Dr. Go Clinic for Women Jl Kebon Kacang 30, No 12, Tanah Abang, Jakarta. 0213143360 08118855872

Tests for Down's syndrome

(*and* Edwards' syndrome, Patau's syndrome, Turner's syndrome, triploidy, DiGeorge syndrome, 1p36 deletion syndrome, Angelman syndrome, Cri-du-chat syndrome, Prader-Willi syndrome)

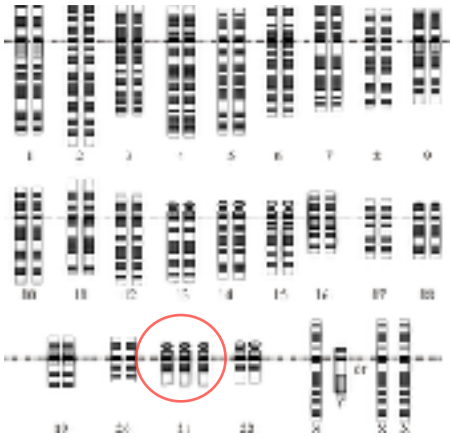
As the public is increasingly aware of Down's syndrome and the availability of tests to screen for Down's syndrome, the clinic offers the latest and most advance technology to perform such a screening test.

The test available can, additionally, screen for other chromosomal abnormalities and microdeletion syndromes.

What is Down's syndrome?

Down's syndrome is also known as trisomy 21. This is a condition whereby the cells in the body contain 47 chromosomes, instead of the normal 46 chromosomes. To be exact, in Down's syndrome there are three sets of chromosome number 21. (see picture to the right)
If the extra set of chromosome occurs in chromosome number 18, the condition is known as Edwards' syndrome (trisomy 18). Similarly, Patau's syndrome is trisomy 13, Klinefelter syndrome is XXY (an extra X chromosome). Turner's syndrome occurs when one set of X chromosome missing (XO, monosomy) in a female.

Down's syndrome (trisomy 21)



The condition whereby the number of chromosomes is not normal (extra or deleted set of chromosomes) is known as aneuploidy. It occurs as a result of an error at the time of cell division.

What causes Down's syndrome?

Down's syndrome occurs sporadically (by chance), *in most cases*. Increasing maternal age increases the risk of Down's syndrome- the older you are the higher the chance of having a Down's syndrome baby.

Maternal age	Incidence of Down's syndrome	Maternal age	Incidence of Down's syndrome	Maternal age	Incidence of Down's syndrome	Maternal age	Incidence of Down's syndrome
15 - 19	1 in 1250	33	1 in 625	38	1 in 175	43	1 in 50
20 - 24	1 in 1400	34	1 in 500	39	1 in 140	44	1 in 40
25 - 29	1 in 1100	35	1 in 350	40	1 in 100	45 and older	1 in 25
30 - 31	1 in 900	36	1 in 275	41	1 in 85		
32	1 in 750	37	1 in 225	42	1 in 65		

It should be noted however that *the incidence of livebirths with Down's syndrome babies* ***is higher among the younger mothers***. The reason being the fact that there are more pregnancies among younger women, and that older women are aware of the increased risk of Down's syndrome- hence taking steps to screen and terminate the affected pregnancy. **Screening for Down's syndrome is equally important for younger mothers.**

Revised December 2018 Written by Dr. Gozali MBBS (London) DFFP (UK) MRCOG (UK)

Hereditary cause of Down's syndrome is also known but contribute to a small percentage (about 4%) of all cases. This is due to *translocation disorder* whereby an affected chromosome, carrying extra genetic materials from chromosome 21, is being inherited from a parent (either father or mother) to the baby. The risk of recurrence (the subsequent baby with a Down's syndrome) is less than 15% (3 % if the father is a carrier and 10-15% if the mother is a carrier).

External factors such as maternal intake of certain food (diet), medicine/drugs, smoking, alcohol, chemicals (monosodium glutamate- MSG, flavourings, colouring, food additives, pesticides), exposure to irradiation (X rays) and travelling (frequent or occasional) are not known to be the causes of Down's syndrome.

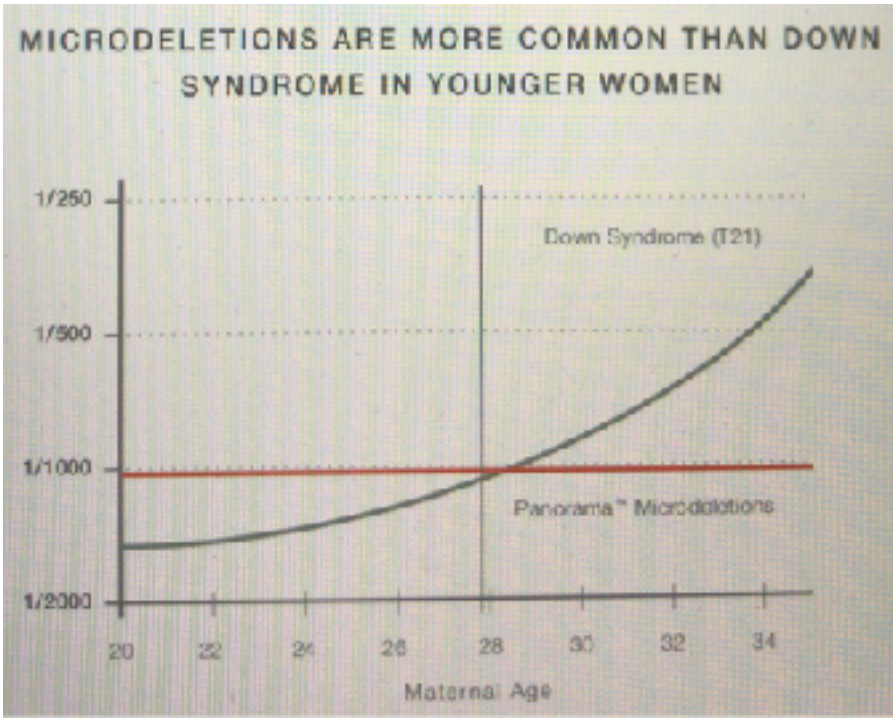
What test(s) are available *in the clinic*?

