University of Basrah
Al-Zahraa Medical College

Ministry of Higher education
and Scientific Research

Block: child health

Lecture: genetic counseling

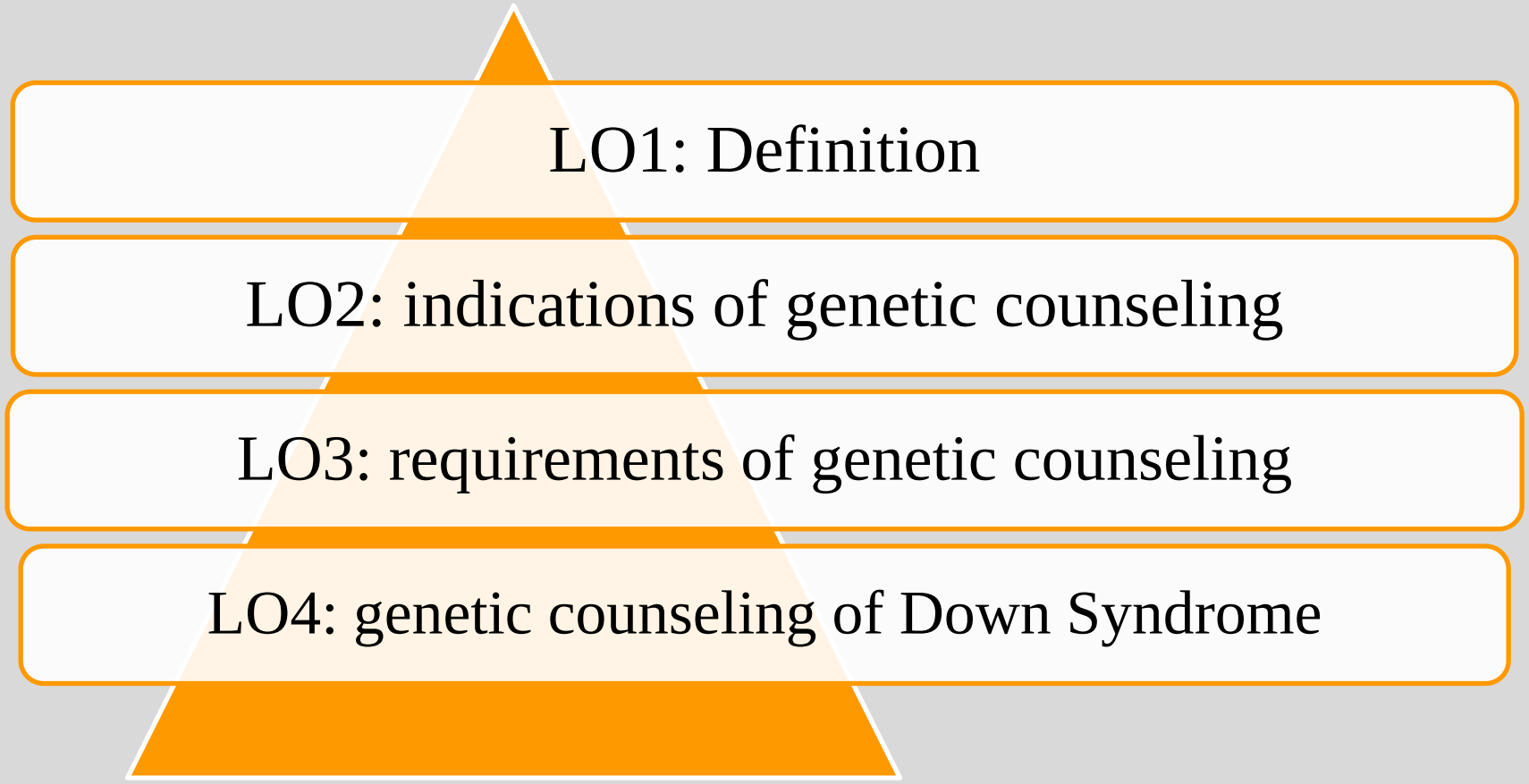
Lecturer: Dr. Miami Kadhim

Block staff:

