Frequently Asked Questions

Q **Do I still have to take this test even though my previous child did not have Down syndrome?**

The occurrence of Down syndrome is generally not related to genetics, but a spontaneous chromosomal aberration. Indeed, fewer than 5% of Down syndrome cases are caused by genetic factors. Therefore, we strongly suggest screening for Down syndrome during every pregnancy.

Q **If the result indicates low risk, does that mean my baby will not have Down syndrome?**

The screening test for Down syndrome is merely a risk assessment. No matter which type of screening test you undergo (i.e. ultrasound or serum screening), there is still a 10~20% chance that Down syndrome will occur. We therefore suggest that you continue to undergo regular checkups.

Q **What should I do if my test result indicates that my baby is at high risk of Down syndrome?**

The screening test for Down syndrome is a risk assessment that helps pregnant mothers determine whether an amniotic fluid examination is needed. A higher risk value does not necessarily reflect chromosomal abnormalities in the fetus, so there is no need to become overly panicked. Any decisions regarding additional examination procedures should still be made based on your doctor's advice.

Choosing a Down Syndrome Screening Test

The most significant differences between Down syndrome babies and other babies are related to physical appearance. However, intellectual disabilities and a lifelong inability to live independently are often more worrisome for parents. To prevent giving birth to a baby with Down syndrome, we strongly recommend that you take a Down syndrome screening test. If you have any questions, please consult your attending physician.

There are currently a number of Down syndrome screening methods to choose from. Please consult with your physician to determine which method is the most suitable for you:

	First Trimester Maternal Blood Down Syndrome Screening Test	**New** Second Trimester Maternal Serum Quad Screening Test
Stage in which screening is performed	8~13^{+6} weeks (Best at 12 weeks)	15~20 weeks
Test indicator	• PAPP-A • free β-hCG • Nuchal translucency (NT)	• free β-hCG • AFP • uE3 • inhibin A
Detection rate (5% false positive)	82~87%	83%

References: 1. *Obstet Gynecol.* 2007 Jan; 109(1):217-27. 2. *J Med Screen.* 2003;10(2):56-104.

Understanding the Results of a Down Syndrome Screening Test

Risk value ≧1/270: high risk

This result indicates that the fetus has a high risk of Down syndrome, and further testing is recommended (e.g. Non-Invasive Prenatal Screening (NIPS) or amniotic fluid examination).

Risk value <1/270: low risk

This result indicates that the fetus has a low risk of Down syndrome; however, regular checkups are still recommended to ensure fetal health.

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Maternal Serum Screening for Down Syndrome

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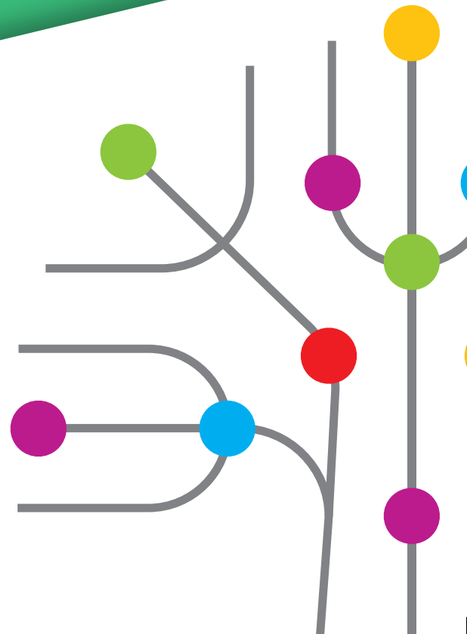


The Most Common Genetic Disorder to Affect Babies

Down syndrome is the most common chromosomal abnormality and one of the primary causes of intellectual disability. Humans have 23 pairs of chromosomes, including 22 pairs of autosomes and 1 pair of sex chromosomes (which determines gender). Any changes in these chromosome numbers result in fetal abnormalities. Babies affected by Down syndrome (approximately 1 in 800~1000 newborns) presented an extra third copy on their chromosome 21.

Individuals with Down syndrome look different from normal individuals and may also suffer from intellectual disabilities, developmental delays, low muscle tone, muscle weakness, and congenital heart disease, conditions that described above are likely to require long-term care.

