GENETIC COUNCLING AND PRENATAL DIAGNOSIS

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Genetic Counselling

INTRODUCTION

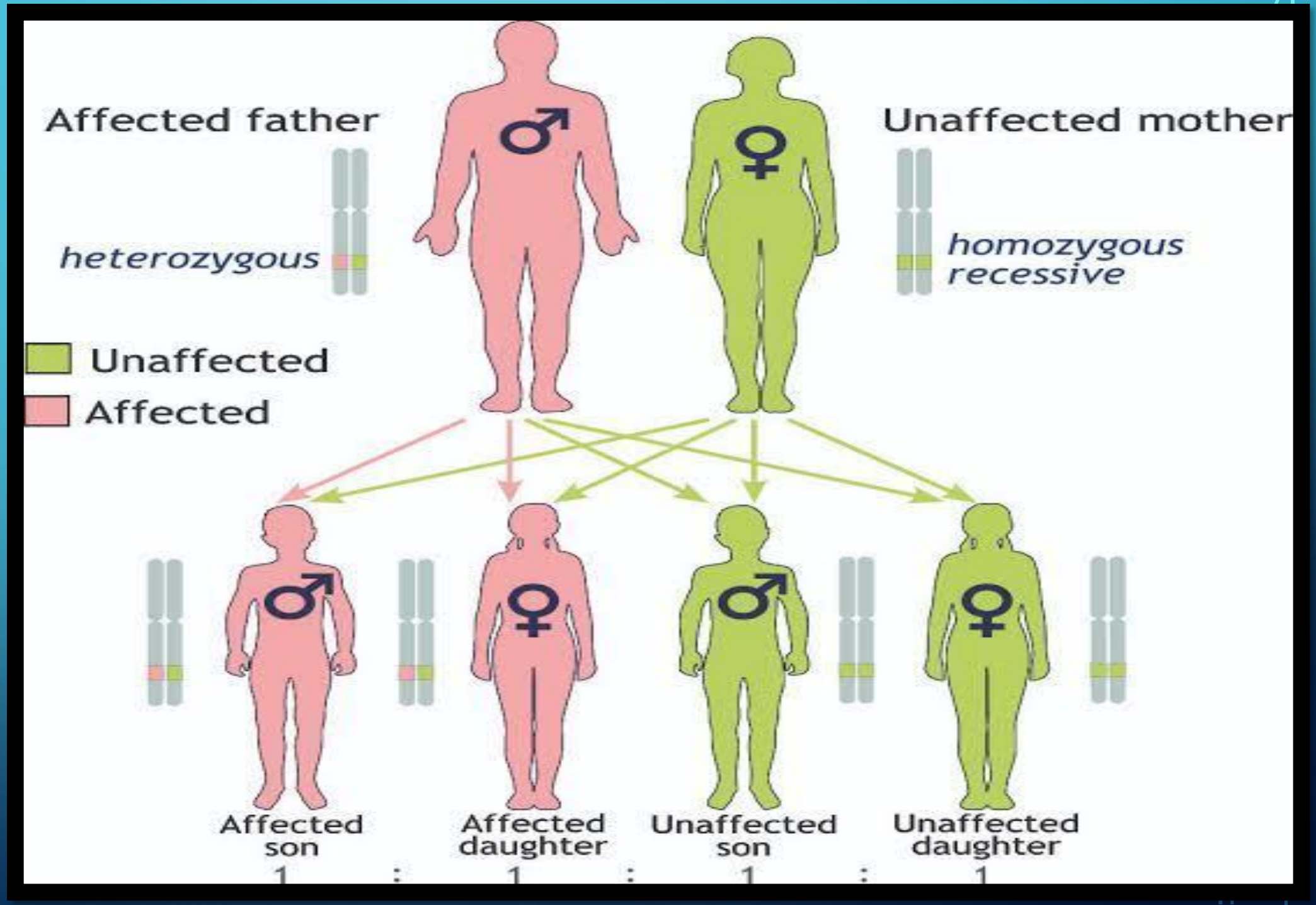
Genetic counseling is important in dermatological practice. Estimation of genetic risks requires accuracy.

Prenatal diagnosis (PND) may be chosen for severe genodermatoses. It is essential that PND be based on ethical considerations.

PURPOSE OF GENETIC COUNSELLING

Purpose of genetic counsellingProvide concrete, accurate information about inherited disorders.

- Reassure people who are concerned that their child may inherit a particular disorder that the disorder will not occur.
- Allow people who are affected by inherited disease to make informed choice about future reproduction.
- Educate people about inherited disorder and the process of inheritance.
- Offer support by skilled health care professionals to people who are affected by genetic disorders.


Affected father
heterozygous
Unaffected mother
homozygous recessive
Unaffected
Affected
Affected son
Affected daughter
Unaffected son
Unaffected daughter
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INDICATION

1.Advanced parental age:

Maternal age =35 yrs

Paternal age =50 yrs.

