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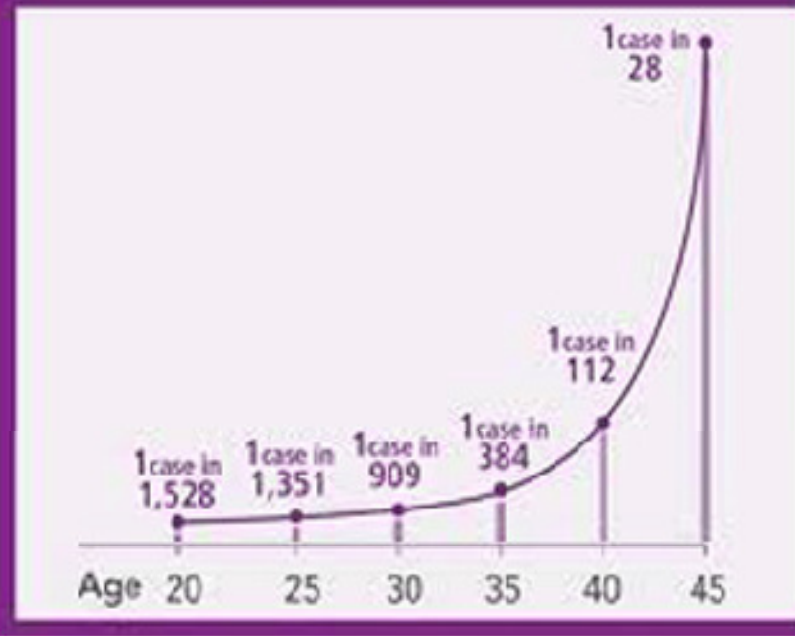
- Cause Discovered by Jerome Lejeune
- Genetic condition in which a person has 47 chromosomes instead of the usual 46
- There is an extra copy of chromosome 21, therefore Trisomy 21





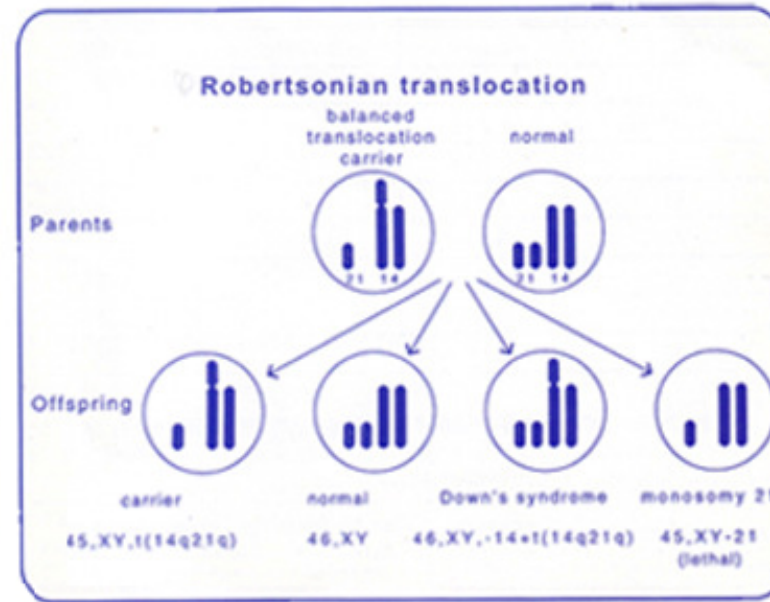
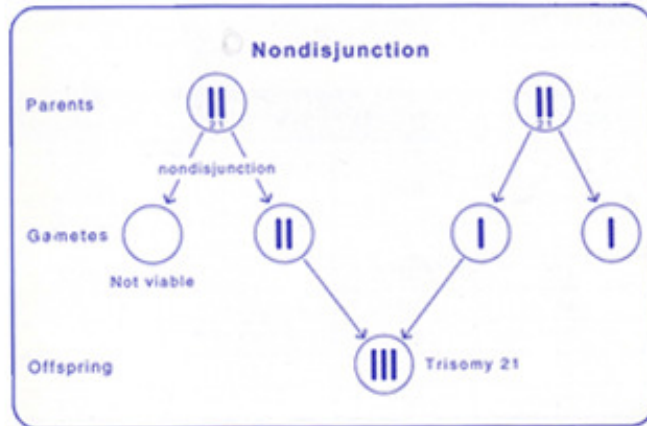
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PROBABILITY OF GIVING BIRTH TO A BABY WITH TRISOMY 21 BY WOMAN'S AGE



