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# DOWN'S SYNDROME

- Down's Syndrome is the most common single cause of human birth defect.
- Down's syndrome is known to occur in one in every 600 pregnancies around the world
- Half of the fetuses with Down's syndrome will miscarry and only 50 % will result in a live birth.





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Features of Downs Syndrome



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Described by Langdon Down in 1866

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by Jérôme Lejeune

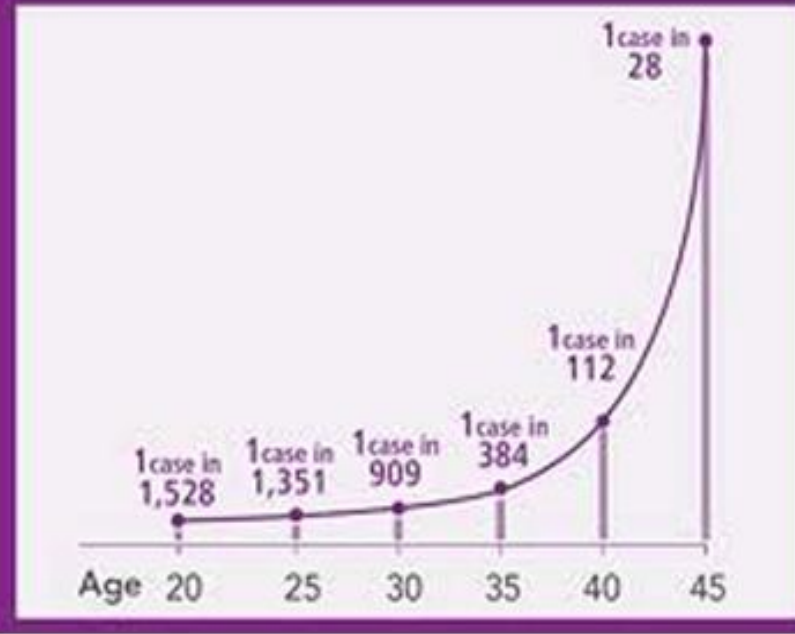
- Cause Discovered by Jerome Lejeune
- Genetic condition in which a person has 47 chromosomes instead of the usual 46
- There is an extra copy of chromosome 21, therefore Trisomy 21



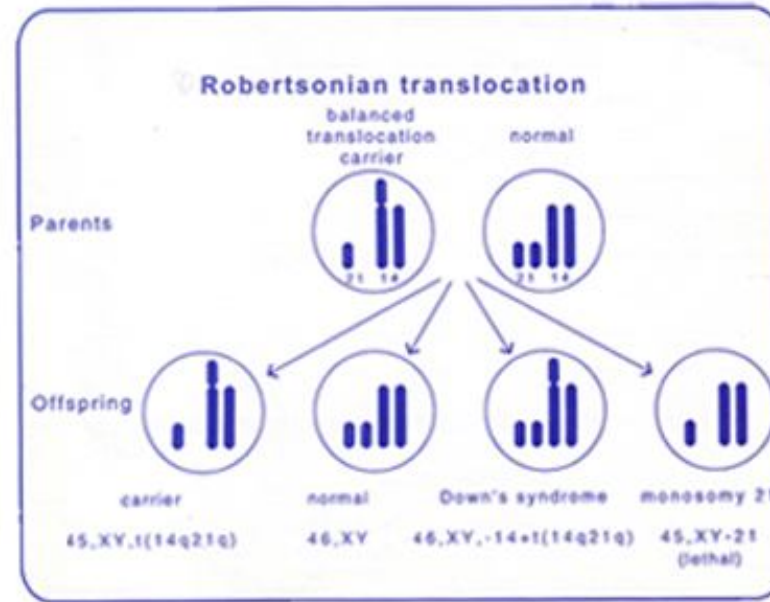
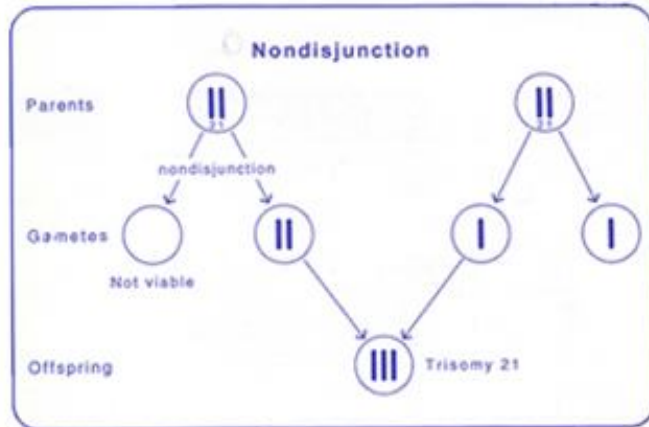


