Prenatal Diagnosis of Inherited Bleeding Disorders

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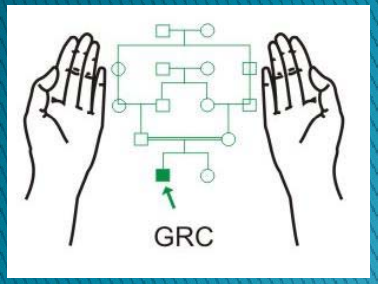

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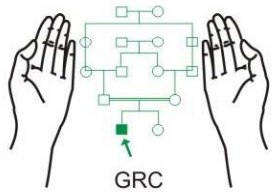
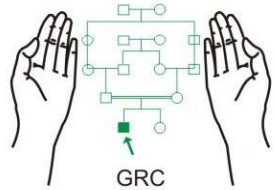
Inherited Bleeding Disorders

- Presence since birth
- Another affected family member
- But not always!



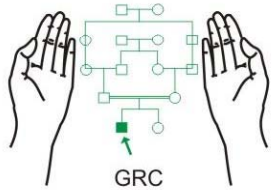
Inherited Bleeding Disorders

- X-Linked
  - Haemophilia-A
  - Haemophilia-B
- Autosomal
  - von Willebrand Disease
  - Uncommon coagulation factor deficiencies
  - Platelet Function Disorders



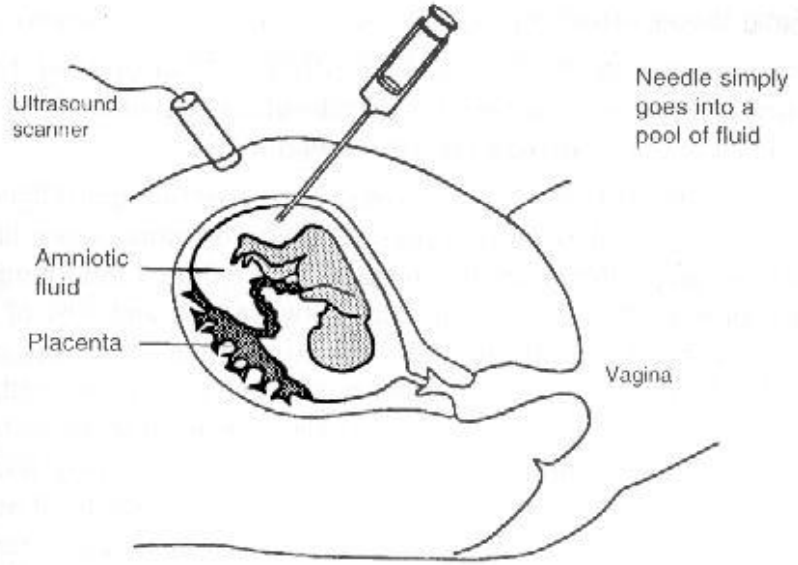
Non Invasive Prenatal Diagnosis

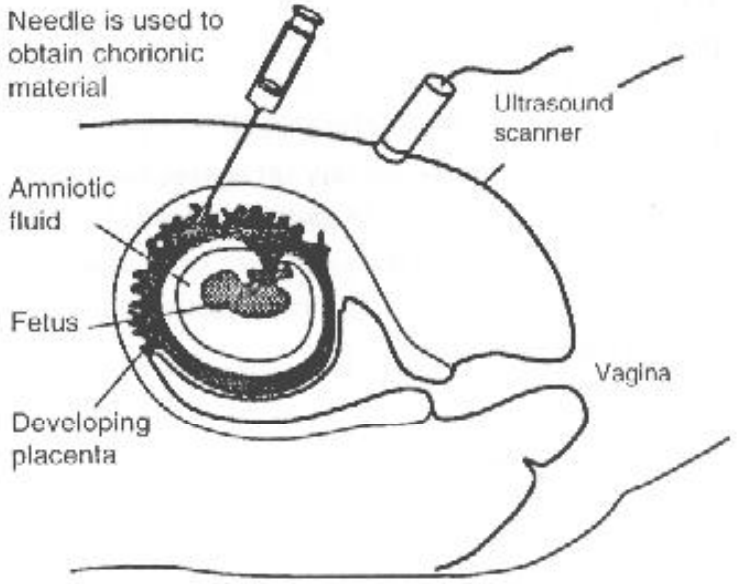
- Fetal Sex determination for X-Linked Disorders
- Ultrasonography
  - 70.3% at 11 weeks
  - 98.7% at 12 weeks
  - 100% at 13 weeks
    - (Efrat et al, 1999)
- Fetal DNA in maternal blood
  - 100% after 10 weeks
    - (Hromadnikova, et al 2003)
- Pre-Implantation Genetic Diagnosis (PGD)

