

Down Syndrome

A Guide for Pregnant Women



This brochure provides information about Down Syndrome. It is design to respond to questions most frequently asked by pregnant women.



آغا خان یونیورسٹی ہسپتال، کراچی

The Aga Khan University Hospital, Karachi.

P.O. Box 3500, Stadium Road, Karachi-74800, Pakistan,

Website: www.aku.edu

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What is Down Syndrome?

In Down Syndrome, the baby is born with 47 chromosomes instead of 46 in each body cell. This results in physical malformations - small, short flat-bridged nose; wide-set, slanted eyes and some degree of mental retardation. The risk of Down Syndrome is about 1 in 700 pregnancies.

What are Chromosomal Abnormalities?

Chromosomes are like books/information manual for the functioning of the cells in the body. They carry the hereditary material that determines the individual's physical characteristics. In order to function normally, human cell should have 46 chromosomes (23 pairs). Any change in the number or "map" of these chromosomes can result in chromosomal abnormalities.

Who is at Higher Risk?

- Every woman has a small risk of chromosomal abnormalities. The screening for this abnormality can be performed as early as 12 weeks via ultrasound for Nuchal Translucency Measurement.
- The risk increases as the woman grows older(>35 years)

What Tests are done to Assess the Risk?

- Nuchal Translucency Ultrasound measures the fluid behind the neck of the foetus. On the basis of that measurement, risk assessment for Down's syndrome can be done.
- By using Nuchal Translucency Measurement, 80% cases of Down Syndrome can be detected.

- After the scan, the estimated risk of Down Syndrome will be discussed with you. It is up to you and your husband to decide if you would like to have a confirmatory test, like amniocentesis, or chronic villous sampling
- Irrespective of whether you will have an invasive test or not, it is recommended that you have a 20 weeks ultrasound to look for abnormalities and markers for chromosomal abnormalities.

How is this Scan Done?

- This is a trans-abdominal scan. No special preparation like full bladder etc, is required for this ultrasound.

Is There Any Other Test For Screening?

- If you book after 12 weeks, the risk of Down Syndrome can be assessed by a blood test called Triple Test, although it is considered to be less accurate than Nuchal Translucency Measurement.
- The risk assessment can be repeated in around 20 weeks by performing a detailed anomaly ultrasound and by looking for the subtle indications of Down Syndrome. Following this, the risk can be re-assessed.

What Happens if The Ultrasound Shows Increased Risk of Down Syndrome?

- If the scan shows increased risk of Down Syndrome, this risk can be confirmed by an invasive test like Amniocentesis or by Chorion Villus Biopsy.
- Amniocentesis is the test where water around the baby is tested and in the same way if placenta is sent for the test it is called Chorion Villus Biopsy.

- When the diagnosis is confirmed then further options about the pregnancy can be explored and discussed with your doctor.

For further information, please call:

Consulting Clinic 4
Aga Khan University Hospital
Karachi
Phone : 4930051 - Ext : 1036-79

Fetomaternal unit
Aga Khan University Hospital
Karachi
Phone : 4930051 - Ext : 2293

