IS MY BABY GOING TO BE FINE?

Pregnancy is a joyous milestone in many couple's relationship, but sometimes, the news is swiftly followed by worry. An expecting mother whose previous two children have a genetic disorder, for example, may want to know whether the child she is carrying is healthy.

This month, consultant obstetrician and gynaecologist Professor Dr Nazimah Idris joins us to offer us some insight into developments in technology that allow us to detect early the state of the child we are carrying, and whether such knowledge will be beneficial to both the mother and child.

WORD **LIM TECK CHOON**
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TO KNOW OR NOT TO KNOW?

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First, let's address the burning questions many parents must have: should they even know the condition of the in the mother's womb?

Professor Dr Nazimah Idris says that the ultimate decision naturally lies with the parents. Some parents want to know, and hence they wish for all the possible tests to be done. Others don't, and that's perfectly fine. "It's entirely up to the parents," she says.

To help parents make their decision, she shares the kind of abnormalities that screening can uncover.

Structural abnormalities are physical abnormalities such as improperly developed limbs, heart abnormalities, and such.

Chromosomal and genetic abnormalities include thalassemia and Down syndrome. These are abnormalities involving the genetic materials of the baby. Such genetic abnormalities may be passed on from parent to baby, or they may occur as a result of improper cell division during the development of the foetus.

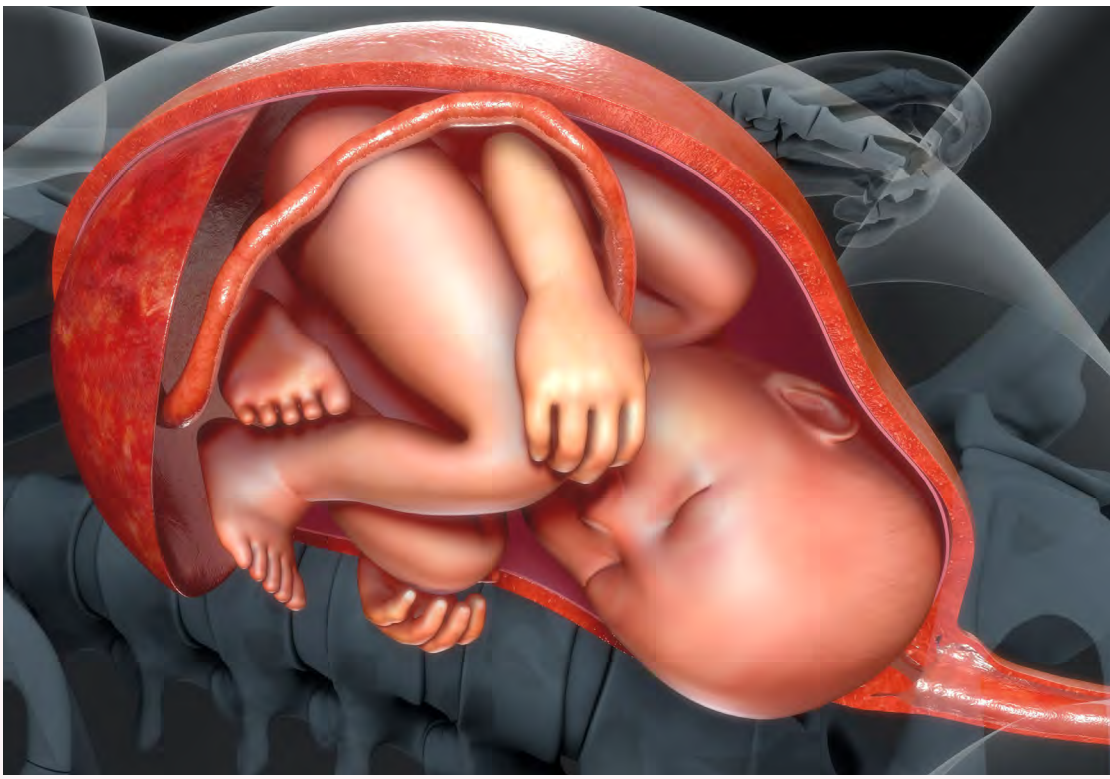
These abnormalities fall under different types and severity. Prof Dr Nazimah shares that some abnormalities may be lethal, such as those involving improperly-developed vital organs such as kidneys. "A lethal anomaly is when you know that the foetus is not compatible with life—the baby is not going to make it after birth," she explains.

