A Common Dysmorphology

Hitesh Tailor

Paediatrics Module

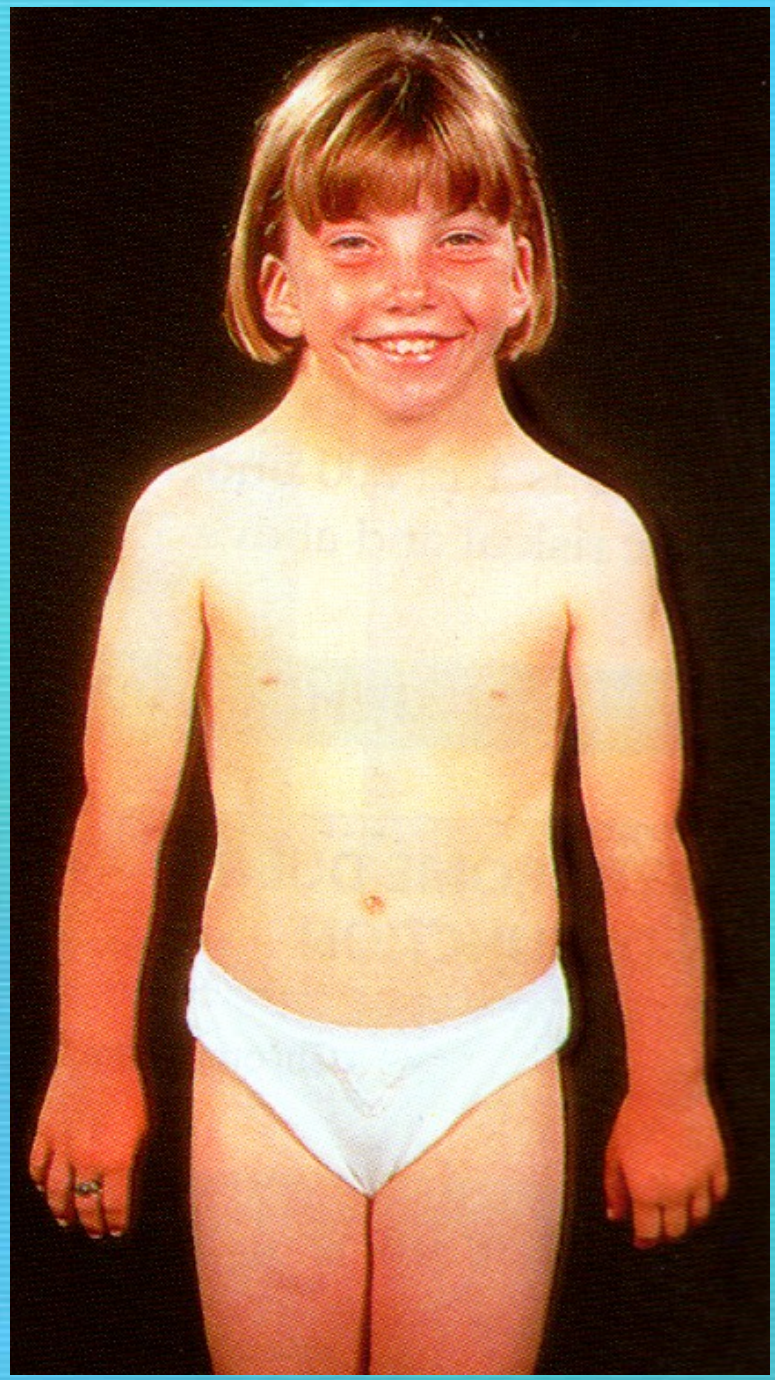
Chase Farm Hospital

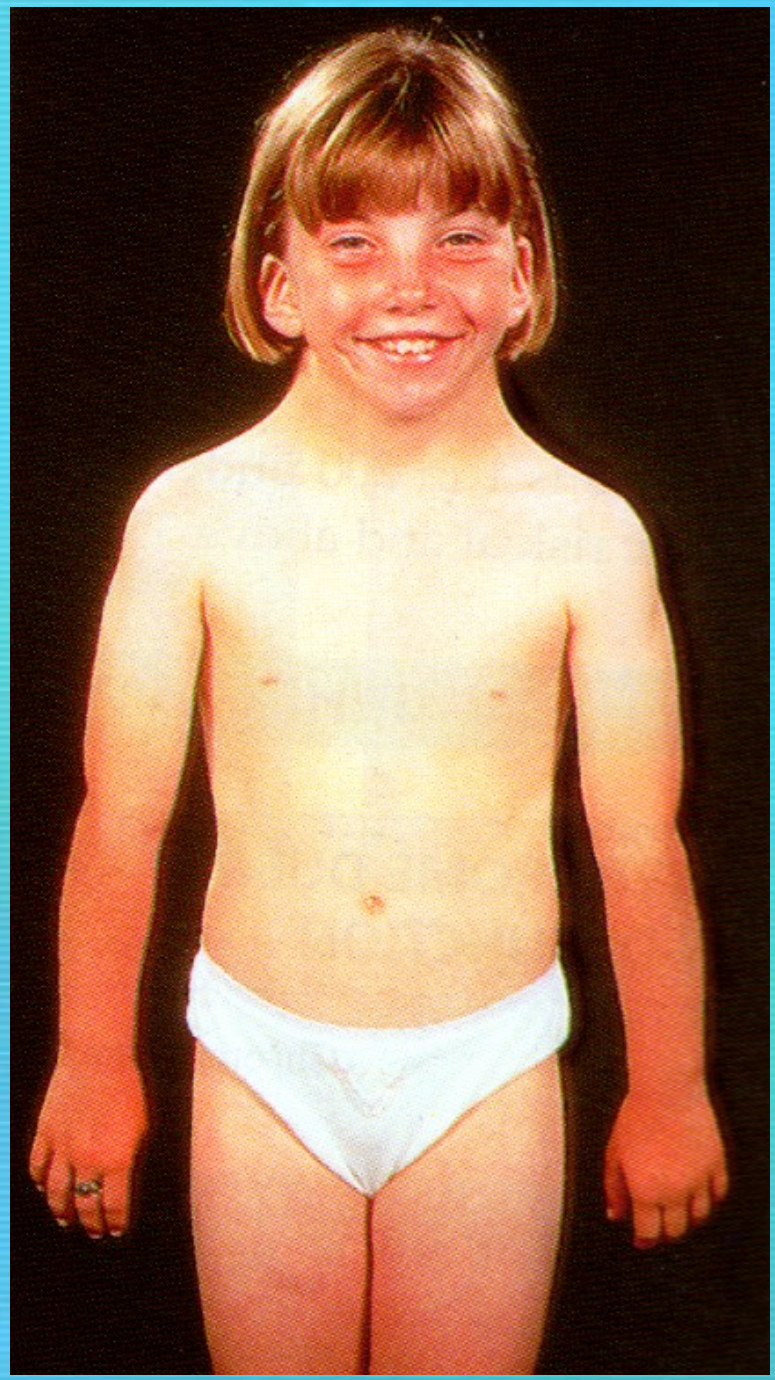
1st Dec 2009