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### PROBABILITY OF GIVING BIRTH TO A BABY WITH TRISOMY 21 BY WOMAN'S AGE



- Incidence of giving birth to a Down Syndrome baby increases with advancing maternal age ( Greater than 35 years )



- Most common cause is incorrect division of the chromosomes in the baby
- Low recurrence risk

- There is a problem in one of the parents genes

(Confirmed by Blood Tests)



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← Flattened Back of Head

← Small Low Set Ears

← Thick Skin Behind the Neck

← Small Chin

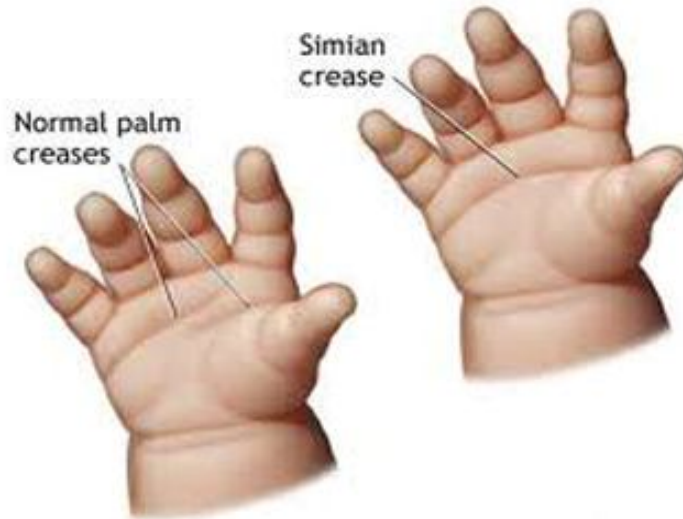




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### Trisomy 21 - Hand Features





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← Wide Gap Between Big toe and  
Second Toe

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Short stature





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1. Congenital Heart Defects : “ hole in the heart” - ventricular septal defect and atrial septal defect
2. Gastro-intestinal blockages
3. Low muscle tone
4. Varying degrees of mental retardation
5. A Blood test in the child can be done to check for the extra chromosome and confirm the diagnosis





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Risk of having another baby with Down's syndrome is 1 % unless there is a problem in the parents chromosome makeup

### High risk women :

1. women with a previous Down syndrome baby
2. Age > than 37 years
3. Parents who are carriers of an abnormal chromosome ( 21 )
4. Family history of Down syndrome babies

All High risk women should be advised to have their baby's chromosomes tested for any abnormalities by Pre-natal Invasive tests



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## Pre - natal Invasive procedures



