DOWN SYNDROME
Diagnosis Management Prevention

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DOWN SYNDROME
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HISTORY

- Till 1866 : Many recognized similar characteristics of the syndrome in many patients
- In 1866 : JOHN LANGDON DOWN, an English physician "Father" of the syndrome, described the condition as a distinct and separate entity
- In 1959 : the French physician JEROME LEJEUNE identified it as a chromosomal condition
- In 2000 : an international team of scientists identified approximately 329 genes on chromosome 21

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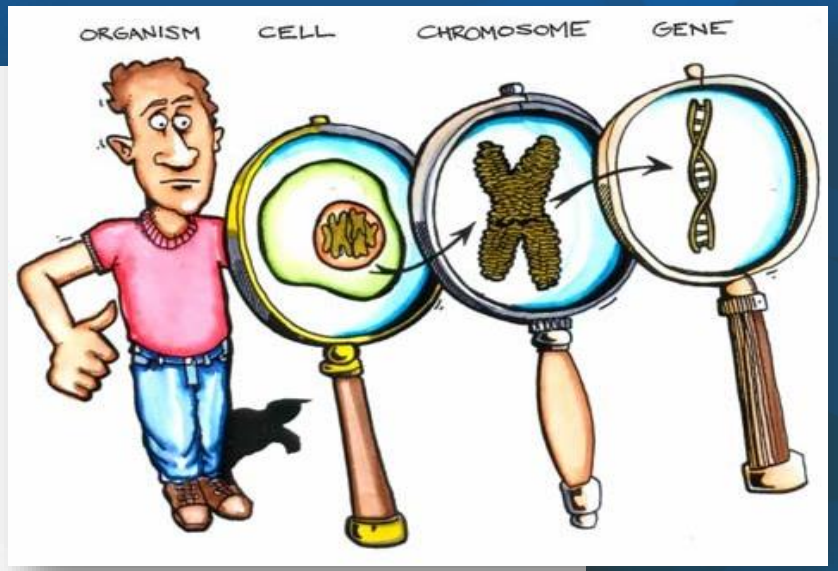
THE ‘BODY BREAKDOWN’



The **body** is made of **organs**
like skin, brain

organs are made of **tissues**
like cardiac, white brain matter

tissues are made of **specialized cells**
like nerves, pancreatic cells

cells contain a **nucleus, mitochondria and vacuoles**
the nucleus is the information center of the cell

nucleus contains **chromosomes**
chromosomes are strands of DNA,
which codes genetic information

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CHROMOSOMES

- Human cells contain 46 chromosomes

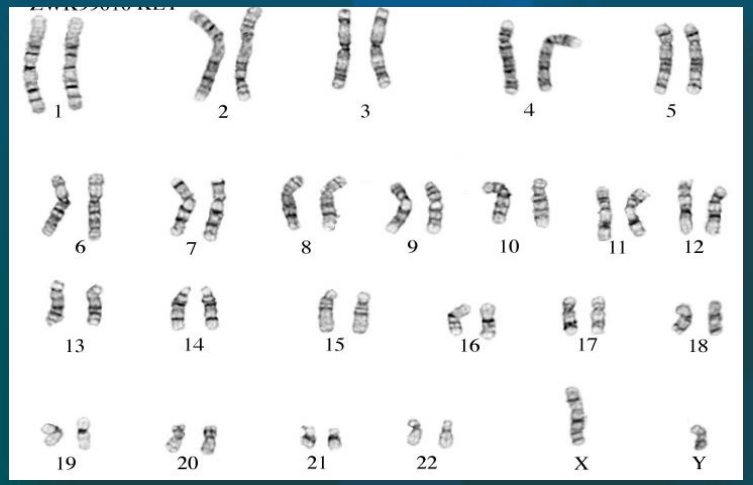
- 22 pairs of chromosomes - autosomes
- 2 sex chromosomes (X,Y)

XY – in males
XX – in females

Two Types of Chromosomal abnormalities

Chromosome Structure

Chromosome Number



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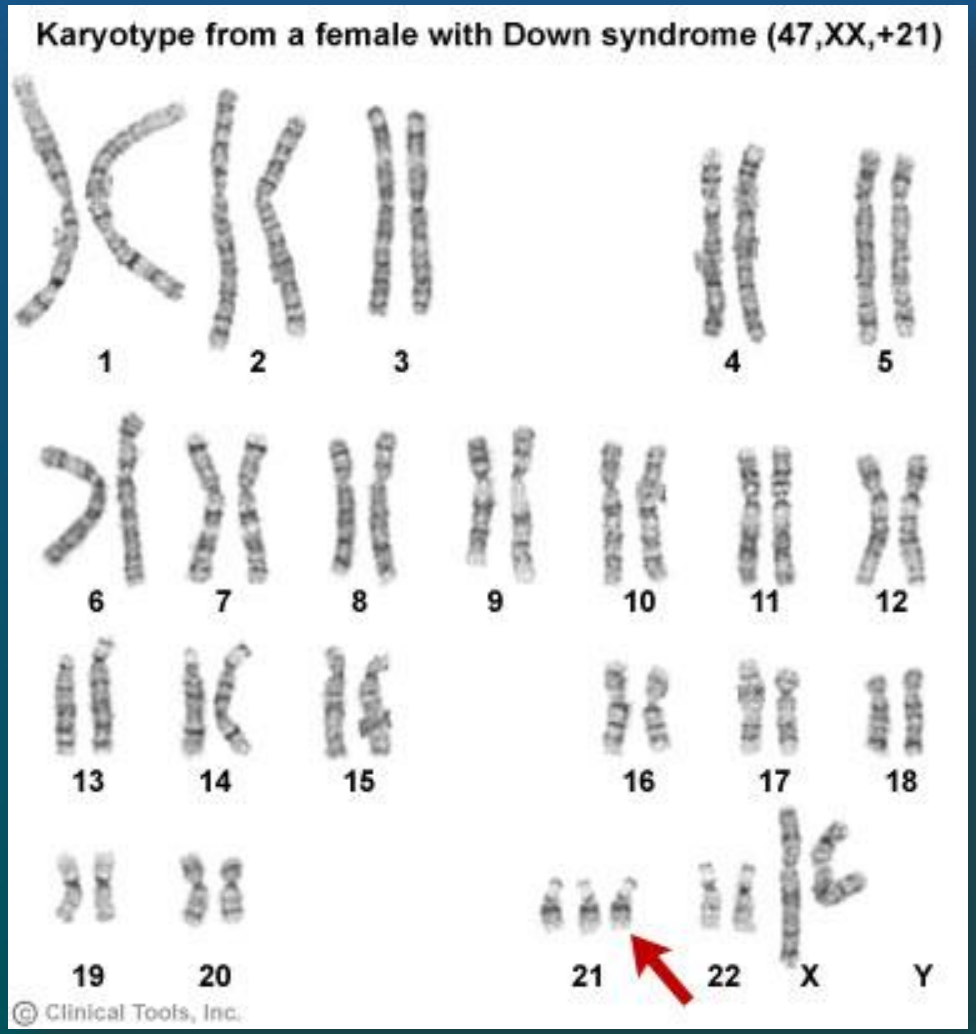
CHROMOSOMAL – ANEUPLOIDIES NUMERICAL