UCL

Barnet and Chase Farm Hospitals NHS

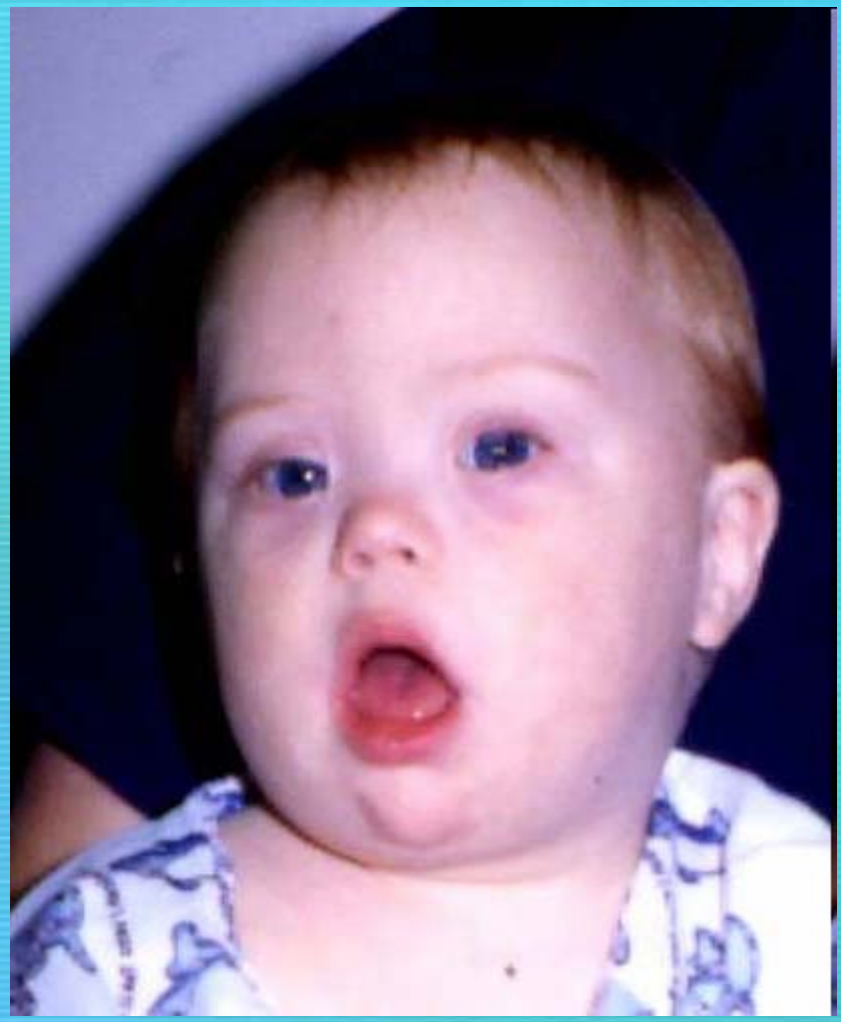
NHS Trust

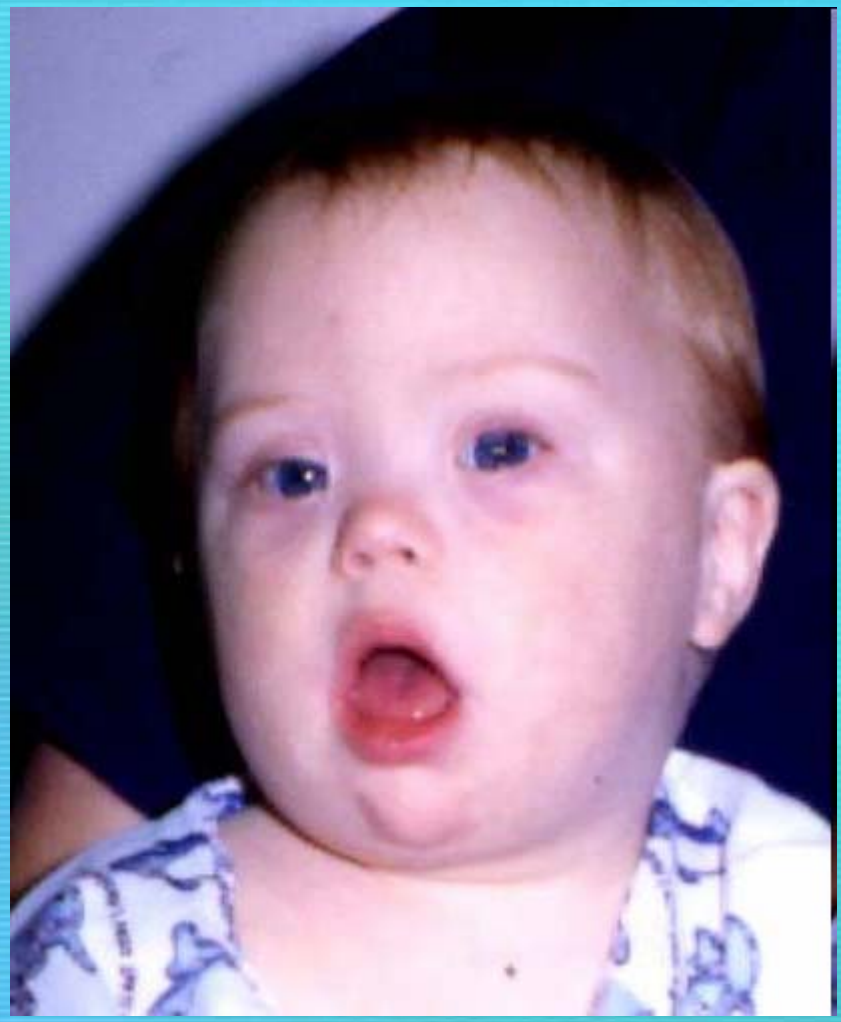




Turner’s Syndrome

