Down Syndrome

Example of an Autosomal Trisomy
Topics to be covered in this session:

• What is Down syndrome?
• How it is formed?
• Cytogenetic types & percentage of each type
• Writing the karyotype in each type
• Phenotype vs. Genotype
• Clinical features
• Special situations in Dx of Down syndrome
• How to deal with a child with Down syndrome
• Management
• Prenatal diagnosis
Apply the same principles here for other chromosomal abnormalities:

“they shall not be discussed here – study them carefully”

Other autosomal trisomies
1. Edward Syndrome
2. Patau syndrome

Sex chromosomal abnormalities
1. Monosomy – Turner syndrome
2. Trisomy –
   a. Klinefelter Syndrome
   b. Super female (XXX) Syndrome
Objectives:
By the end of this session, the students are expected to:

1. Define Down syndrome as an example of autosomal trisomy
2. Recognize why it is important and common
3. List the cytogenetic types of Down syndrome
4. Recognize the importance to specify the cytogenetic type in every child
5. Discriminate the common from the less common clinical features
6. Describe briefly the steps of making the diagnosis and management
7. Appreciate the role of the doctor in counseling of such families
What is Down Syndrome
How it is formed?
What are the cytogenetic types?
How to write the karyotype in each type?
Do different cytogenetic types have different clinical features?

i.e. can we know the defect from the clinical features?

i.e. is there any distinguishing clinical feature for any of the various cytogenetic types?
What are the clinical features of Down Syndrome?
Is there any special situation in Diagnosis of Down syndrome?
How to deal with a child with Down syndrome (i.e. Management)?
Is there any method to diagnose Down syndrome babies before they were born?

i.e. Is there a prenatal Diagnostic method?
THANK YOU