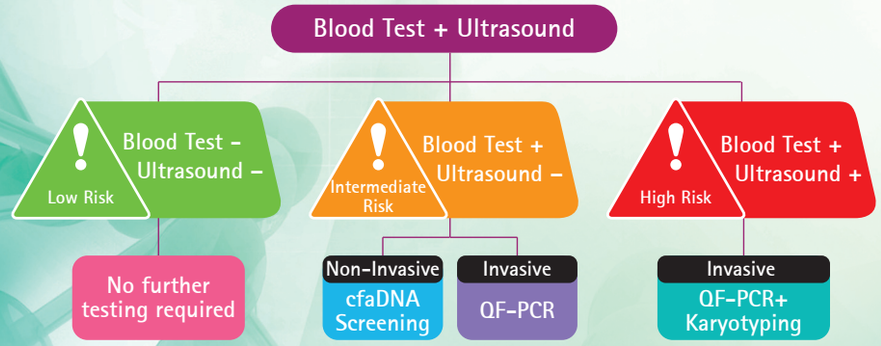
## What Do The Test Results Mean?

**The result is based on the combination of blood test and ultrasound scan which gives 3 possibilities: Low Risk, Intermediate Risk, and High Risk.**




**Low Risk:** If the result shows low risk, your doctor may suggest you to avoid further screening/diagnostic tests. However, this does not mean there is no risk of chromosomal conditions.

**Intermediate Risk:** If the result indicates intermediate risk for chromosomal conditions, your doctor may suggest follow-on tests which aim to detect not all but only certain targeted (7 - 12) conditions, which collectively represent 99.9% of clinically relevant chromosomal conditions seen in unborn babies at this risk level:

**Cell Free DNA (cfDNA) Test** – For those who wish to avoid an invasive procedure may opt for cfDNA test which provides very high detection rates **(>99% for Down's syndrome)** with only a small aliquot of blood. It however does not detect microdeletions or duplications and also may require further invasive testing to confirm a positive diagnosis

**Quantitative Fluorosense PCR (QFPCR) Test:** This is a rapid diagnostic test which provides quick results with **100% detection rate and 0% false positive rate** detecting whole chromosomal aneuploidies (Trisomy 21, 18, 13, sex chromosome aneuploidies) as well as several clinically relevant microdeletions and duplications. However samples used for testing require an invasive collection procedure which may carry a **0.5% risk of miscarriage.**

## What is Prenatal Chromosomal Screening?

Prenatal chromosomal screening is a safe and simple screening test performed in pregnant mothers to assess the risk of chromosomal conditions in the baby inside the womb. It is usually performed in the first trimester and requires only a small amount of mother's blood for the screening process and hence safe for the mother and the baby.

## Why Is This Screening Recommended?

About **1.3 lakh** babies are born with certain chromosomal abnormalities in India every year[1] [2], the highest in the world. Every year, about **23,000 - 29,000** babies are born with Down syndrome[3] a condition which affects the normal growth and development of the baby. A prenatal chromosomal screening is a safe way to check for chromosomal conditions in babies before birth and it helps the parents to make an informed decision on the continuance of pregnancy. Thus, it is **globally recommended**[4] for all pregnant women.