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

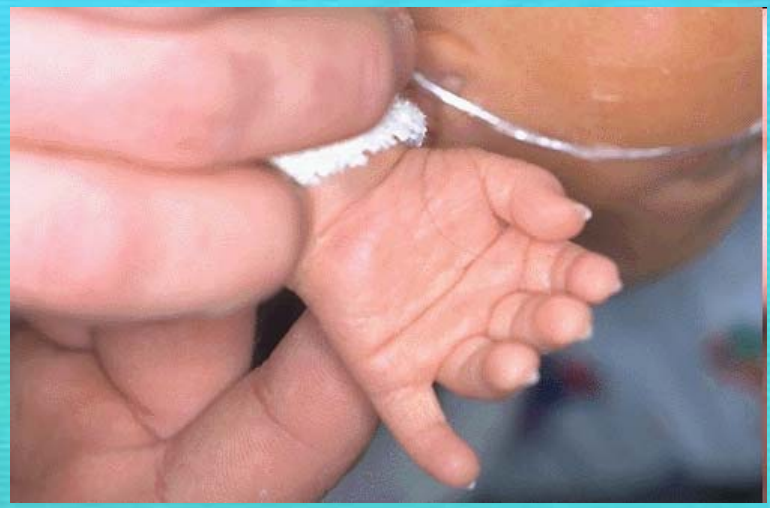
- Flat nasal bridge
- Epicanthic folds

On examination

- Flat nasal bridge
- Epicanthic folds