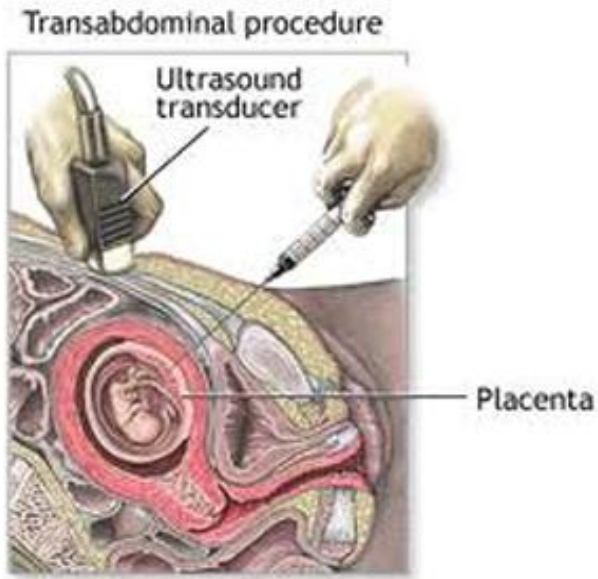
- Done under continuous ultrasound guidance
- Needle is inserted in the mother's womb
- Testing of fetal tissue samples





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## Chorionic Villus Sampling



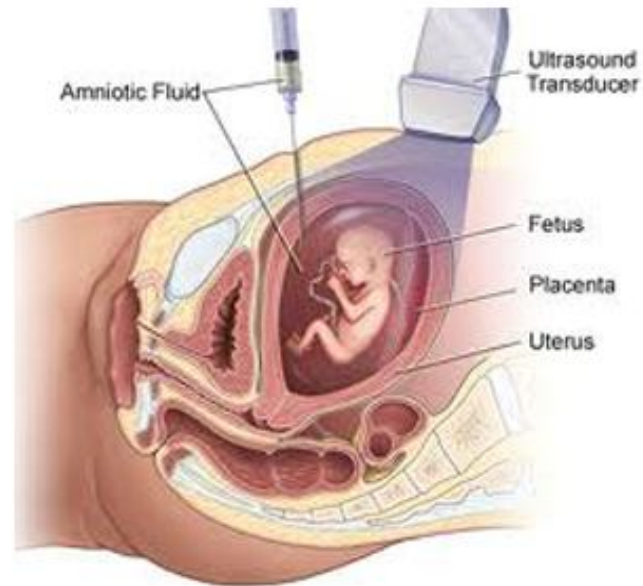
- Done between 11 to 13 weeks of pregnancy
- Removing a piece of placenta ( Afterbirth )





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## Amniocentesis



- Done between 16 to 20 weeks of pregnancy
- Removing the fluid around the baby

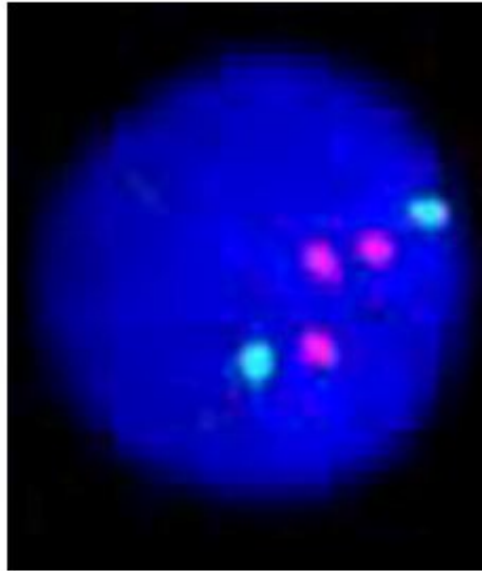






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## Baby's Chromosomes are studied by (1) FISH ( fluorescent in situ hybridization )

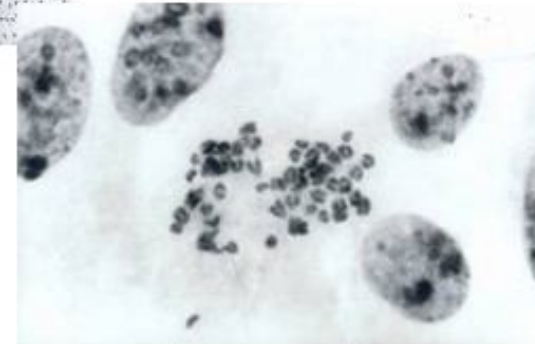
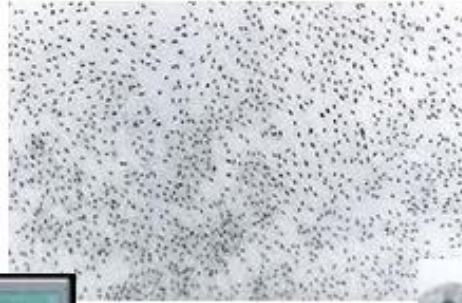


