

DOWN SYNDROME SCREENING

What is Down Syndrome ?

Down syndrome is caused by a baby having an extra chromosome 21 in each cell (figure 1). This condition is characterised by distinctive facial features and varying degree of mental retardation. Other structural defects are common such as congenital heart disease and gastrointestinal defects. Some of them may perform simple work but long term care is usually required. The chance of having baby with Down syndrome increase with increased maternal age (figure 2).

Figure 1 : Chromosome for Down syndrome

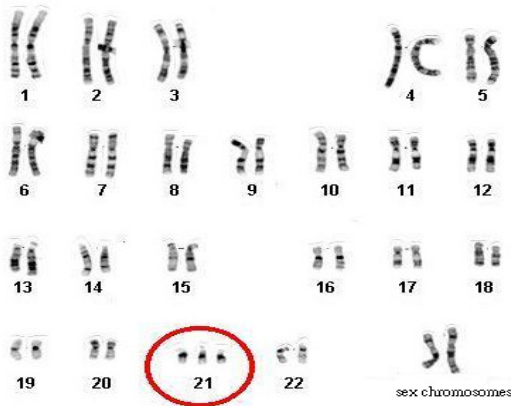


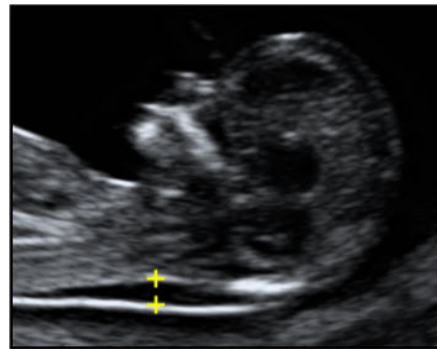
Figure 2 :The chance of baby with Down syndrome

Maternal age	Risk of Down syndrome
20	1 / 1527
25	1 / 1352
30	1 / 895
35	1 / 356
40	1 / 97

1. 1st Trimester Down Syndrome Screening

Assessment the risk of Down syndrome by a combination of maternal age, fetal Nuchal translucency thickness (figure 3) and maternal serum free β -human chorionic gonadotropin and pregnancy-associated plasma protein-A (PAPP-A) at 11-14 weeks of gestation. It can detect 90% of Down syndrome.

Figure 3 : Fetal Nuchal translucency thickness



2. 2nd Trimester Down Syndrome Screening

This screening is performed at about 16-20 weeks of gestation. It measures the hormonal levels of alpha fetal protein (AFP) and human chorionic gonadotrophin (hCG) from the pregnant mother. In combining with maternal age, it recalculates the risk of a woman carrying a Down syndrome baby. The detection rate for Down syndrome is 70%.

3. Chorionic Villus Sampling or Amniocentesis

Fetal tissue including chorionic villus or amniotic fluid are taken out for culture to determine whether the fetus is Down syndrome. These procedures carry a risk of miscarriage of 1%.

Remark: 1st or 2nd trimester for Down syndrome screening only calculate the risk of Down syndrome. It is not a diagnostic procedure.

An increased risk does not mean a chromosome abnormality has been diagnosed in the fetus. It only indicates that further tests need to be considered. The options for further testing may include an chorionic villus sampling or amniocentesis for fetal chromosome analysis. These tests will diagnosed Down syndrome .

A low risk result indicates that the risk that the fetus has Down syndrome is not high. It does not completely eliminate the possibility that the fetus may have Down syndrome or other chromosome abnormalities.