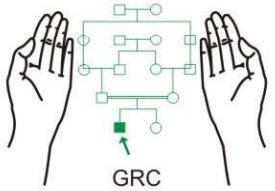


Invasive Prenatal Diagnosis

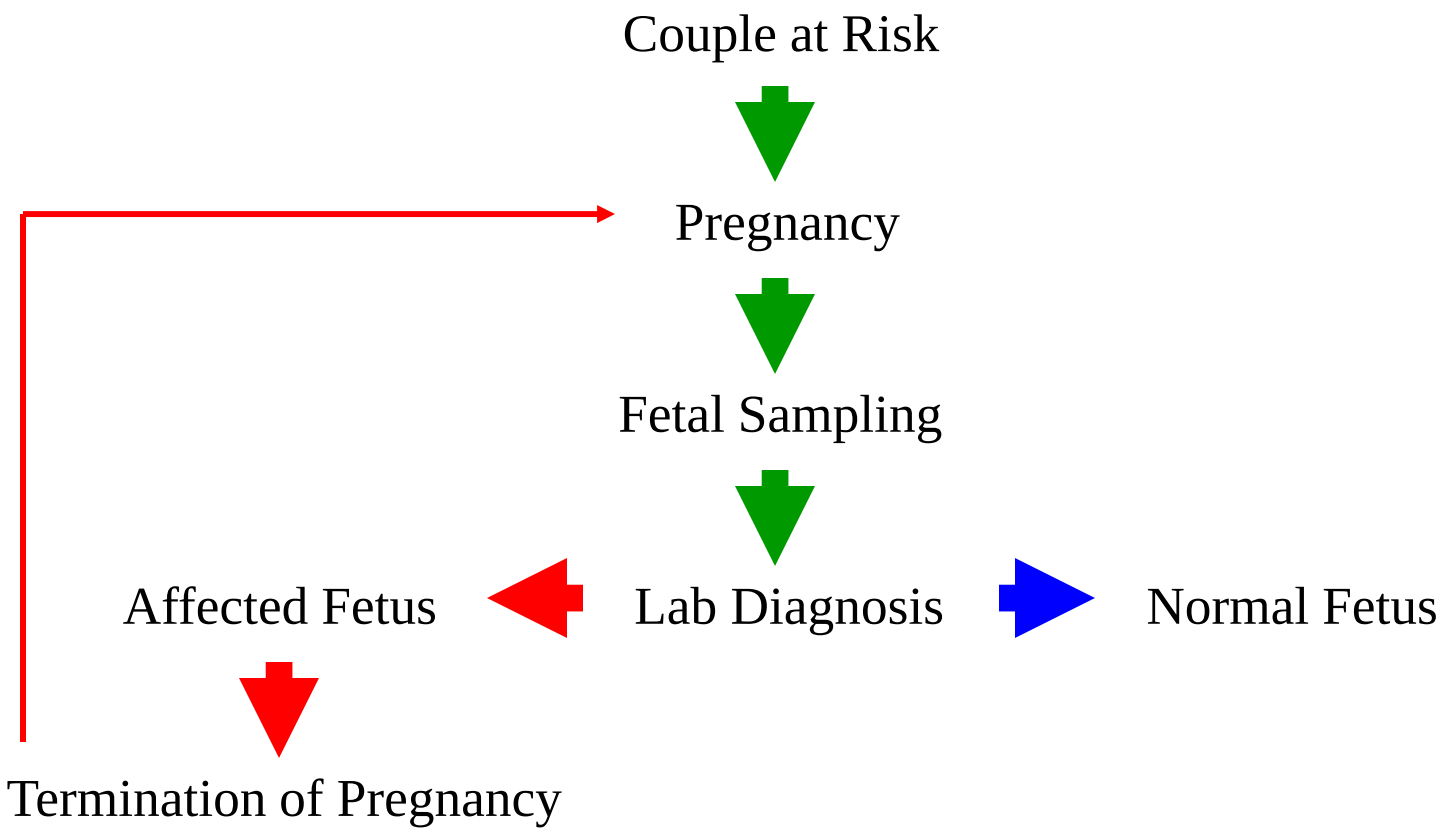
- Chorionic Villus Sampling (CVS)
- Amniocentesis

