Is my baby normal?

We all expect our babies to be healthy and normal, but in reality congenital malformations occur in 2-3% of all pregnancies. Some anomalies can be prevented while others, unfortunately, cannot.

Preventing abnormalities

Intake of folic acid from conception can reduce the incidence of neural tube defects such as spina bifida. Diabetics with good control of blood glucose (to be started *before* conception) can also prevent the development of foetal malformation found in babies of diabetic mums. Exposure to harm (irradiation and chemicals/drugs) must be avoided. Stopping smoking and excessive alcohol consumption are important. Patients with existing disease(s) should consult the clinic before getting pregnant regarding their medical treatment- the dosage may be reduced or the medicine changed to a safer one.

Congenital anomalies

Abnormalities that are found in newborns are known as congenital abnormalities. These anomalies can be a single structural abnormality (such as a heart defect) or a collection of several abnormalities forming a syndrome (such as Down's syndrome).

A syndrome is usually a more serious disorder, and may be related to chromosomal abnormailities. The most well known and most common chromosomal abnormality is Down's syndrome (Trisomy 21). Generally, it occurs in 1 in 700 pregnancies. The risk varies with maternal age- the older you are the higher the risk becomes. At 30 years old, the risk is 1:900; at 36 years old- 1:275; at 40 years old- 1:100.

Other chromosomal abnormalities include lesser known but equally serious syndromes are: Edwards' syndrome/trisomy 18 (1 in 3000), Patau's syndrome /trisomy 13 (1 in 5000), Turner syndrome/monosomy X (1 in 2500), Klinefelter syndrome/XXY (1:1000), triple X syndrome/XXX (1 in 800) and Jacob's syndrome/XYY (1 in 650).

Majority of congenital anomalies are, however, non-chromosomal. The largest risk being congenital heart defects (6 in 1000), followed by limb defects (4 in 1000), anomalies of urinary system (3 in 1000) and nervous system defects (2 in 1000).

Abnormalities caused by genetic disorders are less common. New technologies have allowed the detection of *some* of these abnormalities, including microdeletion syndromes (1 in 1000). More well known genetic disorders like cystic fibrosis, Duchenne muscular dystrophy, androgen insensitivity syndrome (male infant appearing as a female), congenital adrenal hyperplasia (uncertainty with the gender of the baby at birth- female infant looking like a male or vice versa) and haemophilia cannot be screened for.

There are also other abnormalities with multifactorial or unknown causes. These include autism, cleft palate/hare-lip and psychiatric disorders.

Screening for abnormalities

Various methods have been developed to detect or screen for congenital anomalies. Despite best attempts, babies are still born with malformations. The reasons are many- the abnormalities being too small to be detected, the test being not good enough (not sensitive), pregnant mum declined being screened, access to the screening test not available, absence of in-utero treatment and termination of pregnancy not an option (unwilling to do so or too late in pregnancy to do so).

Proper counselling with regards to the screening tests must be done with sensitivity. The ultimate choice (to screen or not to screen) lies with the parents, as some of the abnormalities may need the consideration of terminating the pregnancy. Equally important is the location of delivery should the baby requires immediate treatment for any detected anomalies.

Screening for chromosomal abnormalities (aneuploidies)- ***the following tests are available in the clinic***:

- 9 weeks onwards- **non invasive prenatal test (NIPT)** looks at placental (which is similar to foetal) chromosomes found within maternal blood. It screens for chromosomal abnormalities resulting in the following syndromes- Down's, Edwards', Patau's, Turner's, Jacob's, Klinefelter, triple X. Additionally, with the same test, some microdeletion syndromes (Di George, Prader-Willi, Angelman, Cri-du-chat and 1p36 deletion) can be screened for. It will also inform you the gender of your baby. The clinic sends the maternal blood to **Panorama, USA** for NIPT.

- 11-13 weeks GA- **combined first trimester screening test**. In this test, ultrasound measurement of the nuchal translucency (NT) together with maternal biochemistry (PAPP-A and hCG) are used to derive a *risk ratio* of having a baby with Down's syndrome (as well as Patau's and Edwards' syndromes). The clinic sends the maternal blood to **Pathlab, Singapore** for this test.

- 15-20 weeks GA- **triple test** makes use of maternal biochemistry levels (AFP, hCG and E3) to assess the risk of having a Down's syndrome (as well as Edwards' and Patau's syndromes) baby. It can also assess the risk of neural tube defects. The clinic sends the maternal blood to **Pathlab, Singapore** for this test.

Amniocentesis is diagnostic but it is associated with 1% miscarriage risk, even if the baby is found to be normal. It is best done at 16 weeks GA and above.

Screening for structural abnormalities- the foetal anomaly (or morphology) scan
This is a very detailed ultrasound scan looking at individual organs of the baby. Among the important organs being examined are the brain (hydrocephaly or microcephaly), the heart (heart valve defects) and backbone (spina bifida). Functional abnormalities (blindness, deafness, autism, hyperactivity) cannot be examined. Finger/toe counting may or may not be performed because it is not considered as a serious/lethal anomaly. The anomaly scan is best done at 20-22 weeks GA.

Abnormalities	Tests available in the clinic
Down's syndrome, Edwards' syndrome, Patau's syndrome, Turner's syndrome, Klinefelter syndrome, Jacob's syndrome, triple X syndrome	NIPT (Panorama, USA) at 9 wk onwards or Combined first trimester test (Pathlab, Singapore) at 12 wk or Amniocentesis at 16 wk onwards
Microdeletion syndromes (Di George, Prader-Willi, Angelman, Cri-du-chat and 1p36 deletion)	NIPT (Panorama, USA) at 9 weeks onwards
Structural abnormalities	Foetal anomaly scan at 20 wk
Functional abnormalities	No test currently available (during pregnancy)

Abnormalities	Incidence		Abnormalities	Incidence
Congenital heart defects	6 in 1000		Di George syndrome	1 in 2000
Limb defects	4 in 1000		Turner's syndrome	1 in 2500 girls
Anomalies of urinary system	3 in 1000		Edwards' syndrome	1 in 3000
Nervous system defects	2 in 1000		1p36 syndrome	1 in 5000
Jacob's syndrome	1 in 650 boys		Patau's syndrome	1 in 5000
Down's syndrome	1 in 700		Prader-Willi syndrome	1 in 10,000
Triple X syndrome	1 in 800 girls		Angelman syndrome	1 in 12,000
Klinefelter syndrome	1 in 1000		Cri-du-chat syndrome	1 in 20,000

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Dr. Gozali has worked as an obstetrician and gynaecologist in the UK for 20 years. During that time he has been at various teaching hospitals including those of University of London and University of Oxford. He has also worked as clinical lecturer at the University of Oxford.