- 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

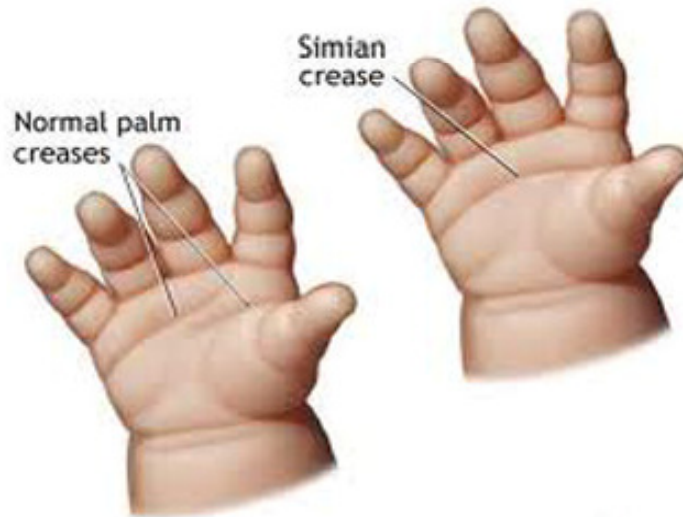
← Small Chin



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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



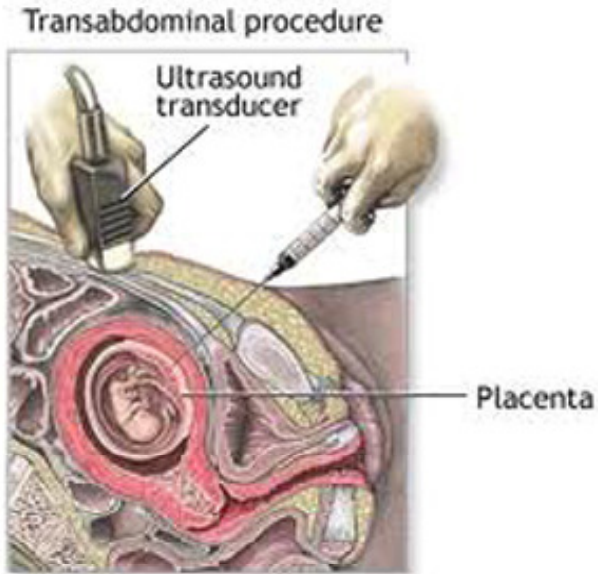
- Done under continuous ultrasound guidance
- Needle is inserted in the mothers womb
- Testing of fetal tissue samples





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Chorionic Villus Sampling



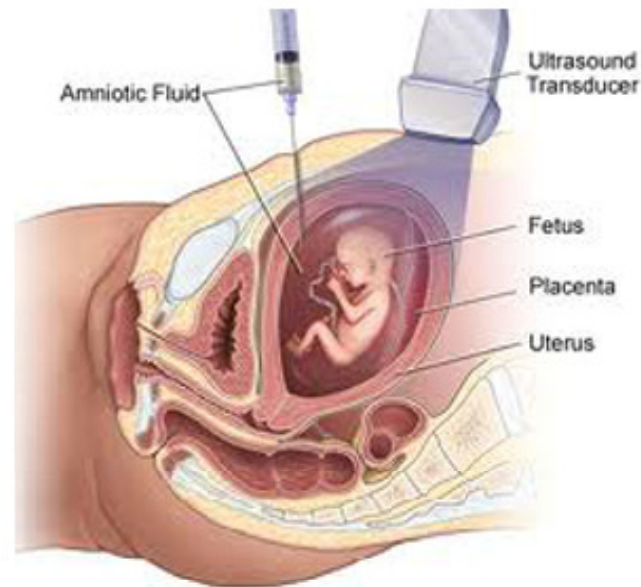
- Done between 11 to 13 weeks of pregnancy
- Removing a piece of placenta (Afterbirth)