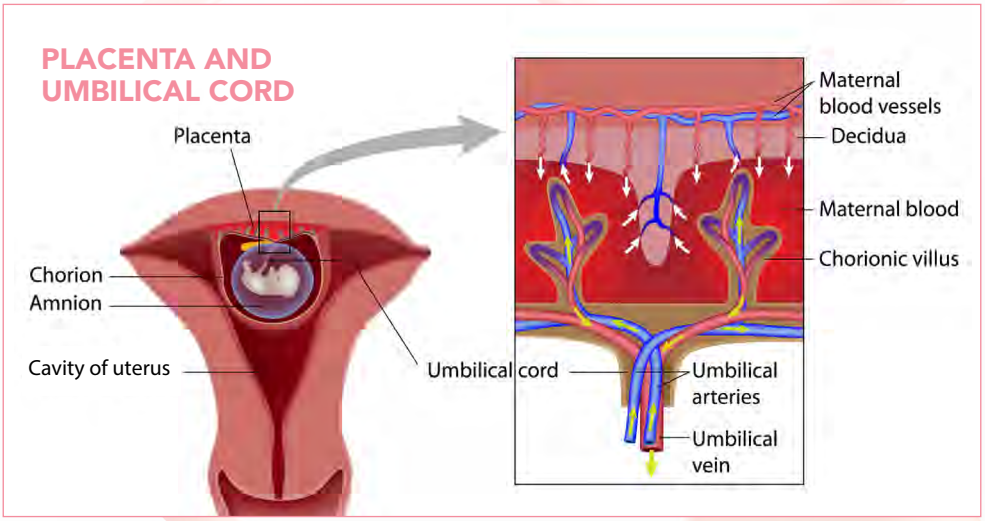
On the other hand, some abnormalities are non-lethal, such as a range of structural abnormalities (limb disabilities, clubfoot and cleft palates, etc) and even more complex conditions such as cardiac defects. Prof Dr Nazimah explains that some of these abnormalities can be addressed and even fixed—some easier than others, naturally—by specialised surgeries and procedures after the baby is born. Genetic or chromosomal anomalies that cause conditions such as Down syndrome are also considered non-lethal.

HOWEVER, THESE SCREENINGS ARE NOT THE BE-ALL AND END-ALL

Prof Dr Nazimah shares that it is possible for a foetus detected with a high risk of abnormality to be born perfectly normal.

Hence, test results can be used as part of the parents' decision-making process, but parents should not jump to quick conclusions based on the result. A better route would be to involve the healthcare team in the decision-making process, so that the parents can evaluate insight and opinion from qualified medical professionals before the making the best educated decision for themselves and the child they are expecting.





THE SCREENING PROCESS

The process of scanning and detecting possible abnormalities in the foetus typically begins with the screening of an expectant mother. "This is usually in the form of a nuchal translucency scan or non-invasive prenatal testing," says Prof Dr Nazimah.

The nuchal translucency scan

The **NT scan**, as the **nuchal translucency scan** is also known as, is an ultrasound commonly offered to mothers during the 1st trimester of their pregnancy.

WHAT IT IS. The NT scan measures the amount of fluid present under the skin at the back of the baby's neck. The size of the area containing the fluid is called the nuchal translucency.

WHAT IT DETECTS. The scan assess the baby's risk of having Down syndrome and other chromosomal disorders. High nuchal translucency could be an indication of the presence of these chromosomal disorders.

However, the NT scan is not a diagnostic tool. It is only a screening procedure. There is a small chance that it can give a false positive result, which is to say, it may suggest that something is amiss when the baby is actually perfectly fine. Hence, the doctor will conduct follow-up tests to confirm an abnormal NT scan result.