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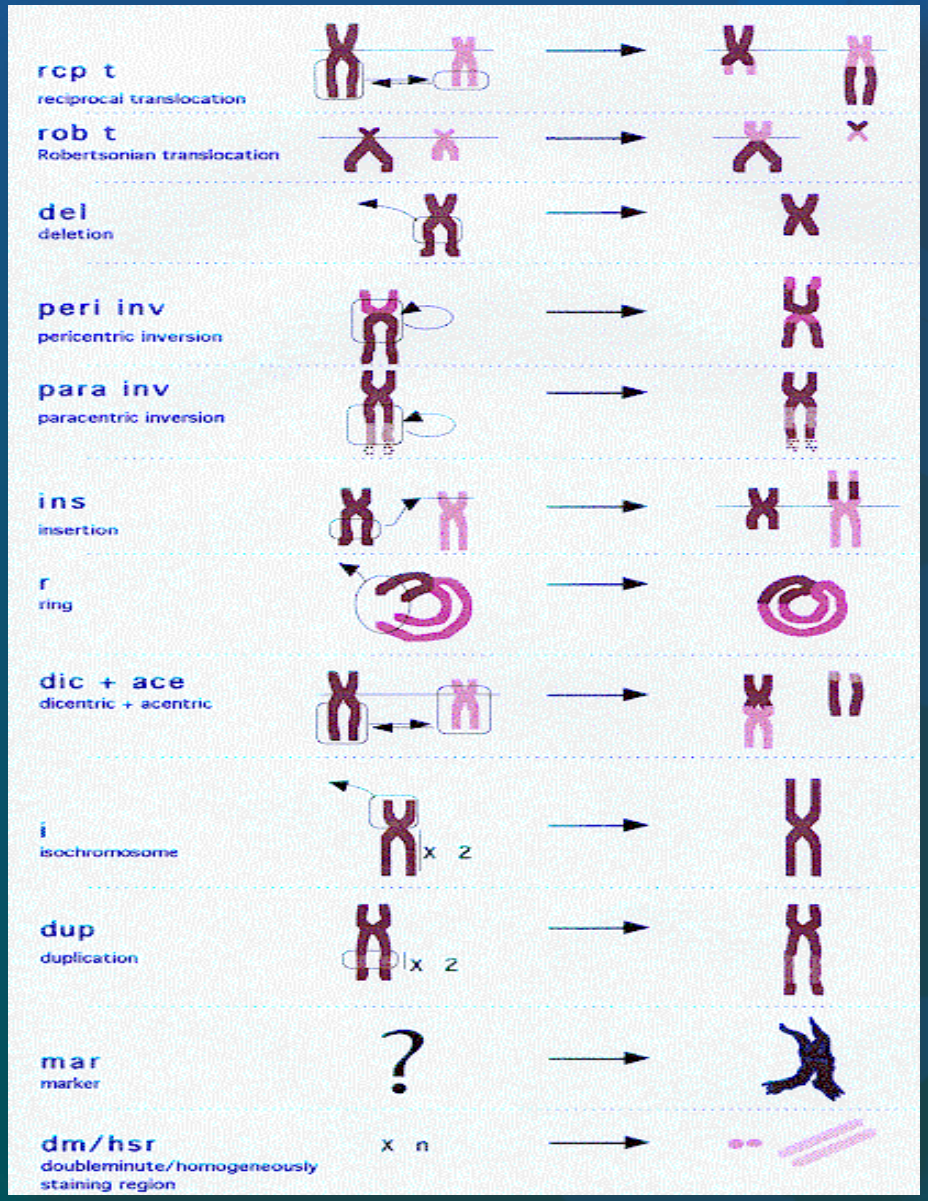
STRUCTURAL REARRANGEMENTS

- Deletion
- Duplication
- Inversion
- Translocation
- Addition
- Insertion
- Ring chromosome
- Isochromosome