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Amniocentesis



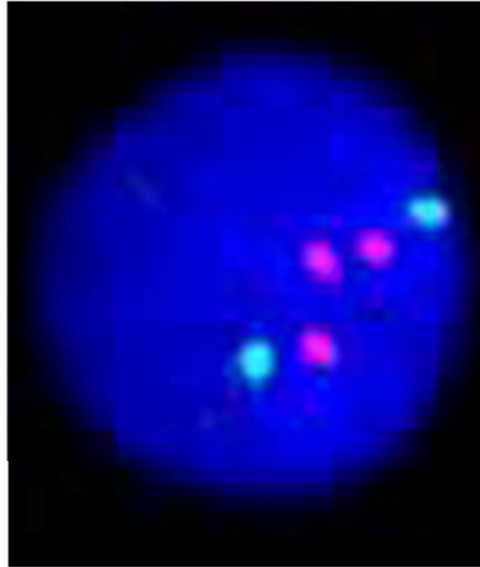
- Done between 16 to 20 weeks of pregnancy
- Removing the fluid around the baby





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

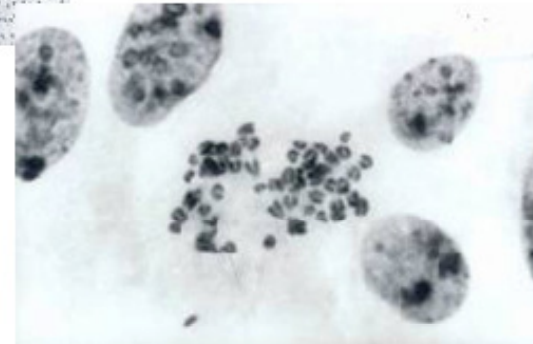


- Results are ready in 48 hours
- 99% accurate
- Coloured probes identify the three chromosomes of chromosome 21



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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 baby's
neck - (1) **Nuchal translucency**
(normal < than 3 mm)



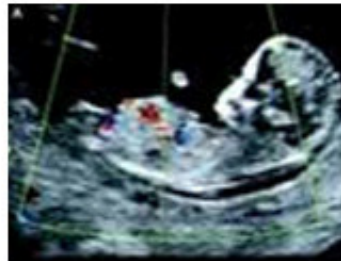
Abnormal skin fold



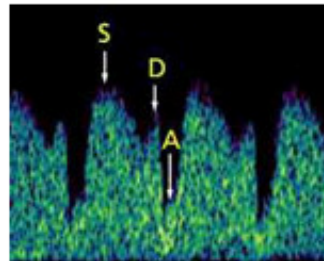
(2) **Nasal bone** present



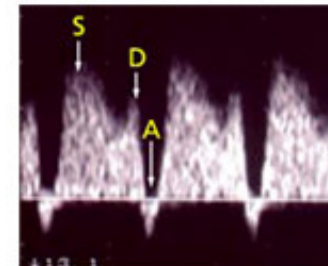
Nasal bone absent



(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 Downs Syndrome : PAPP - A is reduced
Free beta hCG is raised above normal





Screening tests between 16 to 22 weeks of pregnancy

Triple Marker Test

Abnormality	AFP	hCG	μ E3
Neural Tube Defects	↑	—	—
Trisomy 21	↓	↑	↓
Trisomy 18	↓	↓	↓

DOWN'S 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



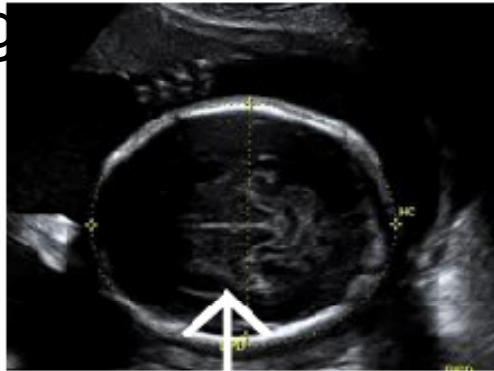


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Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy (Markers for Downs Syndrome)