- Low set ears

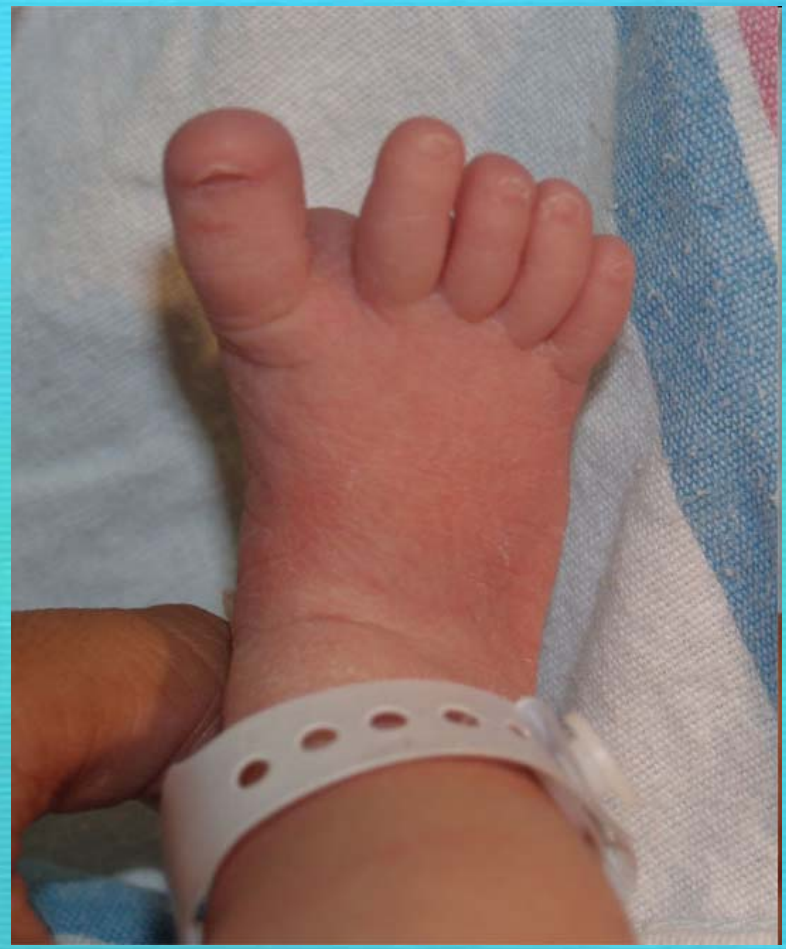
On examination

- 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

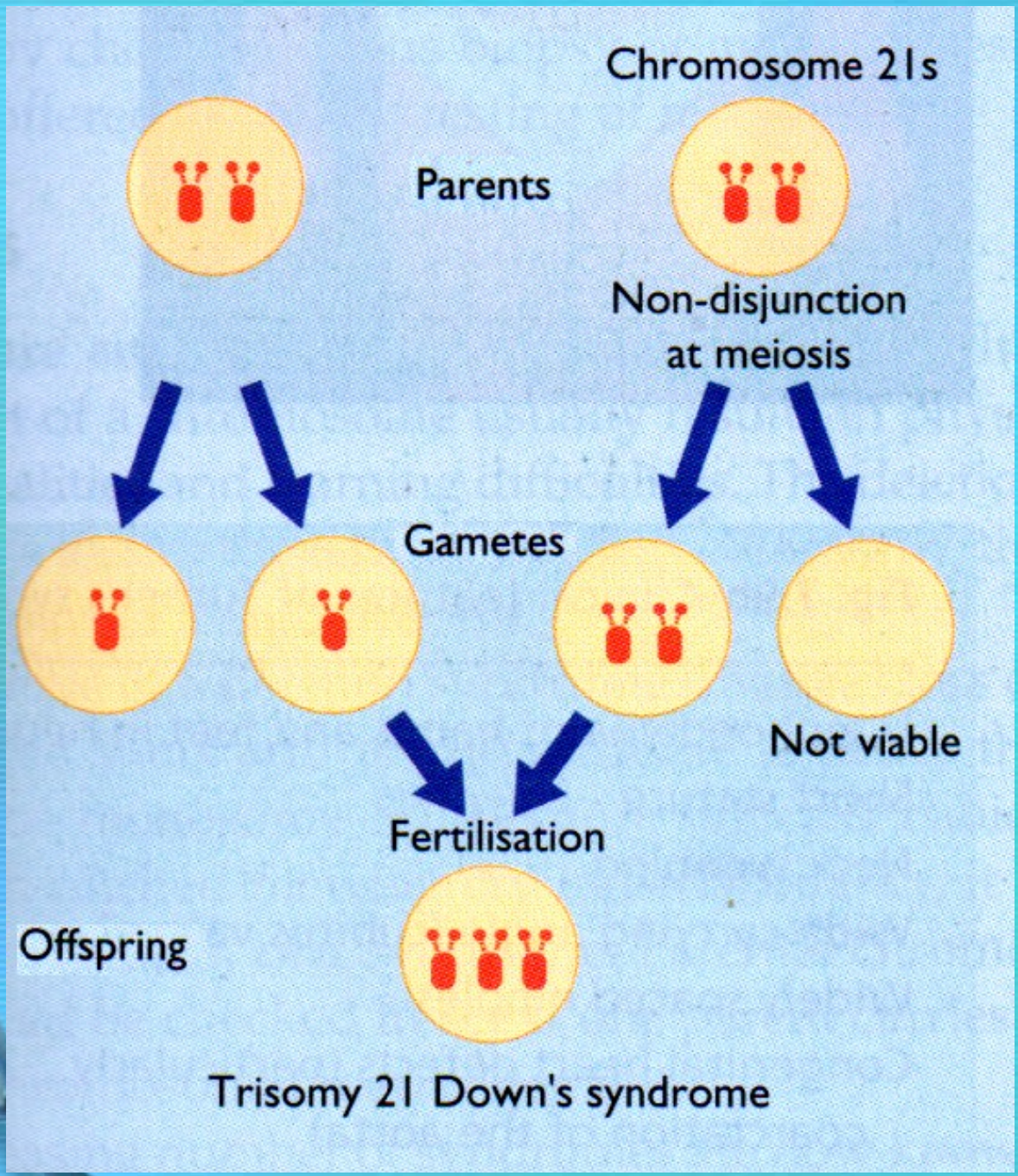
Less obvious features

- Eyes
  - Brushfield spots
  - Refractive errors
- Sparse hair
- Atlantoaxial instability
- Congenital Heart Defects: PDA, VSD
  - Usually the cause of mortality