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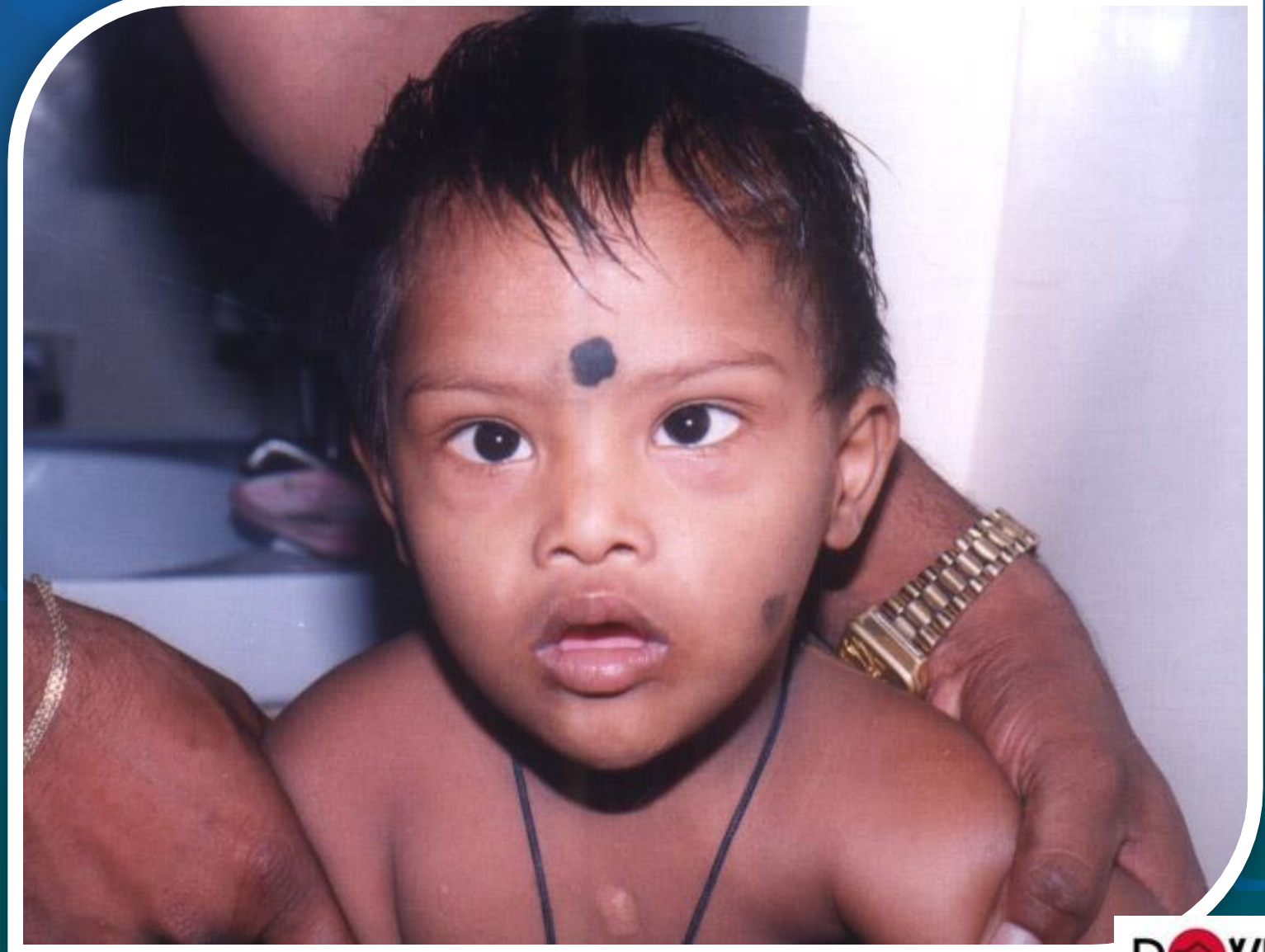

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What will be the genotype ?

NON-DYSJUNCTION OR TRANSLOCATION OR MOSAICISM ???

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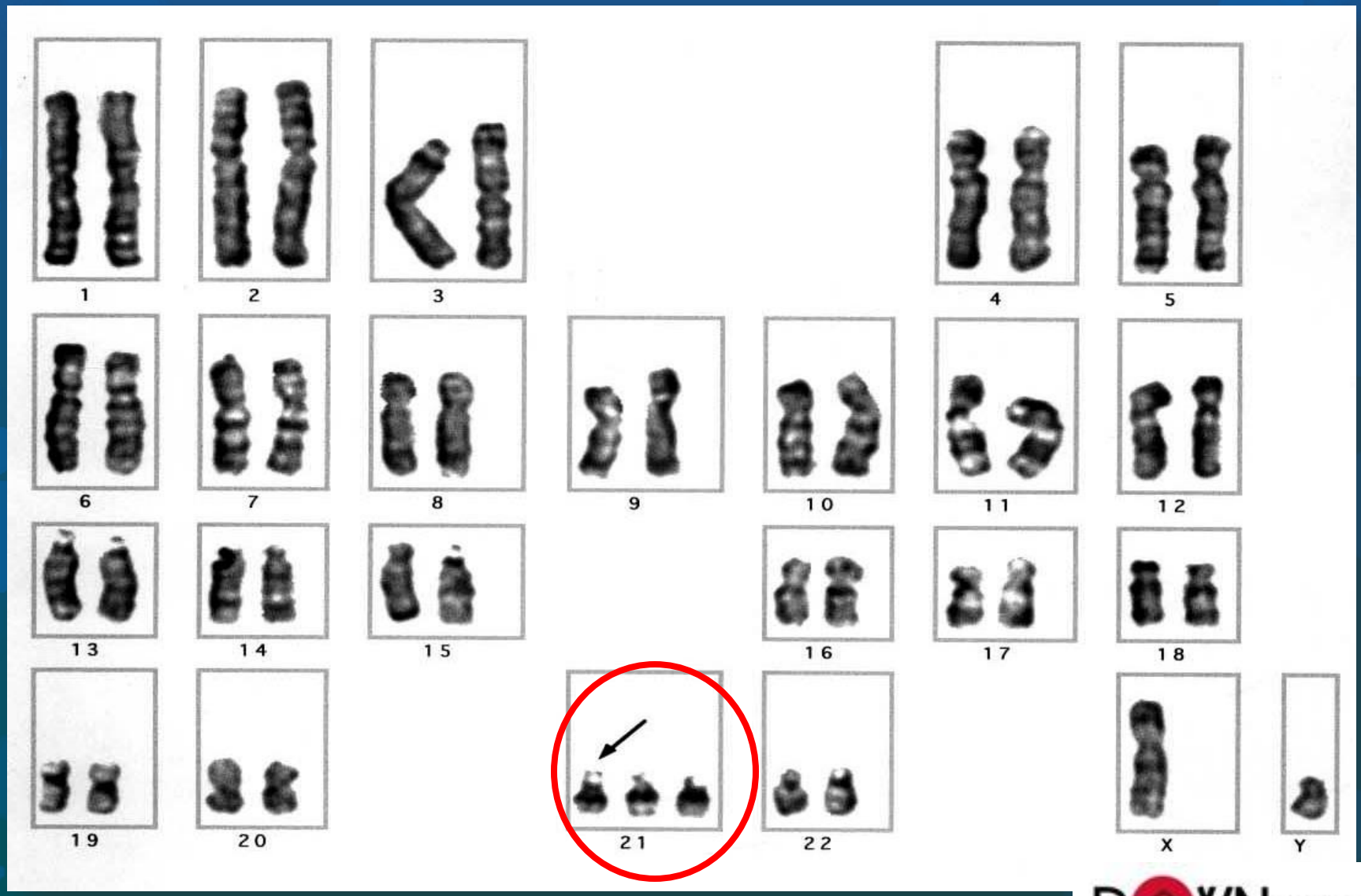
NON –DYSJUNCTION (> 95%)



