Lecture 3 ***Dr. miami***

Genetic counseling and Prenatal diagnosis

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. SCA

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 (1/1oo).

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.

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