DOES A LOW RISK IONA® TEST RESULT MEAN MY BABY IS COMPLETELY HEALTHY?

Low risk results do not eliminate the possibility that your pregnancy may have other chromosomal abnormalities, birth defects, or other conditions, such as neural tube defects. In addition, a low risk result does not guarantee a healthy pregnancy or baby.

WHO CAN I SPEAK TO FOR MORE INFORMATION ON THE IONA® NIPT?

If you are considering NIPT, it is important to consult your managing doctor. Additionally, you can seek genetic counselling from a registered Genetic Counsellor to discuss the merits and pitfalls of all types of antenatal testing options.

For genetic counselling, contact Sarah Walters at 012 678 1350/62 or walterss@ampath.co.za

For any other queries, please email: nipt@ampath.co.za
WHAT IS NON-INVASIVE PRENATAL TESTING (NIPT)?
During your pregnancy, the health of your baby is typically monitored by your doctor, using a variety of screening tests. These can include ultrasound scans and maternal serum tests. These screening tests assist your doctor in determining whether your baby is at risk of health conditions, including genetic conditions, such as Down Syndrome.

If your pregnancy is identified to be at risk of being affected with a genetic condition, your doctor might refer you for an invasive diagnostic procedure such as an amniocentesis. An amniocentesis obtains cells from your baby’s amniotic fluid in order to perform a genetic test. While invasive testing is regarded as the gold-standard for prenatal genetic testing, it is associated with a small risk of miscarriage. Non-invasive prenatal testing (NIPT) makes use of a blood sample obtained from a pregnant woman, to test her baby for certain genetic conditions. Being non-invasive, NIPT avoids the risks associated with invasive procedures.

HOW DOES NIPT WORK?
During a pregnancy, DNA from the placenta of the developing baby is released into the maternal bloodstream. Using a maternal blood sample, it is possible to measure the amount of DNA that is present. By calculating the ratio of DNA from the placenta, the risk of a baby being affected with a chromosomal condition (such as Down Syndrome) is calculated.

WHAT DOES NIPT TEST FOR?
A trisomy occurs, when a baby inherits three copies of a chromosome, instead of the usual two copies. Similarly, a monosomy occurs when one copy of a chromosome is inherited. Both trisomies and monosomies are associated with adverse pregnancy outcomes – either miscarriage or a genetic condition.

THE IONA® NIPT
The IONA® test is a non-invasive prenatal test (NIPT), which is able to determine the risk of a baby being affected with a chromosomal condition, but does not pose any risk to the pregnancy.

Using a maternal blood sample from 10 weeks’ gestational age, the IONA® test is able to determine the risk of a pregnancy being affected with:
- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards Syndrome)
- Trisomy 13 (Patau Syndrome)

Additionally, sex chromosome analysis can be performed on singleton pregnancies, for sex chromosome conditions, including Turner Syndrome (XO) and Klinefelter Syndrome (XXY).

Foetal sex determination is optional for singleton pregnancies only.

IS THE IONA® TEST AN OPTION FOR ME?
The IONA® test is available for:
- Singleton pregnancies
- Twin pregnancies
- Surrogate or donor pregnancies
- IVF pregnancies
- Women who are at least 10 weeks pregnant

HOW ACCURATE IS THE IONA® TEST?
The IONA® NIPT has an overall accuracy of >99% for trisomies 21, 18 and 13. Although highly accurate, the IONA® test (as with all NIPT tests) is a screening tool, and not diagnostic.

Due to this, when a positive NIPT result is obtained (indicating high risk for a chromosomal condition), it is recommended that confirmatory follow-up testing is performed via invasive testing, such as amniocentesis. If the result of the IONA® test is that the pregnancy is low risk for the chromosome conditions screened, then routine antenatal care is appropriate and an invasive procedure can be avoided.