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Nondisjunction

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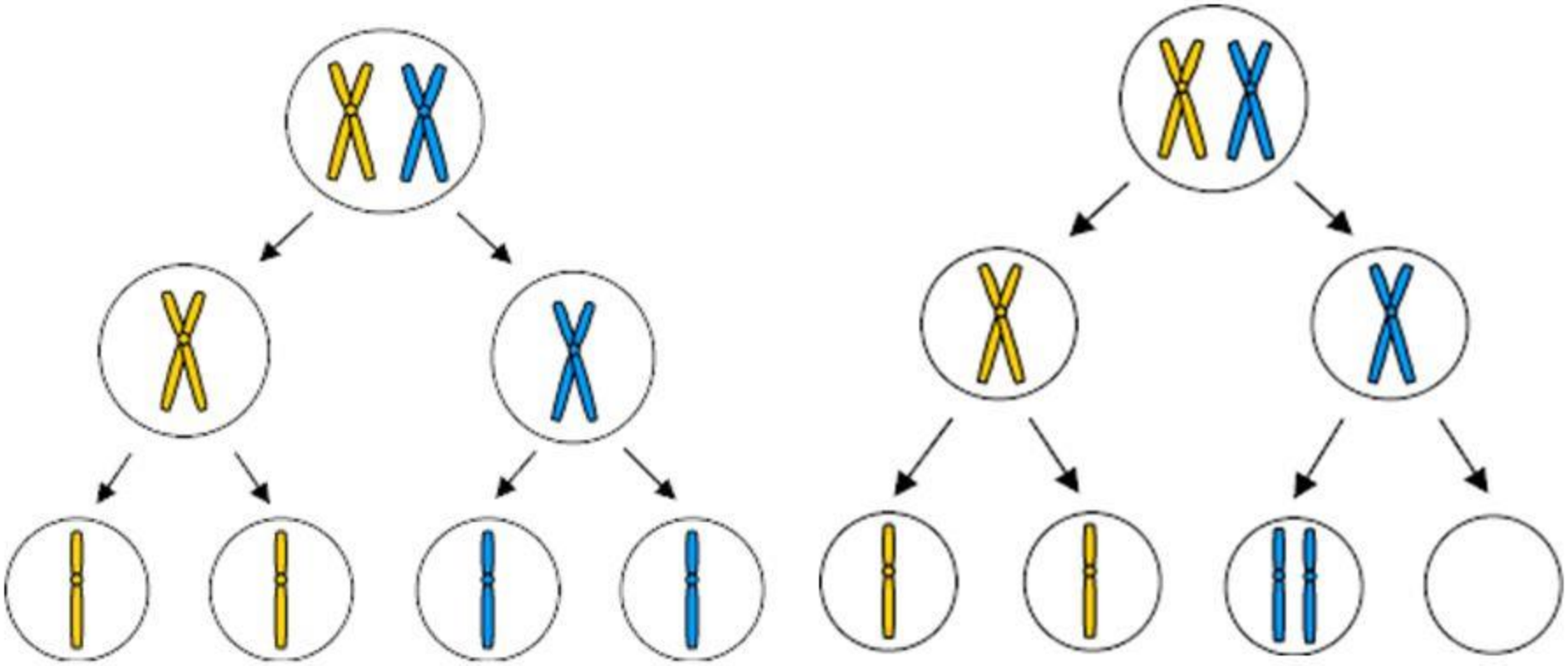
- The failure of homologous chromosomes to separate properly during meiosis.