Screening Test

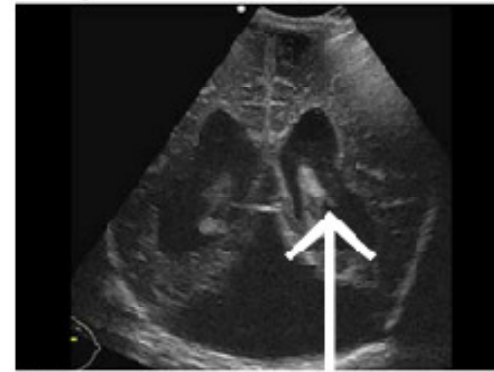
Normal



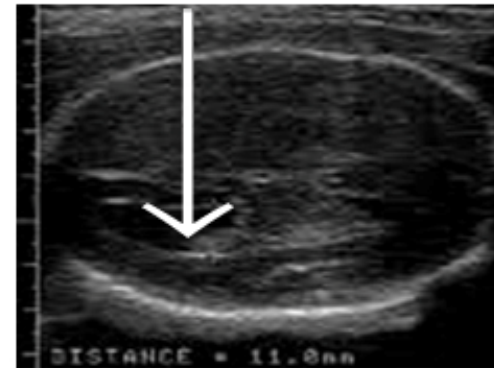
Ventricle of baby's brain



Thickened skin fold behind
baby's neck



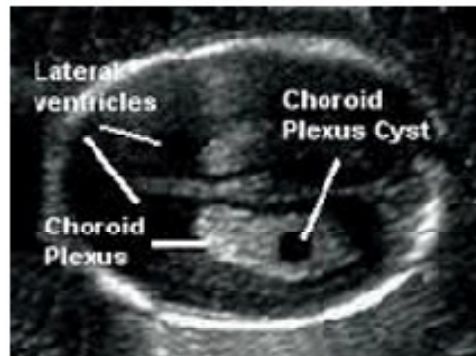
Enlargement of Ventricle
of baby's brain



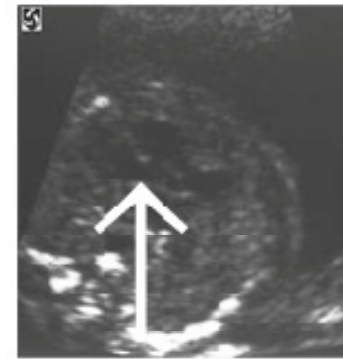


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Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy (Markers for Downs Syndrome)



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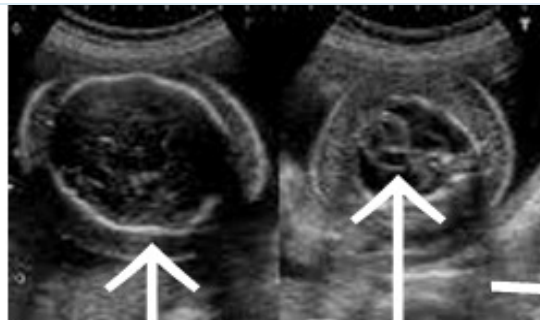
Detailed Ultrasound of the anatomy of the baby at 20 weeks of Pregnancy (Markers for Downs Syndrome)



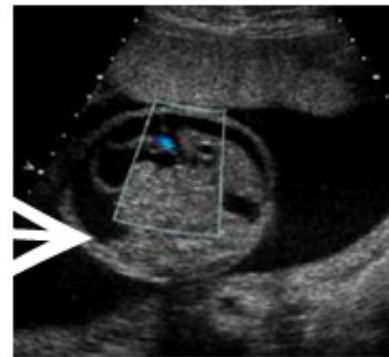
Bright intestine of baby



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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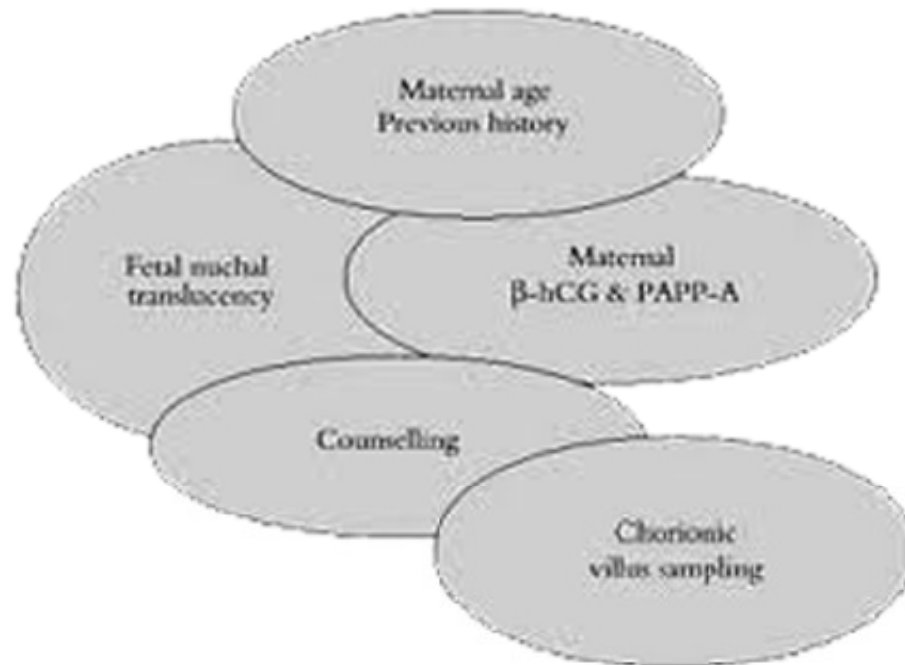