Babies with Down syndrome have an extra copy of chromosome 21

Mother's Age and Down Syndrome

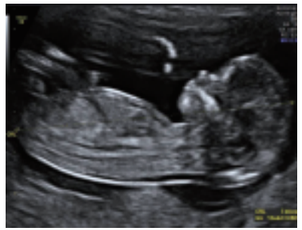
Every pregnant mother is at risk of giving birth to a baby with Down syndrome; however, this risk increases with maternal age. Although the risk that a young mother will give birth to a baby with Down syndrome is lower, but younger mothers account for the majority of the birthing population, therefore, younger mothers should take more awareness to prenatal checkups. To ensure a healthy baby, we recommend that all pregnant women choose one of the following screening tests based on their personal needs regardless of age.

First Trimester Maternal Blood Down Syndrome Screening Test + Nuchal Translucency

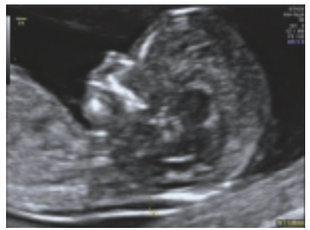
The First Trimester test is taken at 8~13^{+6} weeks of gestation. The risk of Down syndrome in the baby is estimated based on the levels of PAPP-A[1] and free β-hCG[2] in the mother's blood as well as on other soft markers (maternal age, weight, and medical history).

To increase the accuracy of results, we recommend including ultrasound measurements (at 11~13^{+6} weeks) of crown-rump length (CRL) and nuchal translucency (NT) as part of screening. The latter refers to the gap between the skin of the fetus and the underlying tissue at the nape of the neck. This gap is significantly larger in babies with Down syndrome. When ultrasound measurements are combined with serum screening, the detection rate of Down syndrome is 82~87%.

The First Trimester Maternal Blood Screening Test provides pregnant mothers not only assesses the risk that their baby will suffer from Down syndrome, but also examines the risk of Edwards syndrome[3] or Patau syndrome[4] at the same time. A value of less than 1/270 indicates low risk, whereas a value equal to or greater than 1/270 indicates high risk. For high risk cases, further examinations are recommended in order to make a more conclusive determination as to whether the baby has Edwards syndrome or Patau syndrome.



Ultrasound CRL measurement



Ultrasound NT measurement

New Second Trimester Maternal Serum Quad Screening Test

To enhance the quality of Down syndrome screening for pregnant mothers, SOFIVA has completely replaced β-hCG with free β-hCG, which increases the detection rate of the Quad Screening of Down syndrome. The New Second Trimester Maternal Serum Quad Screening Test detects the levels of free β-hCG, AFP[5], uE3[6], and inhibin A in venous blood from the mother at 15-20 weeks of pregnancy and combines this data with other clinical parameters to assess the risk of Down syndrome in the baby. The detection rate of this screening procedure can reach up to 83%!

In addition to providing risk values for Down syndrome, the New Second Trimester Maternal Serum Quad Screening Test also determines risk values for Edwards syndrome and neural tube defects (NTD)[7]. For Edwards syndrome, a value less than 1/270 indicates low risk, and a value equal to or greater than 1/270 indicates high risk. For NTD, a value less than 2.5 indicates low risk, while a value equal to or greater than 2.5 indicates high risk. If a screening test result shows high risk for any of these conditions, we recommend further examinations in order to promote health in the baby.

Note 1: PAPP-A – pregnancy-associated plasma protein A

Note 2: freeβ-hCG – free β-subunit of human chorionic gonadotropin

Note 3: Edwards syndrome – Also known as trisomy 18, Edwards syndrome has an incidence rate of 1/8,000. Affected babies have a very low survival rate. Approximately two thirds are stillborn, and of those that survive birth, 50% die within two months and 90% die within a year.

Note 4: Patau syndrome – Also known as trisomy 13, Patau syndrome has an incidence rate of 1/5,000 and is highly fatal. Affected babies are severely deformed, suffer from heart disease, and have abnormally developed brains, faces, kidneys, limbs, and intestines.

Note 5: AFP – Alpha-fetoprotein

Note 6: uE3 – unconjugated estriol

Note 7: The main cause of NTD are disorders in the neural tube during embryo development, which can lead to congenital malformations such as anencephaly, spina bifida, and encephalocele. Infants with anencephaly usually die shortly after birth. Conversely, infants with spina bifida can survive; mild cases are affected by gastrointestinal or bladder disorders, but severe cases may suffer from lower body paralysis.