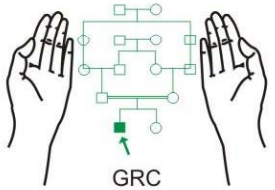


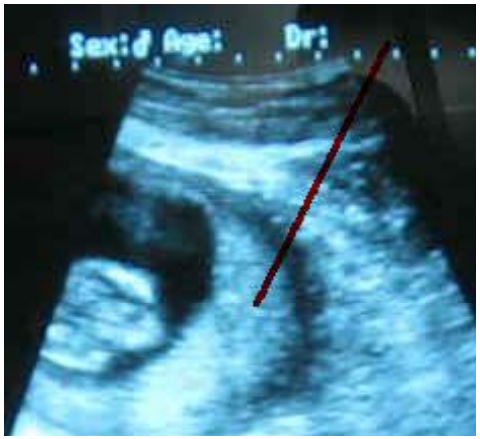


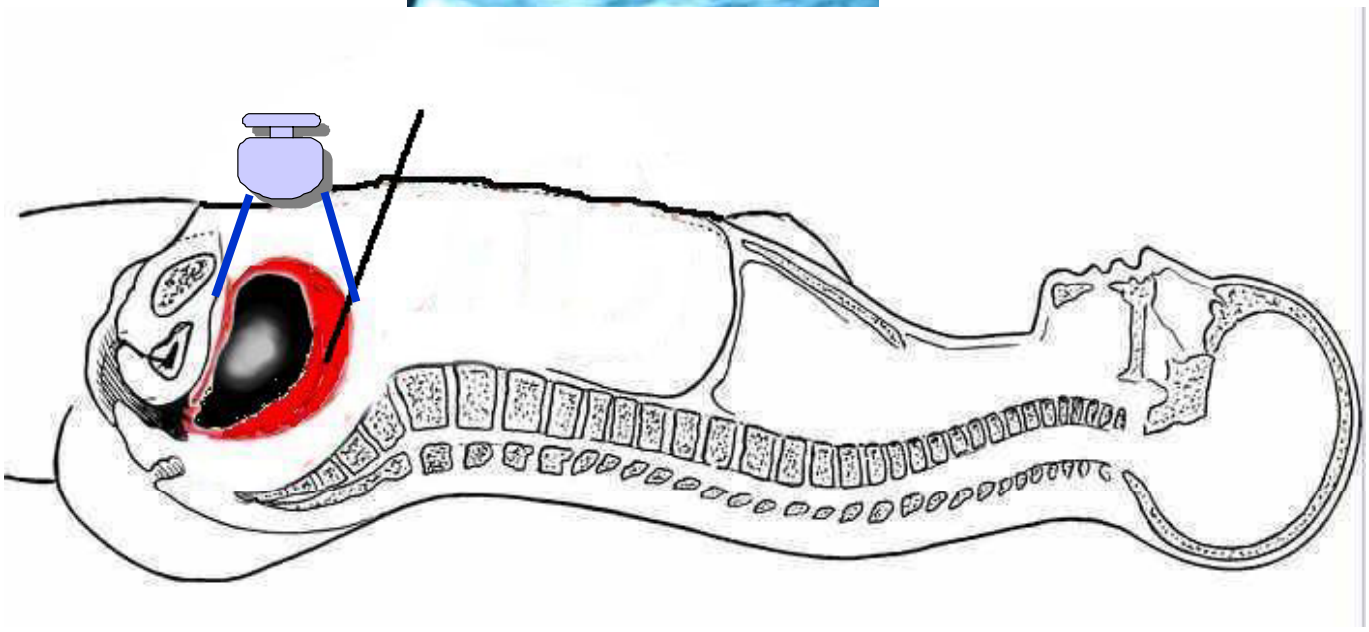


Prenatal Diagnosis









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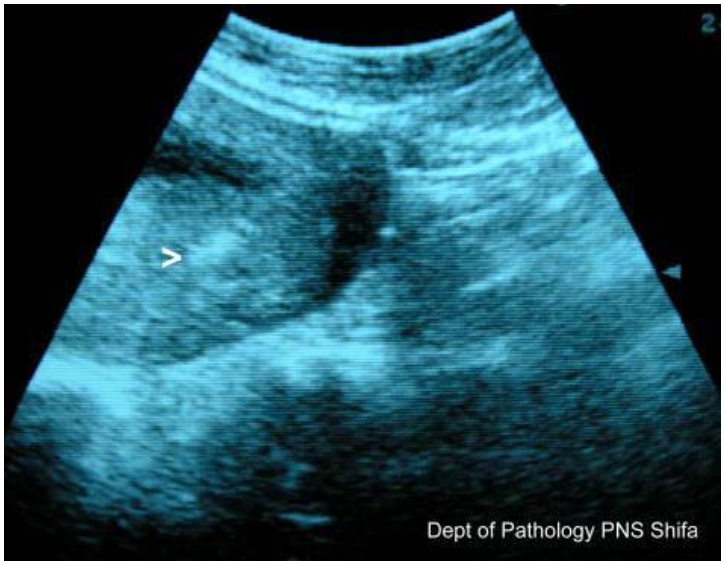