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One Stop Clinics for Assessment of Risk



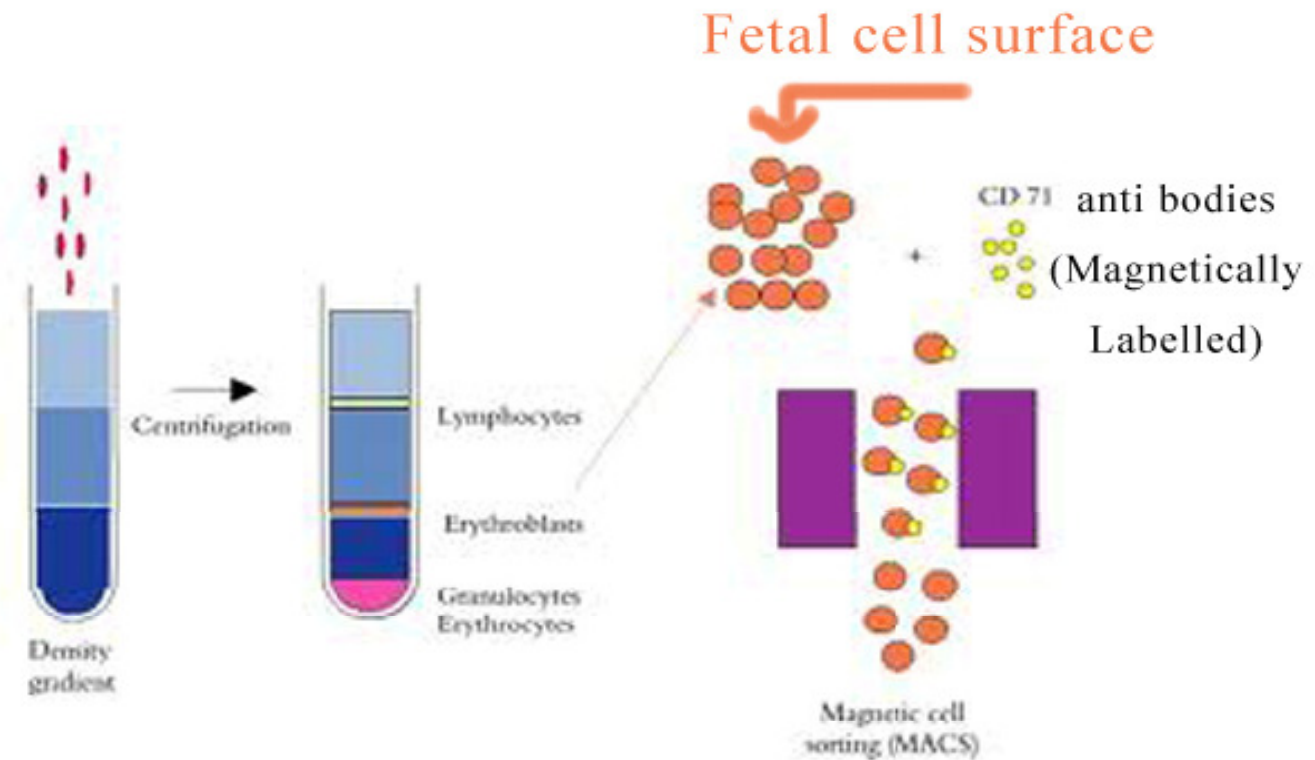
Where the woman can find out her risk of having a Down syndrome baby in one visit to this specialised clinic





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Non Invasive Diagnosis using fetal cells from maternal blood



(Extensive Research being carried out)

The End