- 额外染色体的存在可以导致严重的先天性身心残缺 和发育问题。
- 检测染色体三体综合症之诊断检验如羊膜穿刺术和 绒毛取样术都是侵入性和有可能导致流产的风险。
- 您现在可在怀孕10个星期或以上时选择非侵入性的 NICC®检验以避免流产的风险。

### NICC®是如何检测染色体异常?

- 在怀孕期间,游离胎儿的DNA会通过胎盘里的血管 进入母亲的血液(请参考图一)。
- NICC®檢验只须要10m1母亲的血液样本。从这血液 样本, 胎儿的基因将会被提取然后引用新一代测序 方式(NGS)来加以分析。

### NICC®能筛查些什么?

	100711 - 11		
染色体三体综合症:	唐氏综合症 Trisomy 21 爱德华综合症 Trisomy 18 巴陶氏综合症 Trisomy 13 Trisomy 9 Trisomy 16 Trisomy 22	染色体微缺失症:	1p36 2q33.1 16p12.2 第二型狄喬治综合症 雅各布森综合症 猫叫综合症 (5p) 遺傳性唇顎裂综合症 普瑞德威利综合症/安
性别染色体异常:	特纳氏综合症 (XO) 克氏综合症 (XXY) X三体综合症 (XXX) 超Y综合症 (XYY)	性别:	格曼综合症 男(XY) 女(XX)

#### 注意:

- 双胞胎怀孕结果只限于唐氏综合症:爱德华综合症 与巴陶氏综合症。
- 只要捐赠者/收卵者没有任何已知的染色体异常的 条件下, NICC®检验也可以为辅助怀孕的妇女进行 检验。

#### 谁应该考虑NICC®?

- 希望提前了解胎儿健康状况的孕妇。
- 年龄偏高的妇女(35岁或以上)。
- 个人或家庭有生理缺陷胎儿的家史。
- 曾怀有三体综合症孩子的孕妇。
- 母体血清筛检出现阳性结果。

## NICC®之优势



无风险





简单

# 如何解读NICC®检验结果?

风险	说明
低	<ul><li>胎儿拥有所测试之染色体异常的可能性偏低</li><li>建议为胎儿进行常规检查以监测胎儿之发育</li></ul>
髙	<ul><li>胎儿受影响的风险增加</li><li>进一步的诊断测试可确认高风险结果</li></ul>

- 作为一个筛查性检验, NICC®测试会预测出您的 宝宝是否有染色体异常的风险。
- 您的医生会根据结果向您推荐下一步行动。
- 此检验之结果并不能排除所检测的染色体拥有:
  - 其它异常的可能性
  - 其它遗传性疾病或并发症
  - 胎儿或怀孕期显现任何其他症状
- 胎儿性别预测应通过诊断性检验来断定。

#### 请咨询您的医生以获取更多资料。

1. Gregg et al. (2016). Noninvasive prenatal screening for fetal aneuploidy, 2016 update: American College of Medical Genetics and Genomics. Genet Med., 18(10), 1056-65







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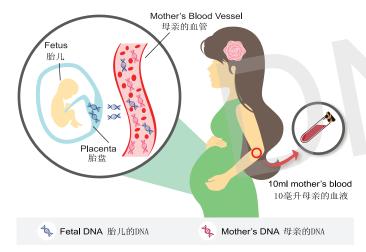
Non-Invasive ChromosomesCheck

Simple, safe and highly accurate Non-Invasive Prenatal Test (NIPT)

#### **Chromosomal Abnormalities: Trisomy**

- A normal cell contains 46 chromosomes arranged in 23 pairs.
- A trisomy is a disorder characterised by having additional chromosome. Having an extra copy of the paired chromosomes will cause abnormal developments such as Trisomy 21 or also known as Down Syndrome.
- Trisomies can cause severe congenital physical disability and developmental problems.
- The risk of having a child with chromosomal abnormalities such as Down Syndrome, Edwards Syndrome and Patau Syndrome increases with mother's age.
- Diagnostic tests for trisomies detection such as amniocentesis and chorionic villus sampling (CVS) are invasive and impose a risk of miscarriage to the pregnancy.
- You can now avoid this risk with NICC® test that can be done any time after 10 weeks of pregnancy.

#### How does NICC® work?



Picture 1: Cell free fetal DNA in mother's blood. 图1: 母亲血液中的游离胎儿DNA。

- During pregnancy, cell free fetal DNA released from placenta into the mother's blood vessel (Picture 1).
- Using only mother's blood, NICC® detects the baby's DNA and measures the risk of chromosomal abnormalities using Next-Generation Sequencing (NGS) method.

#### What does NICC® screen for?

TRISOMIES	Down Syndrome (Trisomy 21) Edwards Syndrome (Trisomy 18) Patau Syndrome (Trisomy 13) Trisomy 9	MICRODELETION SYNDROMES	1p36 2q33.1 16p12.2 DiGeorge Syndrome 2 Jacobsen Syndrome Cri-du-chat Syndrome (5p)
OMES	Trisomy 16 Trisomy 22  Turner Syndrome (XO)		Van der Woude Syndrome Prader-Willi/Angelman Syndrome
SEX CHROMOSOME ANEUPLOIDY	Klinefelter Syndrome (XXY) Triple-X Syndrome (XXX) Jacob's Syndrome (XYY)	GENDER	Male (XY) Female (XX)

#### Notes:

- For Twin Pregnancy, NICC® can only screen for Trisomies 21, 18, and 13.
- NICC® is also suitable for Assisted Pregnancy such as IVF, surrogate and donor egg pregnancies; provided that the surrogate/donor does not have any known chromosomal abnormalities as per the knowledge of the clinician.

# American College of Medical Genetics and Genomics (ACMG), recommends:

Informing all pregnant women that NIPT/NIPS (Non-invasive prenatal screening) is the most sensitive and reliable option for traditionally screened aneuploidies involving chromosome 13. 18 and 21<sup>1</sup>.

#### Who should consider NICC®?

- Any pregnant women who is anxious to have early information about her baby's health.
- Women with advanced maternal age (35 years and above).
- · Personal or family history of birth defect.
- Previous birth of a child with a birth defect.
- · Positive serum screening test.

#### Advantages of NICC®





SIMPLE

#### What does NICC® tell you?

Risks	Indications			
Low	<ul> <li>Likelihood of the baby being affected with the tested condition is low</li> <li>Routine follow-ups should be performed to monitor the baby's growth</li> </ul>			
High	<ul> <li>Baby has an increased risk of being affected</li> <li>Further diagnostic test is recommended to confirm High Risk results</li> </ul>			

- Your doctor would recommend to you the next course of action depending on the NICC® results.
- The result of this test also does not eliminate the possibility of:
  - Other abnormalities of the tested chromosomes.
  - Other genetic disorders,
  - Other complications in the fetus or pregnancy.
- Gender prediction should be confirmed with diagnostic testing.

#### Please consult your doctor for more information

### 染色体异常:染色体三体综合症

- 一般人体的细胞通常拥有46个或23对染色体。
- 染色体三体综合症(例如唐氏综合症)是以额外的染色体为特征的染色体病症。
- 随着孕妇的年龄增长,孩童患有染色体异常(例如: 唐氏综合症,爱德华综合症和巴陶氏综合症)的 风险也会增高。