Non-invasive prenatal testing

This blood test, often abbreviated as **NIPT**, is typically offered to mothers during the 10th week of pregnancy or later.

WHAT IT IS. Blood obtained from the mother is screened for fragments of DNA from the placenta. The DNA in placental cells is usually identical to the DNA of the foetus. NIPT is therefore a way to screen the baby without bothering the baby in the mother's womb.

WHAT IT DETECTS. These fragments of DNA may reveal the presence of chromosomal disorders such as Down syndrome in the baby.

However, the NIPT is not a diagnostic tool. Just like the NT scan, this is a screening procedure. The doctor will conduct follow-up tests to confirm an abnormal NIPT result.

WHAT'S NEXT AFTER SCREENING?

The doctor will use the results of the screening, along with the couple's family history and background, to determine whether the follow-up diagnostic tests would be beneficial to the mother and her baby.

NEXT STEP: DIAGNOSIS

Once it is deemed that diagnostic tests would benefit the expecting parents, the mother can be considered for screening tests such as chorionic villus sampling or amniocentesis.

"These tests are not without risk though. There is a 1% risk of the test leading to a miscarriage," Prof Dr Nazimah shares.

Chiorionic villus sampling

The chorionic villus is a finger-like protrusion found in the placenta. There are many chorionic villi present to allow oxygen and nutrients to pass from the mother to the foetus. Carbon dioxide and other waste materials will also pass from the foetus to the mother through these structures.

WHAT IT IS. In chorionic villus sampling, a sample of the chronic villi is obtained for further testing. This is typically done between the 10th and 12th week of pregnancy.

The doctor can either insert a catheter through the cervix, or insert a needle through the abdomen and uterus to reach the placenta and obtain the sample.

WHAT IT DETECTS. The DNA found in the sample is the same as that in the foetus. Hence, this test can be used to look out for chromosomal abnormalities (Down syndrome, etc) and a few other genetic problems.

Amniocentesis

During pregnancy, a baby is surrounded by protective fluid called amniotic fluid. Present in this fluid are foetal cells and various proteins—substances that can tell us much about the state of the baby.

WHAT IT IS. With the guidance of an ultrasound, the doctor will locate a safe spot to extract amniotic fluid. A needle is used to extract a sample of amniotic fluid. The entire procedure shouldn't last more than 10 to 15 minutes.

This test is usually offered from the 15th week of pregnancy, and has a high degree of accuracy.

WHAT IT DETECTS. This test detects chromosome abnormalities (Down syndrome, trisomy 21, etc) and genetic disorders (cystic fibrosis, etc).

Ultrasound

The ultrasound scan is is used to diagnose structural abnormalities such as heart abnormalities and brain abnormalities, as well as to screen for foetal anaemia in Rh-incompatible pregnancies.

MY BABY HAS AN ABNORMALITY —WHAT CAN I DO NEXT?

If abnormalities are detected, the expecting mother's healthcare team will present treatment options for consideration and discussion. With the advance of medical treatments, it is possible that sometimes what seems like a bleak situation may not appear to be as hopeless as it seems after a thorough discussion with the healthcare team.

However, there may be instances when termination of the pregnancy may have to be considered, such as when the foetus has a lethal abnormality or when the continuation of pregnancy endangers the mother's life.

Dr Nazimah explains that termination in Malaysia is highly regulated and governed by laws. It is only considered when the continuation of the pregnancy is deemed detrimental to the mother, either physically or mentally. "In times like this, we consult a psychiatrist to assess whether the mother may be at risk of severe depression or anxiety after the pregnancy," she adds.

However, she says that some couples also decide to continue the pregnancy, even when the baby is not expected to survive once they are born. "It is an emotional issue. Some couples want to be with their baby for a bit longer, so they carry the pregnancy to term. During this time, they prepare themselves emotionally for what it is to come, and find closure after the delivery."

CAN WE ENSURE THAT THE CHILD WE ARE CARRYING IS FREE FROM ABNORMALITIES?
Get the answers from Prof Dr Nazimah in an exclusive article on the HealthToday website.