### Global Recommendations Include

ACOG
THE AMERICAN CONGRESS OF OBSTETRICIANS AND GYNECOLOGISTS

THE SOCIETY OF OBSTETRICIANS AND GYNAECOLOGISTS OF CANADA

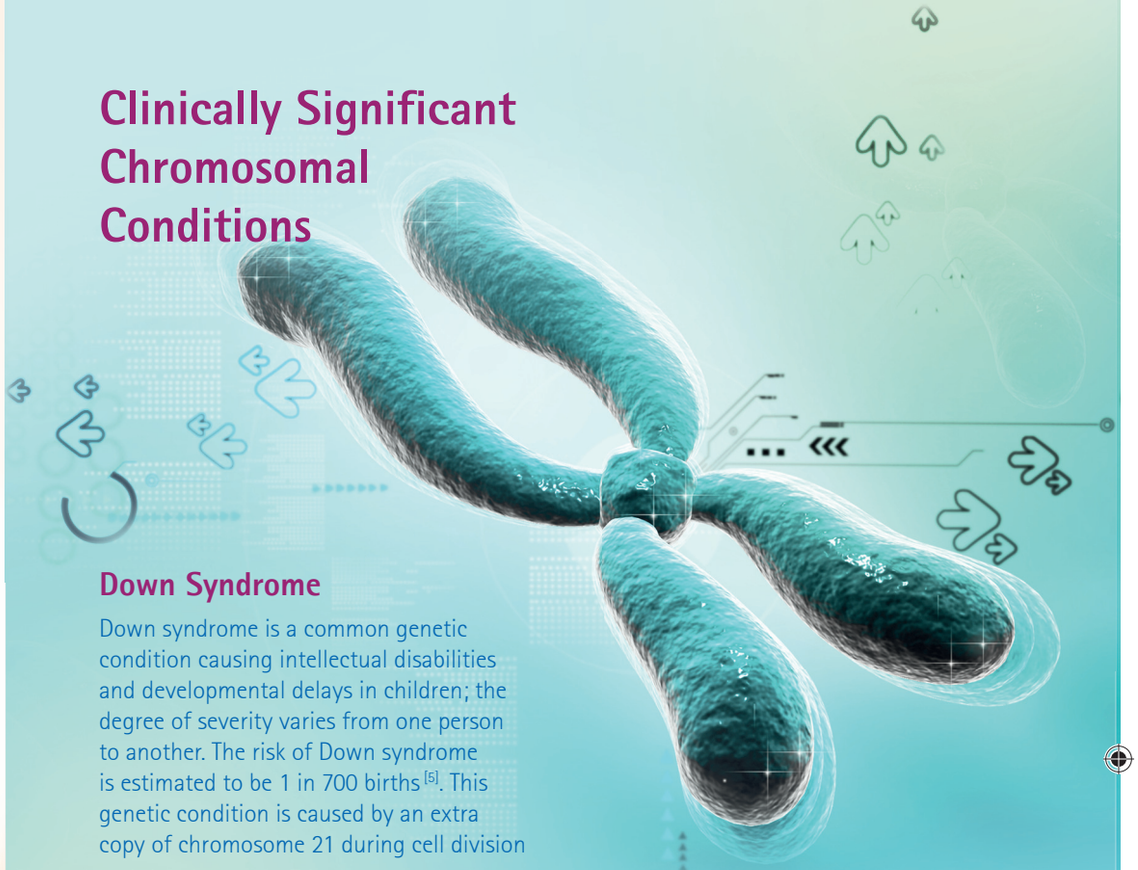
Perinatal Services BC
An agency of the Provincial Health Services Authority

## How Does Prenatal Screening Help?

1. If the screening results are normal, it will ease the anxiety of the expectant parents and give them the comfort of their baby's normal health.

2. It helps the pregnant couple to prepare and make an informed decision; based on further diagnostic tests, they can decide on the continuance of pregnancy.

# Clinically Significant Chromosomal Conditions



## Down Syndrome

Down syndrome is a common genetic condition causing intellectual disabilities and developmental delays in children; the degree of severity varies from one person to another. The risk of Down syndrome is estimated to be 1 in 700 births[5]. This genetic condition is caused by an extra copy of chromosome 21 during cell division and thus known as Trisomy 21.

## Edwards and Patau Syndrome

Babies born with Edwards or Patau syndrome may experience congenital heart disease, brain abnormalities, growth problems and abnormal facial structures. Mostly, these medical conditions result in miscarriage or still-born. Edwards syndrome (Trisomy 18) is the second most common chromosomal abnormality and the risk is estimated to be 1 in 2500 pregnancies[6]. It is caused by an extra copy of chromosome 18 while Patau syndrome (Trisomy 13) is caused by an extra copy of chromosome 13 formed during cell division.

## How Is Prenatal Chromosomal Screening Done?

The screening is performed in the first trimester between 9 weeks and 13 weeks, 6 days of pregnancy. A small amount of blood is drawn from the pregnant mother and tested for proteins produced by the baby inside the womb. An ultrasound test is also optionally performed to improve the accuracy of the test.

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