

**Objective**

To understand definition, requirement and indications of genetic counseling and prenatal diagnosis

**Prenatal diagnosis**

***Indications:***

- Increased risk for malformation
- Chromosomal abnormality or other genetic disorder
- Advanced maternal age >35yr

Many different methods are available, depending on the specific genetic disorder including:

- Ultrasonography for the diagnosis of anatomic abnormalities such as congenital heart defects
- Amniocentesis and chorionic villous sampling are used to obtain fetal tissue for analysis of chromosomal abnormalities, biochemical disorders, and DNA studies.
- Maternal blood or serum sampling is used for some types of screening.
- Fetal blood from umbilical cord can also be used for testing.

## **Genetic counseling**

Genetic counseling is a communication process in which the genetic contribution to health is explained, along with specific risks of transmission of a trait, and options to manage the condition and its inheritance.

### ***Providing accurate information to families requires:***

- 1- Taking a careful family history and constructing a pedigree that lists the relatives with their sex, age and state of health.
- 2- Gathering information from hospital records about affected individual.
- 3- Documenting prenatal, pregnancy and delivery histories.
- 4- Reviewing the latest available medical, laboratory and genetic information concerning the disorder.
- 5- Careful physical examination of affected individual.
- 6- Establishing the diagnosis by diagnostic tests.

### **Indications for genetic counseling:**

- 1- Advanced parental age.

Maternal age > 35yr

Paternal age > 50yr

- 2- Previous child with or family history of:

- congenital abnormality
- dysmorphology
- mental retardation
- isolated birth defect
- Metabolic disorder
- Chromosome abnormality

- Single gene disorder
- 3- Consanguinity
- 4- Teratogen exposure
- 5- Heterozygote screening eg. SCA3
- 6- Repeated pregnancy loss
- 7- abnormal neonatal genetic testing

### **Genetic counseling of Down Syndrome**

Congratulate the parent for having a new baby and assure them that the doctor/ genetic counselor is there to support them.

Why has this happened?

A genetic disorder incidence: 1/600-800 births

#### ***Causes:***

- Trisomy 21 (due to nondisjunction) 95%
- Translocation 4%
- Mosaic 1%

Trisomy 21 ( nondisjunction ) and maternal age

Age(year) Risk

< 25 1/600-800

>35 1/350

>40 1/100

#### **Will it happened again?**

recurrence risk :

\*translocation t( 21; 21) 100%

For : t( 13;21 ), t( 14;21 ), t( 15;21 )

if the mother is the carrier 12%

if the father is the carrier 3%

\*If a couple has a child with Trisomy 21( nondisjunction ), the risk is higher for the next pregnancy ( 1/100 ).

### **Prenatal Diagnosis**

1- Maternal screening

a- First trimester- US for nuchal translucency, blood for PAPP(pregnancy associated plasma protein)(decreased) and hcg ( increased )

b- Second trimester- AFP (increased), hcg, and estriol ( decreased )

2- Diagnostic tests

chorionic villous sampling at 9-11week

amniocentesis at 14-18 weeks

### **IQ ?**

The average IQ is 50-60 but it varies.5

complications

have high risk to develop:

1- Hypothyroidism

2- DM

3- Celiac disease

4- Leukemia

5- Congenital heart disease

6- Hearing loss

7- Epilepsy

Health care and maintenance

- Echocardiogram at time of diagnosis

- Hearing and Vision assessment

- Thyroid function test

- Health supervision for children with Down Syndrome