Aetiology

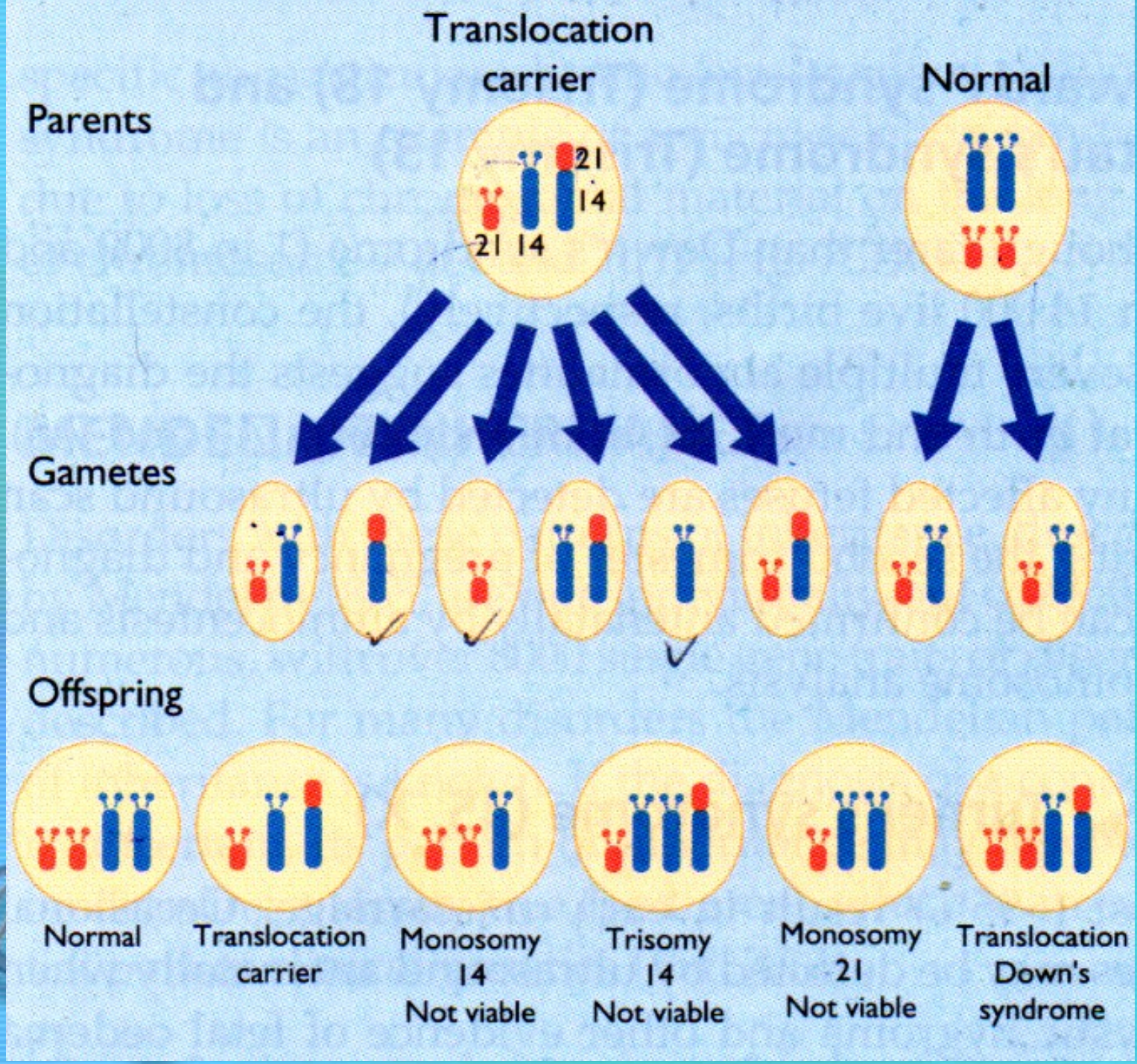
- Trisomy 21

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


Chromosome 21s
Parents
Non-disjunction
at meiosis
Gametes
Not viable
Fertilisation
Offspring
Trisomy 21 Down's syndrome