- Results are ready in 48 hours
- 99% accurate
- Coloured probes identify the three chromosomes of chromosome 21



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## Baby's Chromosomes are studied by (2) Culture test ( Confirmatory Test )



- Baby's Cells are grown in special media
- Results come in three weeks
- The Chromosomes of the baby are studied under the microscope





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- 80 % of women bear children between 20 to 30 years of age and are classified as Low risk.
- They should be advised Screening tests which will predict their risk of having a Downs syndrome baby.
- All women should be Counseled ( the nature of the tests and it's implications ) before they opt for screening tests.





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## Screening tests between 11 to 14 weeks of pregnancy ( by ultrasound )



Ultrasound done through  
the tummy



Skin fold behind babys  
neck - (1) **Nuchal translucency**  
( normal < than 3 mm )



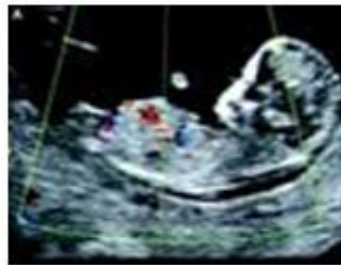
Abnormal skin fold



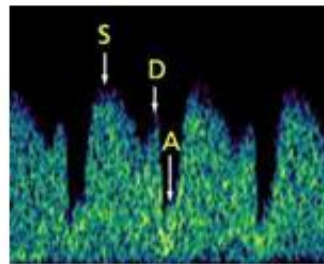
( 2 ) **Nasal bone** present



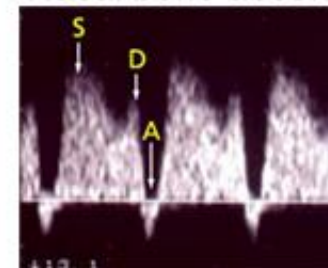
Nasal bone absent



(3) Blood flow in blood vessel  
in baby's tummy



Normal Blood flow



Abnormal Blood flow





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Screening tests between 11 to 14 weeks of pregnancy

### Double Marker Test

- ( 1 ) A blood test performed on the mother's blood
- ( 2 ) Two biochemical markers of pregnancy analysed
  - a) PAPP - A : Pregnancy associated plasma protein
  - b) Free beta hCG : human chorionic gonodotrophin
- ( 3 ) In Down's Syndrome : PAPP - A is reduced  
Free beta hCG is raised above normal





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## Screening tests between 16 to 22 weeks of pregnancy

### Triple Marker Test

Abnormality	AFP	hCG	$\mu$ E3
Neural Tube Defects	↑	—	—
Trisomy 21	↓	↑	↓
Trisomy 18	↓	↓	↓

**DOWNS SYNDROME**

( 1 ) A blood test performed on the mother's blood

( 2 ) Three biochemical markers of pregnancy analysed

- a ) AFP : Alpha feto protein
- b) Free beta hCG
- c) E3 : Estriol





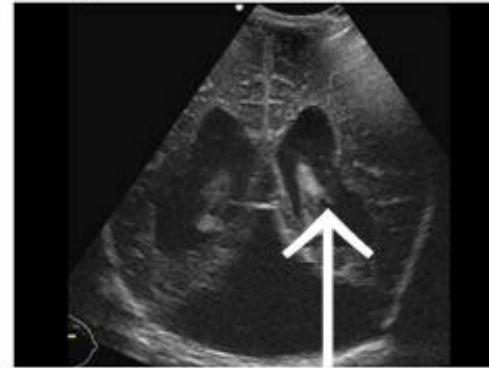
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## Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy ( Markers for Downs Syndrome )