What should happen

Nondisjunction



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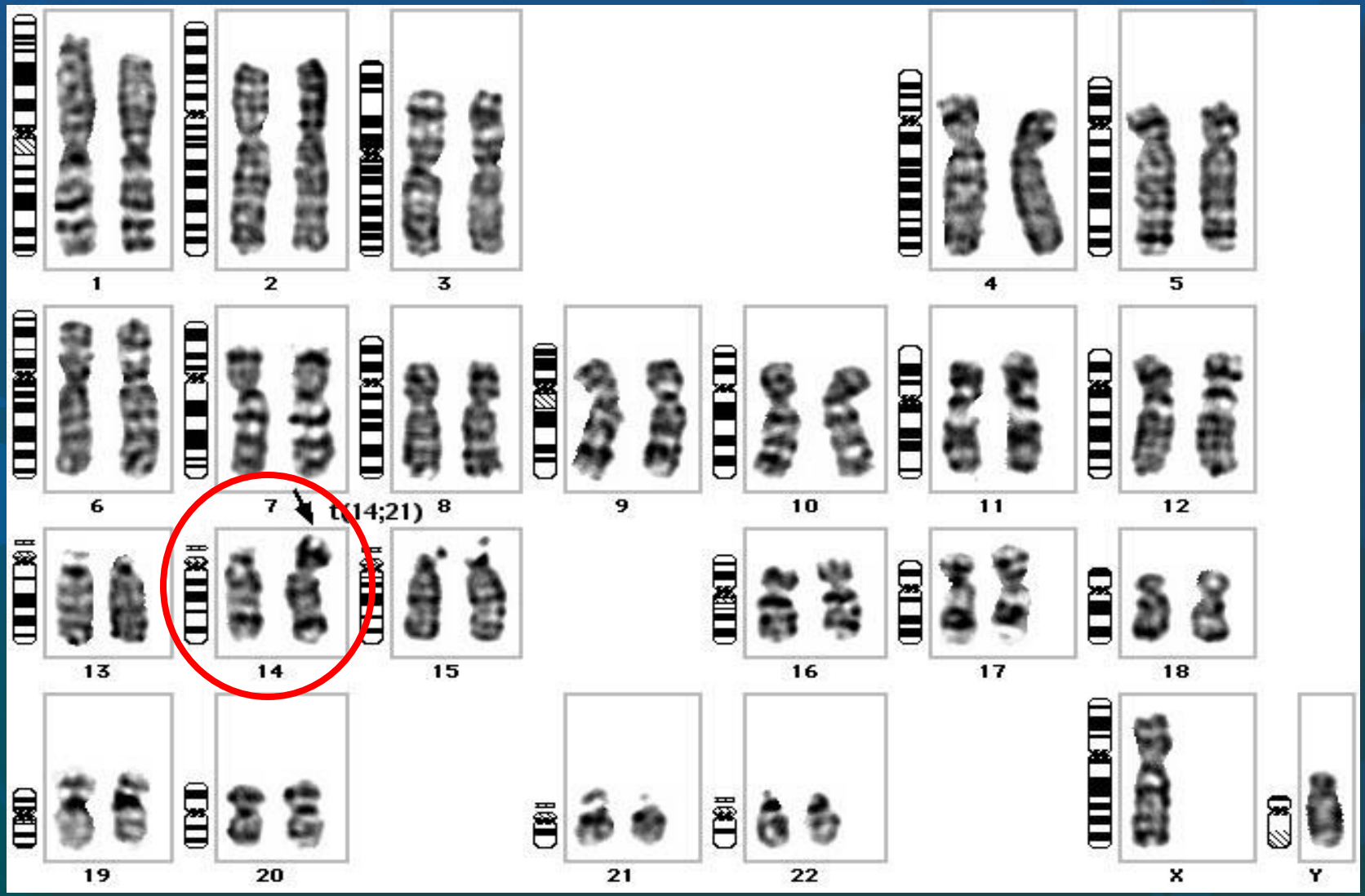
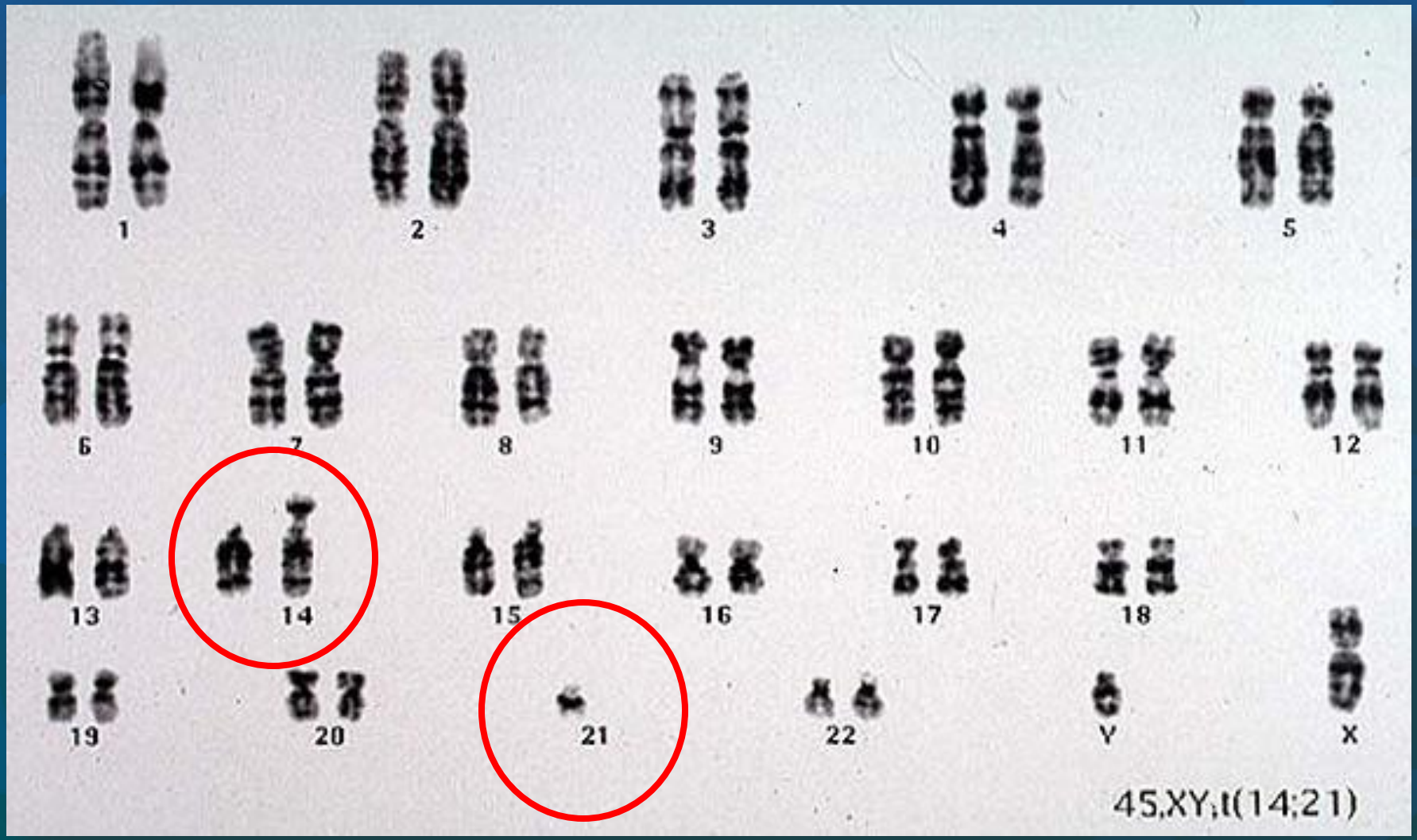
TRANSLOCATION (3-4%)





