Turner’s Syndrome
Normal !!
Down’s Syndrome
Baby MiH

- Twin baby – “Twin II”

- Born 18/11/2009 @ 33^{+4} /40
  - Abnormal CTG & IUGR
  - Emergency C-section
  - 1.54 kg (3.4 lbs) vs. 1.86kg Twin I (4.1 lbs)

- I.V.F.

- Mother
  - 39 years old
  - Primipara
Baby MiH

- In SCBU…

- Polycythaemia neonatorum

- Jaundice

- Absent red reflex (R) eye

- And an unusual physical appearance…
On examination

- Flat nasal bridge
On examination

- Flat nasal bridge
- Epicanthic folds
On examination

- Flat nasal bridge
- Epicanthic folds
- Low set ears
On examination

- Flat nasal bridge
- Epicanthic folds
- Low set ears
- Prominent single palmar crease
On examination

- Flat nasal bridge
- Epicanthic folds
- Low set ears
- Prominent single palmar crease
- Sandal gap
What else to look for?

- **CNS**
  - Hypotonia → mouth open, tongue protrusion
  - Poor moro-reflex

- **Head**
  - Excess skin on back of neck
  - Flat facial profile
  - Slanted palpebral fissures
  - Anomalous auricles

- **Skeleton**
  - Hyperflexible joints
  - Clinodactyly of 5th finger
  - Dysplasia of the pelvis
Less obvious features

- Eyes
  - Brushfield spots
  - Refractive errors

- Sparse hair

- Atlantoaxial instability

- Congenital Heart Defects: PDA, VSD
  - Usually the cause of mortality
Aetiology

• Trisomy 21

• Non-disjunction (94%)
  – Error in meiosis
Chromosome 21s

Parents

Non-disjunction at meiosis

Gametes

Not viable

Fertilisation

Offspring

Trisomy 21 Down's syndrome
Aetiology

- Trisomy 21

- Non-disjunction (94%)
  - Error in meiosis

- Translocation (5%)
  - “Robertsonian”
  - Are the parents a translocation carrier?
Parents

Translocation carrier

Gametes

Offspring

Normal

Translocation carrier

Monosomy 14 Not viable

Trisomy 14 Not viable

Monosomy 21 Not viable

Translocation Down's syndrome

Normal
Aetiology

• Trisomy 21

• Non-disjunction (94%)
  – Error in meiosis

• Translocation (5%)
  – “Robertsonian”
  – Are the parents a translocation carrier?

• Mosaicism (1%)
  – After formation of zygote
  – Error in mitosis
Egg with 23 chromosomes

Sperm with 23 chromosomes

Fertilization

Chromosomes double then the cell divides (mitosis)

Normal cell division continues

Nondisjunction in mitosis (cell does not split evenly)
Cells with +21 continue

These cells die

These cells die

These cells die
Aetiology

- 1 in 800-1000 births
- Male:Female = 1.15:1
- Increasing maternal age
  - 35 years old  1 in 380 (0.26%)
  - 40 years old  1 in 110 (0.91%)
  - 45 years old  1 in 30  (3.33%)
Screening

• History

• End of first trimester (13+6/40)

• Serum screening
  – ↑ β-HCG
  – ↓ PAPP-A

• Imaging
  – Nuchal translucency U/S
Outlook

• Global developmental delay

• IQ: 28-85 (mean 50)

• Increased risk of seizures

• Complications of
  – Eye
  – Heart
  – Hearing
  – GI tract (reflux)
  – Respiratory system (recurrent infections)
  – Spinal cord

• Multi-Disciplinary Approach
Thank you