2. Previous child with or family H/O:

- Congenital anomaly
- Dysmorphism
- Intellectual disabilitY
- Developmental delay
- Isolated birth defect
- Metabolic disorder
- Chromosomal abnormality
- Myopathy/ Neuropathy
- Ambiguous genitalia

3.ADULT ONSET GENETIC DISORDER (PRESYMPTOMATIC TESTING):

CANCER

4.CONSANGUINITY

5. TERATOGEN EXPOSURE

6. REPEATED PREGNANCY LOSS OR INFERTILITY

7.PREGNANCY SCREENING ABNORMALITY

- MATERNAL SERUM A-FETO PROTEIN
- MATERNAL TRIPLE OR QUAD TEST
- FETAL ULTRASONOGRAPHY
- FETAL KARYOTYPE8.HETEROZYGOTE

8.SCREENING BASED ON ETHNIC RISK

SICKLE CELL ANEMIA

TAY-SACHS

CANAVAN

GAUCHER DISEASE

THALASSEMIA

9.FOLLOW UP TO ABNORMAL NEONATAL GENETIC TESTING

TYPES OF GENETIC COUNSELLING

TWO TYPES OF GENETIC COUNSELLING:

(A) PROSPECTIVE GENETIC COUNSELLING:

- THIS ALLOWS FOR THE TRUE PREVENTION OF DISEASE.
- THIS REQUIRES TO IDENTIFY HETEROZYGOUS INDIVIDUALS FOR ANY PARTICULAR DEFECT BY SCREENING.
- EXPLAINING TO THEM THE RISK OF THEIR HAVING AFFECTED CHILDREN IF THEY MARRY ANOTHER HETEROZYGOTE FOR THE SAME GENE.
- IF HETEROZYGOUS MARRIAGE CAN BE PREVENTED OR REDUCED, THE PROSPECTS OF GIVING BIRTH TO AFFECTED CHILDREN WILL DIMINISH.
- EX: SICKLE CELL ANAEMIA
  THALASSEMIA

(B) **RETROSPECTIVE GENETIC COUNSELLING:**• MOST :-GENETIC COUNSELING AT PRESENT IS RETROSPECTIVE, (THE HEREDITARY DISORDER HAS ALREADY OCCURRED WITHINTHE FAMILY).·

EX. MENTAL RETARDATION
PSYCHIATRIC ILLNESS
INBORN ERRORS OF METABOLISM

:-THE METHODS WHICH COULD BE SUGGESTED UNDER RETROSPECTIVE GENETIC COUNSELING ARE:

-CONTRACEPTION
-PREGNANCY TERMINATION
-STERILIZATION

METHODS OF PRENATAL DIAGNOSIS

(A) IMAGING:- ULTRASOUND
MRI

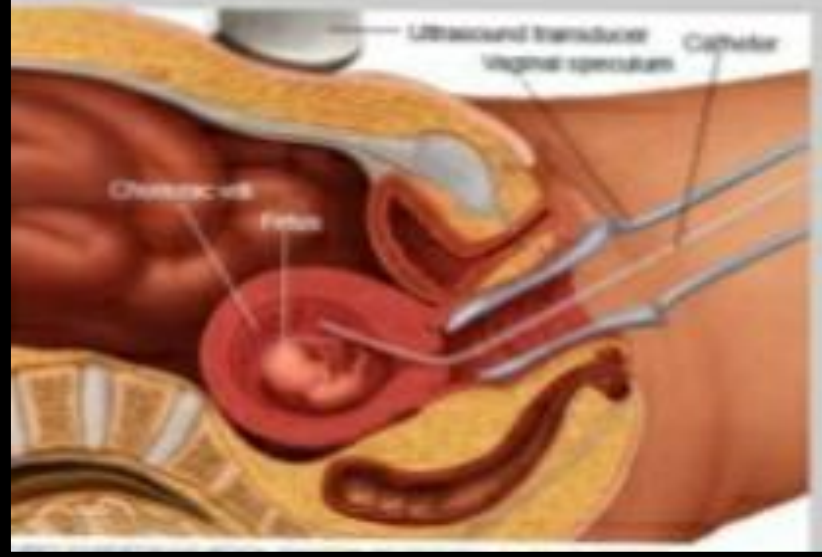
(B) FLUID ANALYSIS:- AMNIOCENTESIS
CORDOCENTESIS

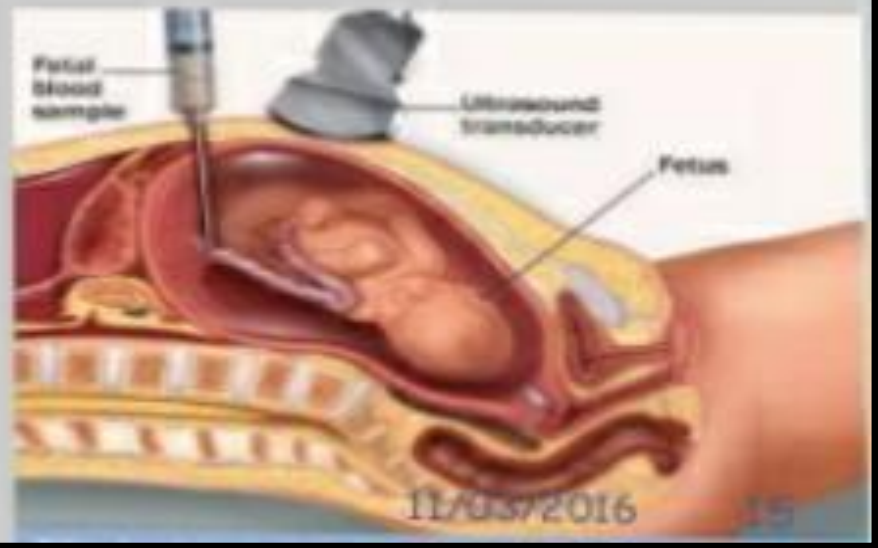
(C) FETAL TISSUE ANALYSIS- CHORIONIC VILLUS SAMPLING

(D) MATERNAL SERUM TESTS:-@FETO PROTEIN
TRIPLE TEST
QUAD TEST

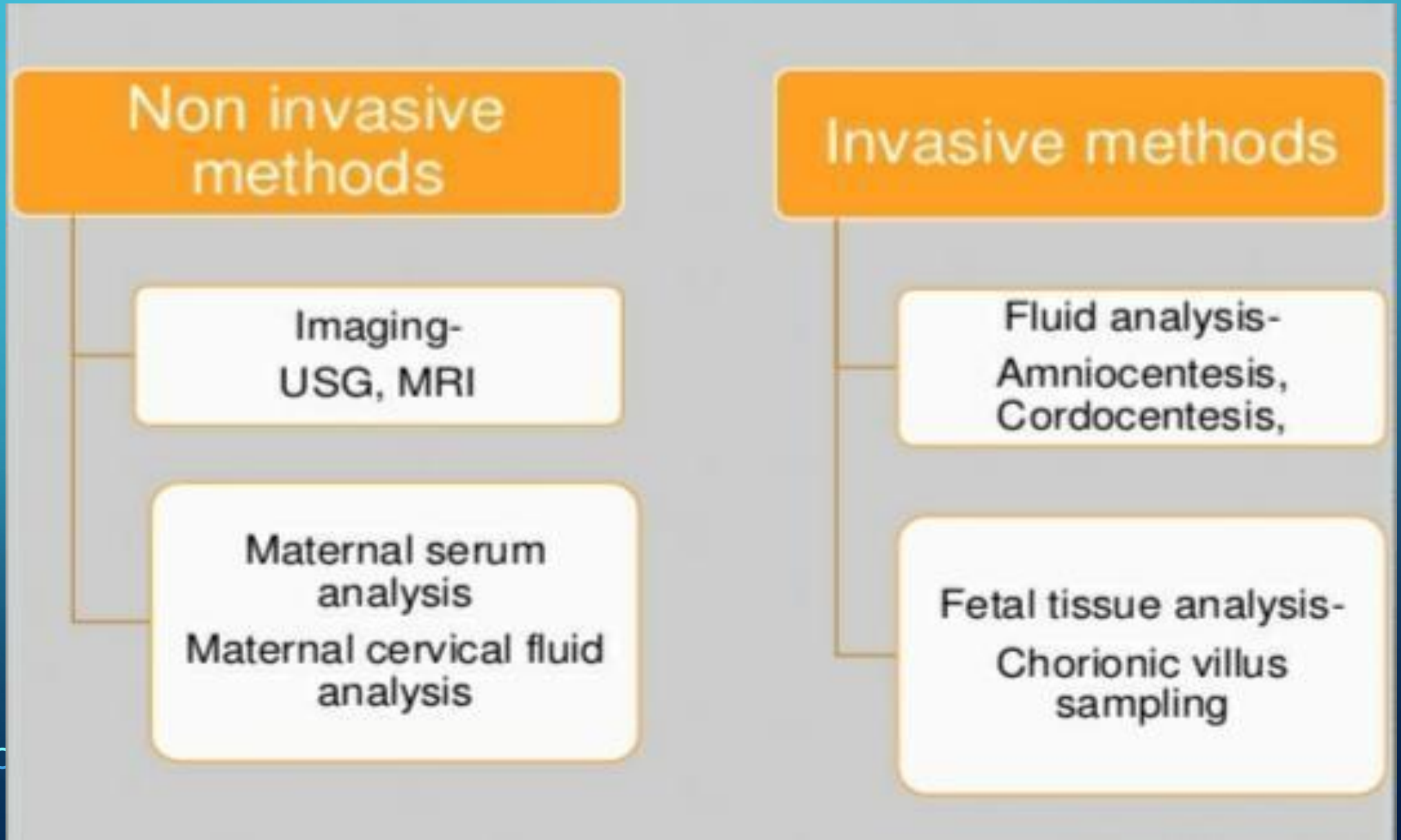
(E) MATERNAL CERVIX:- FETAL FIBRONECTIN
FLUID AND BACTERIAL CULTURE

Prenatal diagnosis





METHOD OF GENETIC COUNSELING



QUESTIONS

1. WHAT IS THE GENETIC COUNSELING?
2. WHAT ARE THE TYPES OF GENETIC COUNSELING?

THANK YOU