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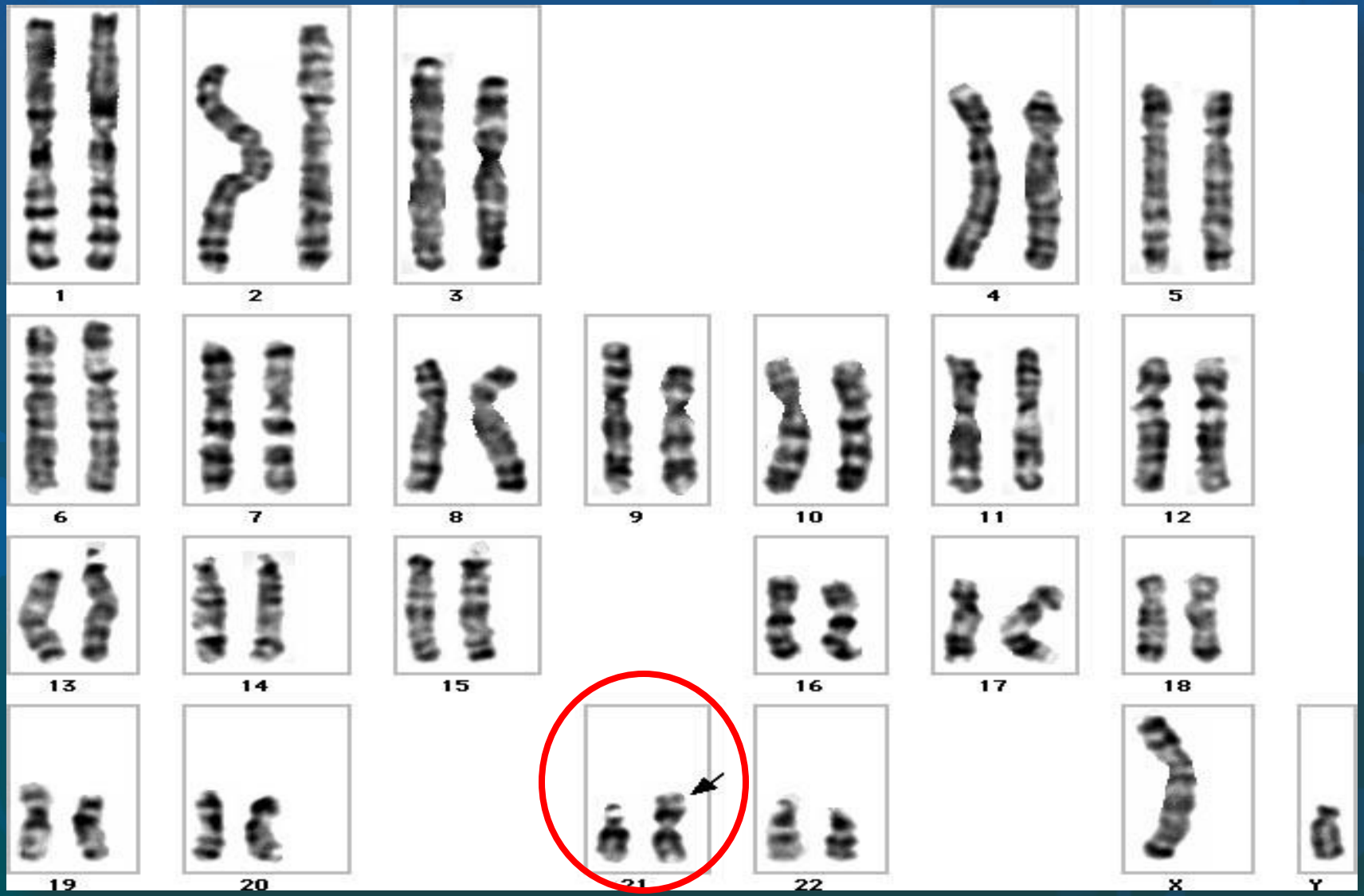