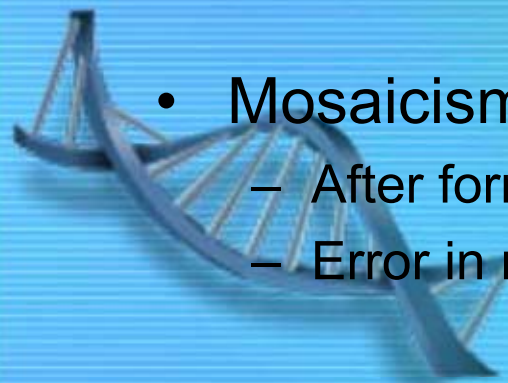
Aetiology

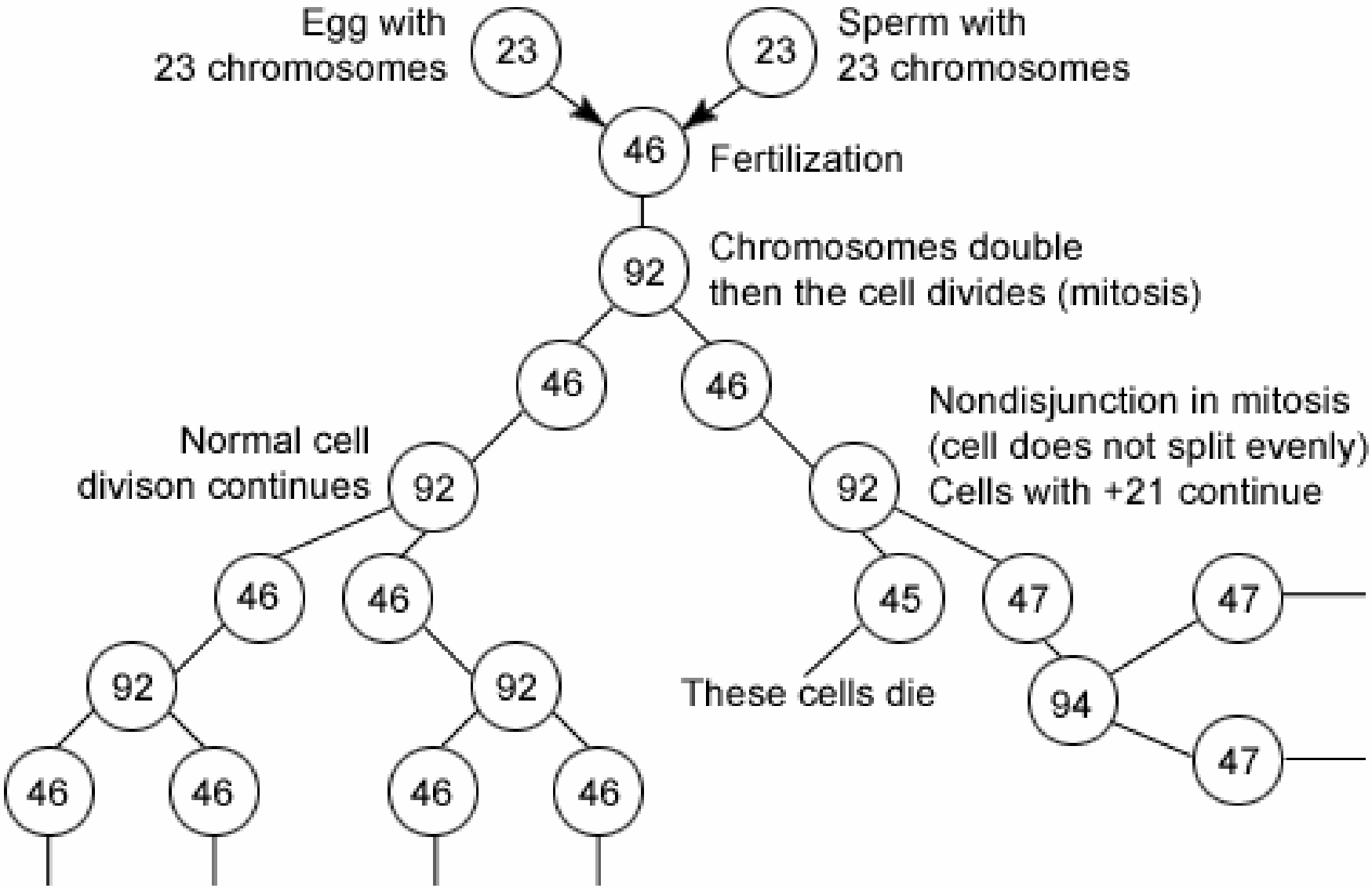
- Trisomy 21
- Non-disjunction (94%)
  - Error in meiosis
- Translocation (5%)
  - “Robertsonian”
  - Are the parents a translocation carrier?


Translocation carrier
Normal
Parents
21
14
21 14
Gametes
Offspring
Normal
Translocation carrier
Monosomy 14 Not viable
Trisomy 14 Not viable
Monosomy 21 Not viable
Translocation Down's syndrome

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



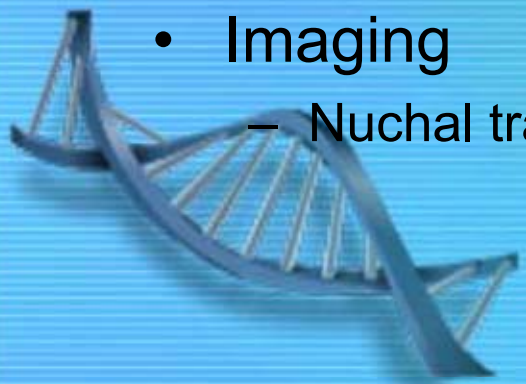

Egg with
23 chromosomes
23
Sperm with
23 chromosomes
46
Fertilization
92
Chromosomes double
then the cell divides (mitosis)
46
46
Normal cell
divison continues
92
92
Nondisjunction in mitosis
(cell does not split evenly)
Cells with +21 continue
46
46
45
47
47
92
92
These cells die
94
46
46
46
46
47

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 (reccurent infections)
  - Spinal cord
- **Multi-Disciplinary Approach**

Thank you