### Screening Test



Normal Ventricle of baby's brain



Enlargement of Ventricle  
of baby's brain



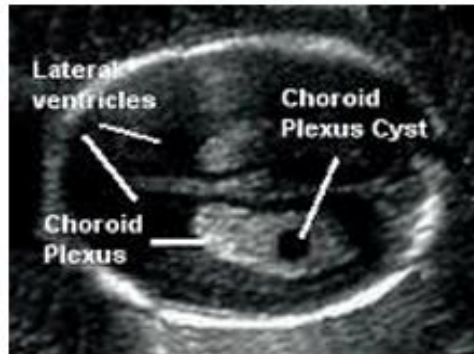
Thickened skin fold behind  
baby's neck



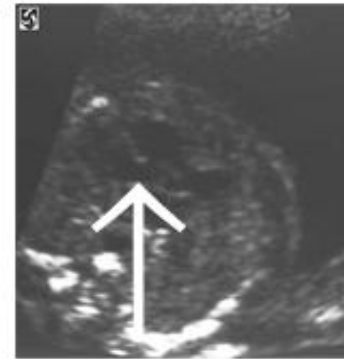


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## Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy ( Markers for Downs Syndrome )



Cyst in baby's brain



Hole in baby's heart



Normal Stomach of the baby



Obstruction of the baby's intestine





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## Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy ( Markers for Downs Syndrome )



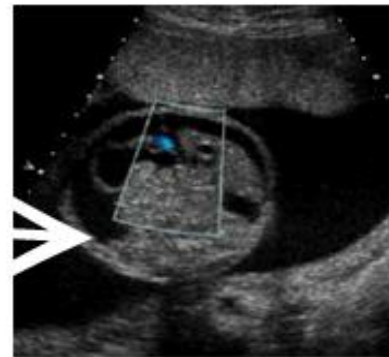
Bright intestine of baby



Collection of fluid in the pelvis  
of the baby's kidney



Fluid collection in the skin  
and inner cavities of the baby





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## 3 Dimensional view of Baby's face having DOWN SYNDROME

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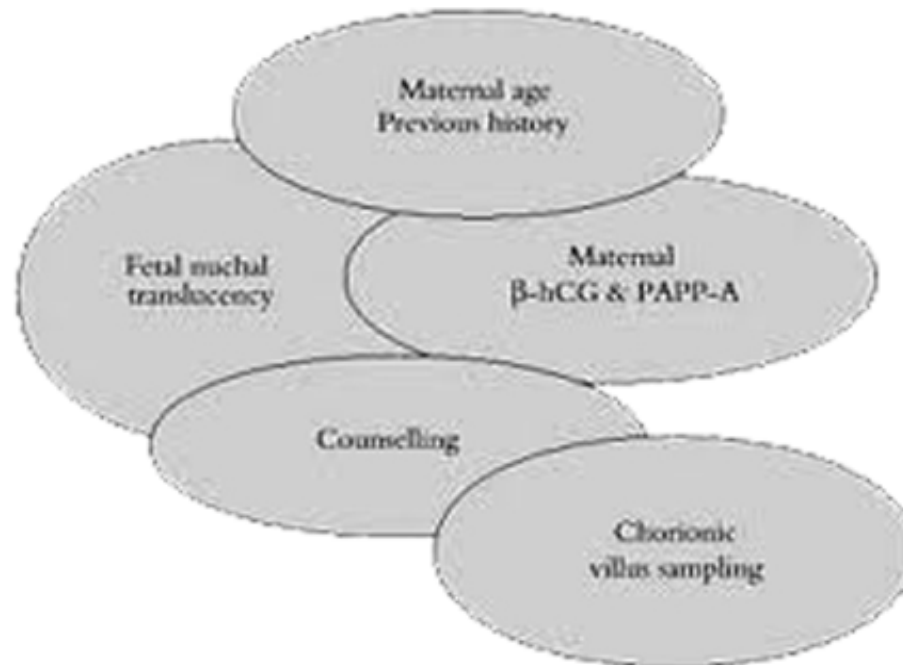