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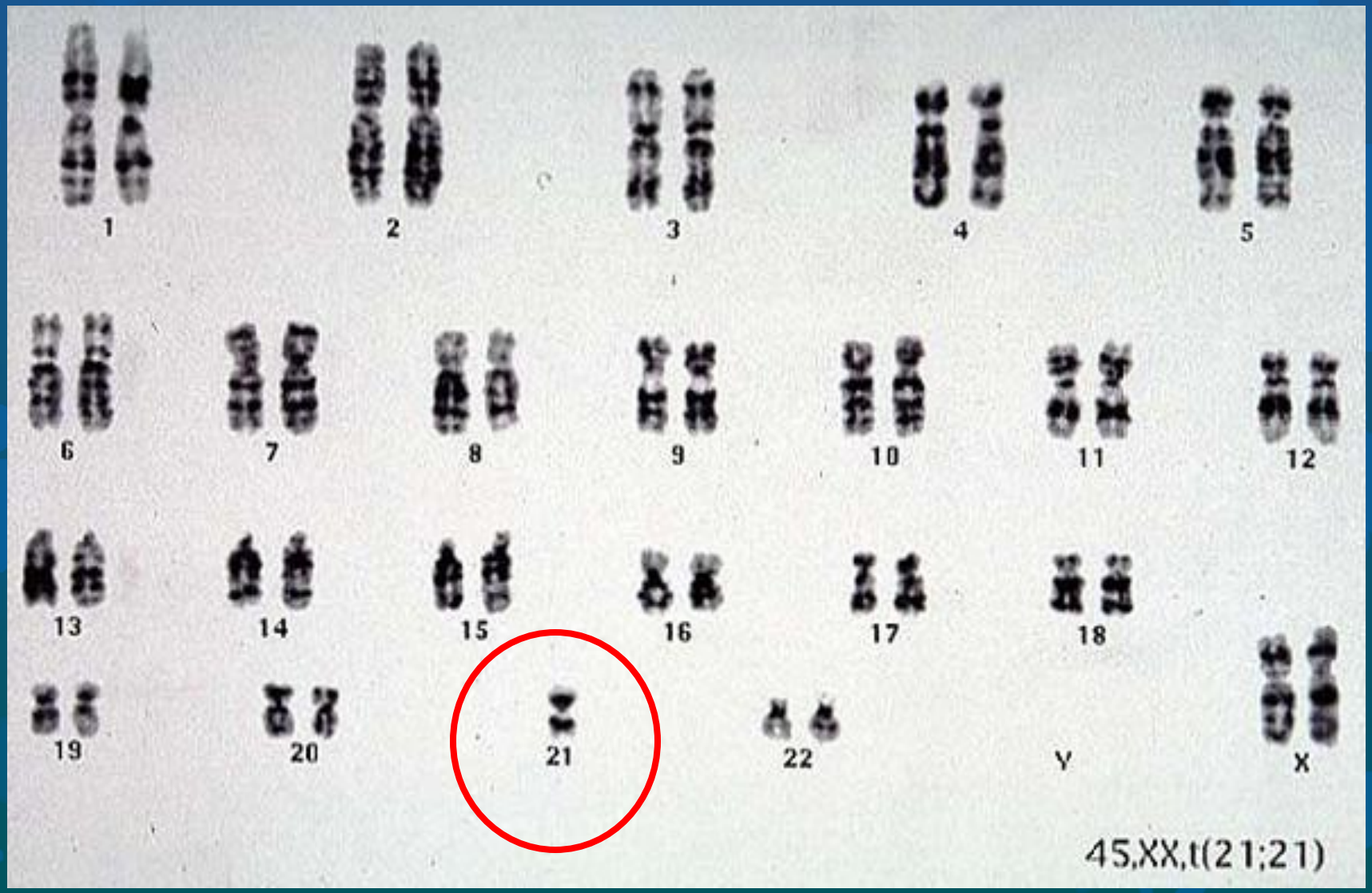
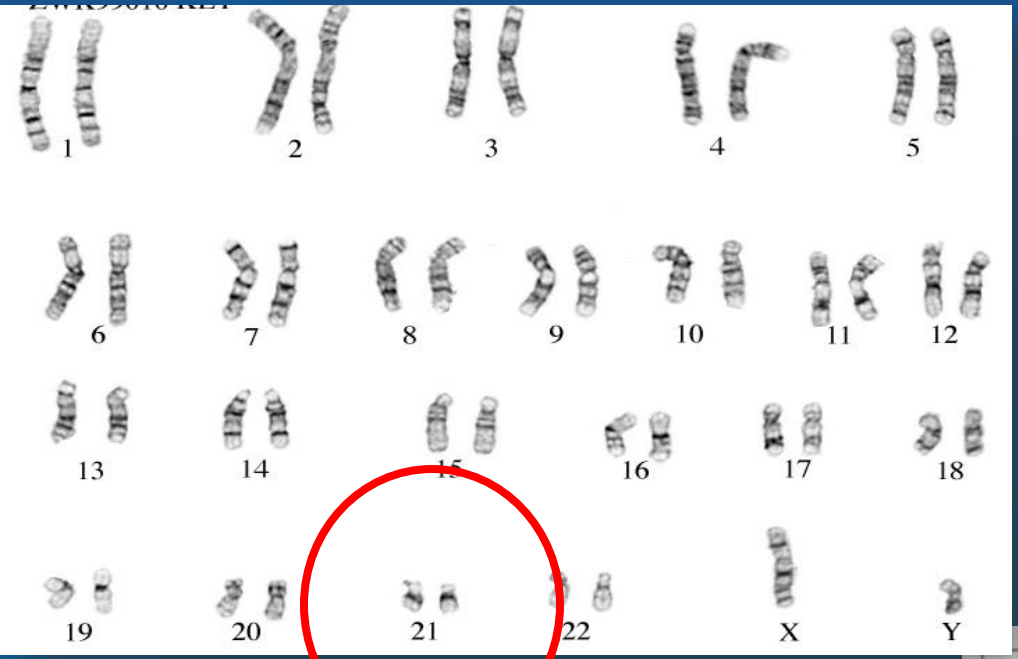


MOSAICISM (1%)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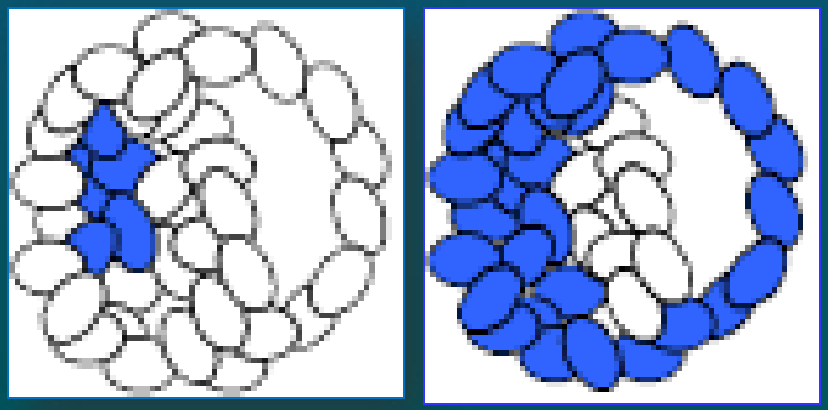
MOSAICISM

- The percentage of mosaicism is not an accurate predictor of outcome
- The percentage of Trisomic cells in the muscle may differ from the in the brain, or the blood or skin
- Great variability in mosaic Down syndrome cases, from very mild features, to severe features



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RECURRENCE RISK

- After 1st child with trisomy21 <1%
- After 2 children with trisomy 21 10%
- If father is a carrier of 14 /21 Translocation <2%
- If a mother is a carrier 14/21 Translocation 10-15%
- If a parent is carrier of 21/21 Translocation 100%
- A mother with T21 become pregnant 50%
- Males with downs usually are sterile