Dr. Jawad Ramadan (supervisor)
Dr. Miami kadhim (leader)
Dr. Rehab abdulwehab (co-leader)
Dr. Ahmad Jaafar (co-leader)
Dr. Ahmad Alsamak (coordinator)
Dr. Dhaighum Imad
Dr. Khalid Ahmad
Dr. Basim Abdulkareem
Dr. Ahmed Ibrahim
Dr. Dheyaa Jameel

Nelson Textbook of Pediatrics, 20th edition.
Nelson Essentials of Pediatrics, 7th Edition 2015
Pediatric Decision-Making Strategies
Illustrated Textbook of Pediatrics
Short Atlas in Pediatrics

Learning outcomes



Genetic Counseling LO1

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

Indications for genetic counseling LO2

1- **advanced parental age**

- **Maternal age >35 yr**
- **Paternal age > 50 yr**

2- previous child with or family history of:

- **Congenital abnormality**
- **Dysmorphology**
- **Mental retardation**

- **Metabolic disorder**
- **Chromosomal abnormality**
- **Single gene disorder**

3- consanguinity

4- teratogen exposure

5-heterozygote screening

6-repeated pregnancy loss

7- abnormal neonatal genetic testing

providing accurate information to families requires: LO3

1- taking a careful family history and constructing a pedigree that lists the relatives with their sex, age and state of health

2- documenting prenatal, natal and postnatal histories

3- careful physical examination

4-reviewing the latest available laboratory and genetic information concerning the disorder

5- establishing the diagnosis by diagnostic tests

Genetic counseling of Down Syndrome

LO4

Congratulate the parents for having a new baby and assure them that the doctor / genetic counselor are there to support them

Why has this happened?

A Genetic Disorder

Incidence: 1/ 600-800 births

- **Trisomy 21 95%**
- **Translocation 4%**
- **Mosaic 1%**

Will it happen again ?

Recurrence Risk

Trisomy 21 (nondisjunction) and maternal age

- **Age (year)** **Risk**

- **< 25** **1/ 600-800**
- **> 35** **1/ 350**
- **> 40** **1/ 100**
- **If a couple has a child with DS the risk is higher for the next pregnancy (1/ 100)**

Translocation

Frequency : 1/ 500

Recurrence Risk

- **T(21;21) 100%**
- **T (14;21) , t (13;21) , t (15;21)**

12% if the mother is the carrier

3% if the father is the carrier

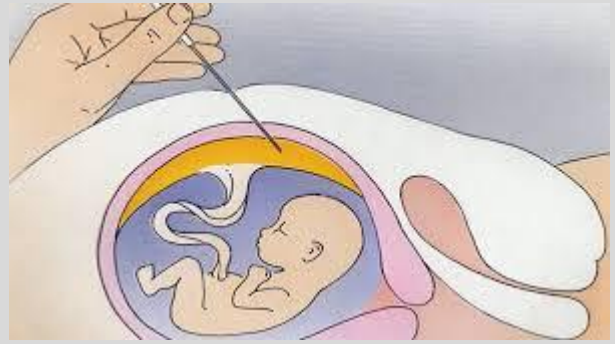
What can be done about it ?

Prenatal diagnosis:

1- maternal screening

- **first trimester - US for nuchal translucency, blood for PAPP(pregnancy associated plasma protein) -A (decreased) and B hcg (increased)**
- **second trimester - AFP (increased), hcg, estriol (decreased)**

2- diagnostic testing - chorionic villous sampling at 9-11weeks, Amniocentesis at 14-18 weeks



I Q ?

The average IQ is 50 – 60 but it varies

complications

High risk for development of:

- **Hypothyroidism**
- **D M**
- **Celiac disease**
- **Leukemia**
- **Congenital heart disease**
- **Hearing loss**
- **epilepsy**

Health care and maintenance

- **Echocardiogram at time of diagnosis**
- **Hearing and vision assessment**
- **Thyroid function test**
- **Cervical spine X ray**
- **Health supervision for children with DS**

Thank You