Various tests have been developed to screen for and also to diagnose Down's syndrome.

The *diagnostic* test is amniocentesis, a procedure whereby a needle is inserted into the uterine cavity to obtain amniotic fluid. The small number of foetal cells in the amniotic fluid are cultured and chromosomes counted in due course. However, the procedure is associated with a miscarriage risk (1% even if the baby is normal). Of note, having performed the amniocentesis, results may not be available because of failure of the foetal cells to grow in the lab. **Amniocentesis is available in the clinic**.

Because of the associated risk of miscarriage with amniocentesis, the clinic recommends alternative test, namely **NIPT** (non-invasive prenatal test). This test involves only taking ***blood*** from pregnant women. The technology will search out the minute amount of foetal sample (cell free DNA) among the "sea" of maternal blood. In depth analysis of the foetal sample will then produce various results among them being Down's syndrome risk.

At the time of print, the clinic offers **Panorama (USA)** to perform NIPT to patients. Among the many advantages, this particular test can be offered to patients as early as **9 weeks** pregnant (with no upper limit). It focuses on the "common" and "relevant" aneuploidies such as trisomies 21,18,13, monosomy (XO) and triploidy.



Additionally, Panorama also offers tests for microdeletion syndromes- DiGeorge, Prader-Willi, Angelman, Cri-du-chat and 1p36 deletion. Together these syndromes occur in around 1 in 1000 pregnancies (see graph on right), regardless of maternal age (cf Down's risk is 1 in 900 pregnancies, and the risk of which increases with maternal age). In fact,

microdeletion syndromes occur more often than Down's syndrome in younger women. Hence the importance of requesting screening tests for microdeletion syndrome for all age group including/especially the younger mothers. The relevance (of requesting microdeletion syndromes) was being able to offer some treatment to the affected babies (example low blood calcium in DiGeorge syndrome).

Because of the (high) cost of NIPT, some patients may opt for other tests. By simply performing ultrasound scan at 12 weeks pregnancy, features of Down's syndrome (nuchal translucency and nasal bone) can be characterised. To improve on the accuracy of the screening test, additional blood test may be requested- combined first trimester screening test (done at 12-14 weeks). If the "combined" test is missed, "triple test" can be requested at 16 weeks pregnancy. If no screening test was done at all, the 20 weeks anomaly scan can also be done to screen for Down's syndrome.

Microdeletion syndromes

Syndrome	Incidence	Lifespan	Mental effects	Heart defects	Other features
DiGeorge	1 in 2,000	Reduced	Mild to moderate intellectual disorder and schizophrenia	Yes	Palate and feeding issues, immune problems, low calcium, seizures
1p36 deletion	1 in 5,000	Infancy to adult	Severe intellectual disorder and behavioural problems	Yes	Limited/no language, hearing loss, abnormal ears, seizures
Prader-Willi	1 in 10,000	Reduced	Mild to moderate intellectual disorder and behavioural problems	No	Hypotonia in babies, insatiable appetite
Angelman	1 in 12,000	Normal	Severe intellectual disorder	No	"Happy" affect, ataxia, microcephaly, no speech, seizures
Cri-du-chat	1 in 20,000	Infancy to adult	Moderate to severe intellectual disorder and behavioural problems	No	Cat-like cry, growth problems, wide set eyes

What to do with a Down's syndrome baby?

Screening test with positive results should be confirmed by a diagnostic test (amniocentesis or chorionic villous sampling). Down's syndrome individuals have a range of abnormalities from mild to severe; some requiring multiple corrective surgeries. The affected IQ is also varied.

Amniocentesis is available at the clinic, and is done when the pregnancy is 16 weeks onwards.

Knowing the diagnosis and therefore preparing for all eventualities is all important for the care of the affected baby. Centres with specialised facilities especially paediatric heart surgery may be needed. There are support groups that are very useful in helping to manage the child.

With increasing advancement in medical technologies and social acceptance of Down's syndrome, termination of pregnancy is being chosen less often.

Dr. Gozali obtained his basic medical degree (Bachelor of Medicine, Bachelor of Surgery) from University College, London (University of London) and post graduate qualification from the Royal College of Obstetricians and Gynaecologists (UK). Other qualifications include a diploma from the Faculty of Family Planning (UK). He is fully registered with the General Medical Council (UK).

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.