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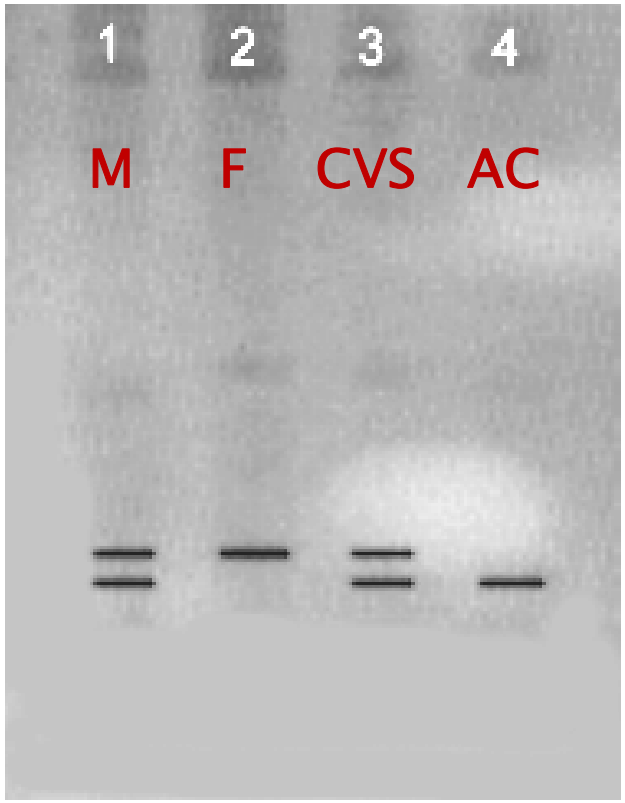
DNA Testing

- Mutation analysis
- Linkage analysis



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