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# One Stop Clinics for Assessment of Risk



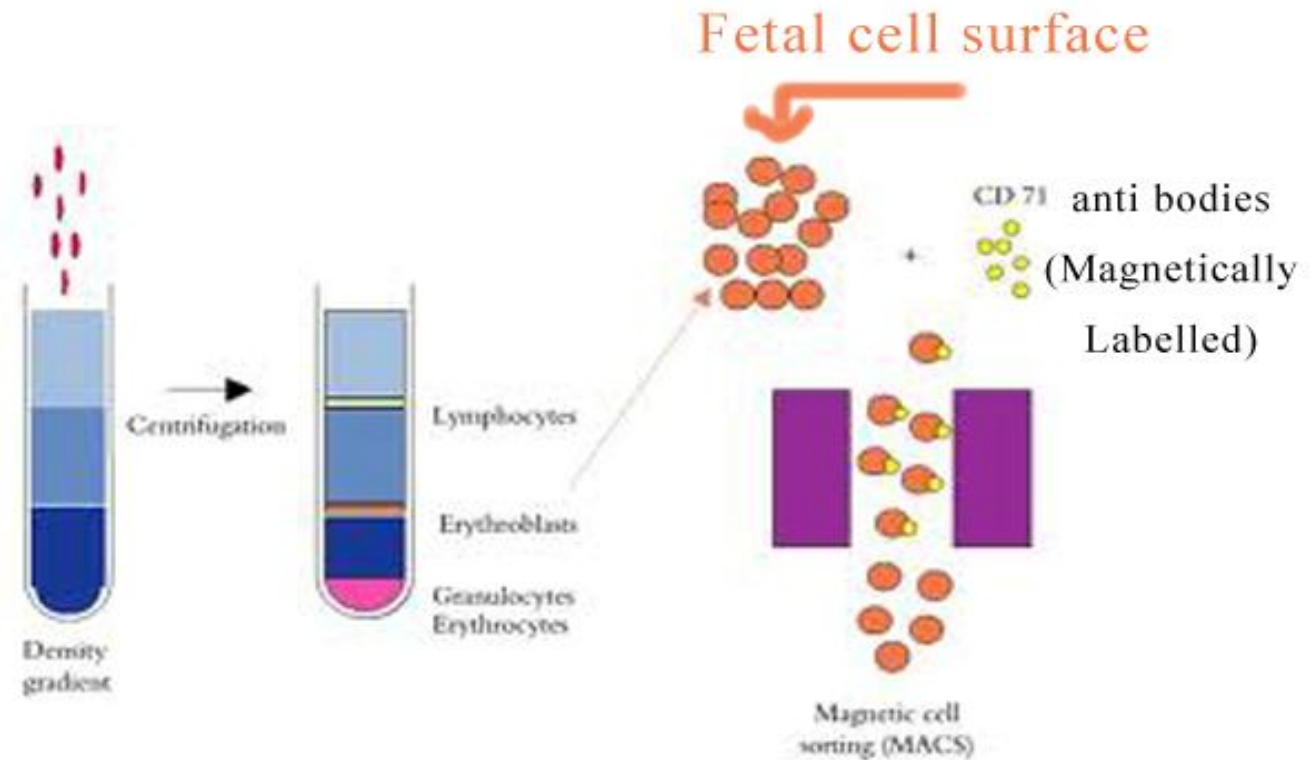
Where the woman can find out her risk of having a Down syndrome baby in one visit to this specialised clinic





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# Non Invasive Diagnosis using fetal cells from maternal blood



( Extensive Research being carried out)

*The End*