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CHILD WITH DOWN SYNDROME

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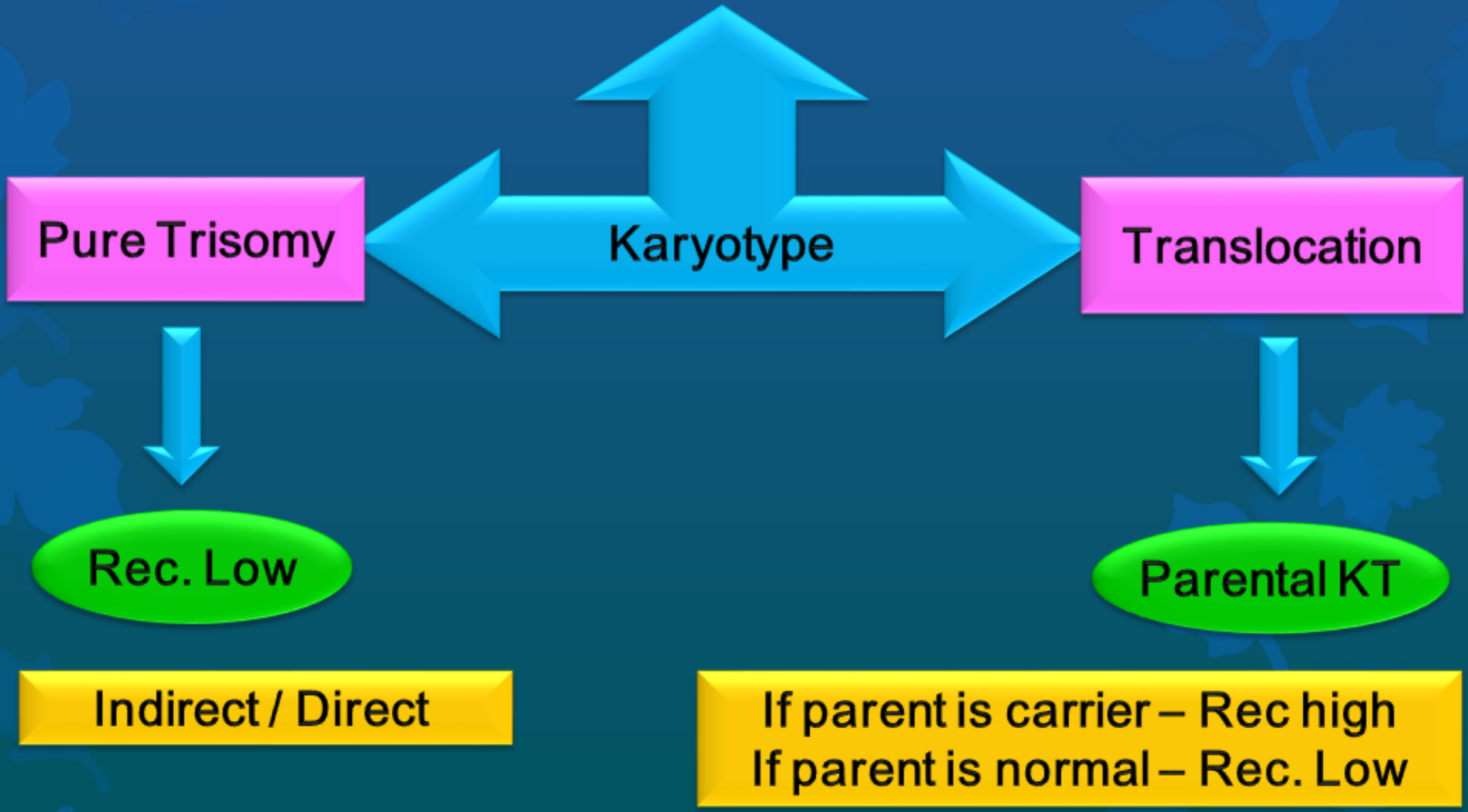

POST NATAL COUNSELING

- When and How to reveal ???
- What to say about the brain growth ???
- How fruitful the offspring is going to be for the family ?
- How am I going to take care of the baby ?
- How am I going to counsel…Optimistic / Pessimistic ???

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CENTRAL NERVOUS SYSTEM

- Global Developmental delay - Early intervention !!!
  - Physio, Speech, Occupational & Special education
- Seizures <9%
- Autism spectrum disorders
- Behavioral disorders
- Depression
- Alzheimer disease

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CARDIO- PULMONARY

- Endocardial Cushing defects 40%, VSD, ASD, PDA, Pulmonary hypertension, Acquired mitral, Tricuspid, or Aortic valve regurgitation
- Obstructed sleep apnea
- Frequent infections
  - Sinusitis
  - Nasopharyngitis
  - Pneumonia

ENDOCRINE

- Short stature
- Hypothyroidism / Hyperthyroidism
- Diabetes mellitus
- Infertility
- Obesity
- Primary gonadal deficiency
- Vitamin D Defficiency

MUSCULOSKELETAL

- Atlantoaxial instability 12%
- Hip dysplasia
- Slipped capital femoral epiphyses
- Avascular hip necrosis
- Scoliosis
- Recurrent joint dislocations
  - Shoulder, knee, elbow, thumb

GASTROINTESTINAL 12%

- Duodenal atresia, Annular pancreas, Tracheoesophageal fistula, Hirschsprung disease, Imperforate anus, Neonatal cholestasis, Diastases of recti
- Celiac disease
- Delayed tooth eruption, periodontal disease

HEARING AND VISION

- Congenital or acquired hearing loss
- Serous otitis media
- Refractive errors (Myopia)
- Congenital or acquired cataracts
- Nystagmus
- Strabismus
- Glaucoma
- Blocked tear ducts

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HEMATOLOGIC

- Transient myeloproliferative syndrome
- Acute lymphocytic leukemia
- Acute myelogenous leukemia

CUTANEOUS

- Hyperkeratosis
- Seborrhea
- Xerosis
- Perigenital folliculitis

- **INFECTIONS AND VACCINATIONS**

Age, in years															
Medical Issues	At Birth or at Diagnosis	6-mo	1	1-1/2	2	2-1/2	3	4	5	6	7	8	9	10	11	12
Karotype & Genetic Counseling	_____															
Usual Preventative Care	_____	____	____	____	____	____	____	____	____	____	____	____	____	____	____	____
Cardiology	Echo															
Audiologic Evaluation	ABR or OAE	____	____	____	____	____	____	____	____	____	____	____	____	____	____	____
Ophthalmologic Evaluation	Red reflex	_____	____		____		____	____	____	____	____	____	____	____	____	____
Thyroid (TSH & T_4)	State screening	_____	____		____		____	____	____	____	____	____	____	____	____	____
Nutrition	_____	____	____	____	____	____	____	____	____	____	____	____	____	____	____	____
Dental Exam[1]					____	____	____	____	____	____	____	____	____	____	____	____
Celiac Screening[2]					____											
Parent Support	_____	____	____	____	____	____	____	____	____	____	____	____	____	____	____	____
Developmental & Educational Services	Early Intervention	____	____	____	____	____	____	____	____	____	____	____	____	____	____	____
Neck X-rays & Neurological Exam[3]							X-ray	____	____	____	____	____	____			

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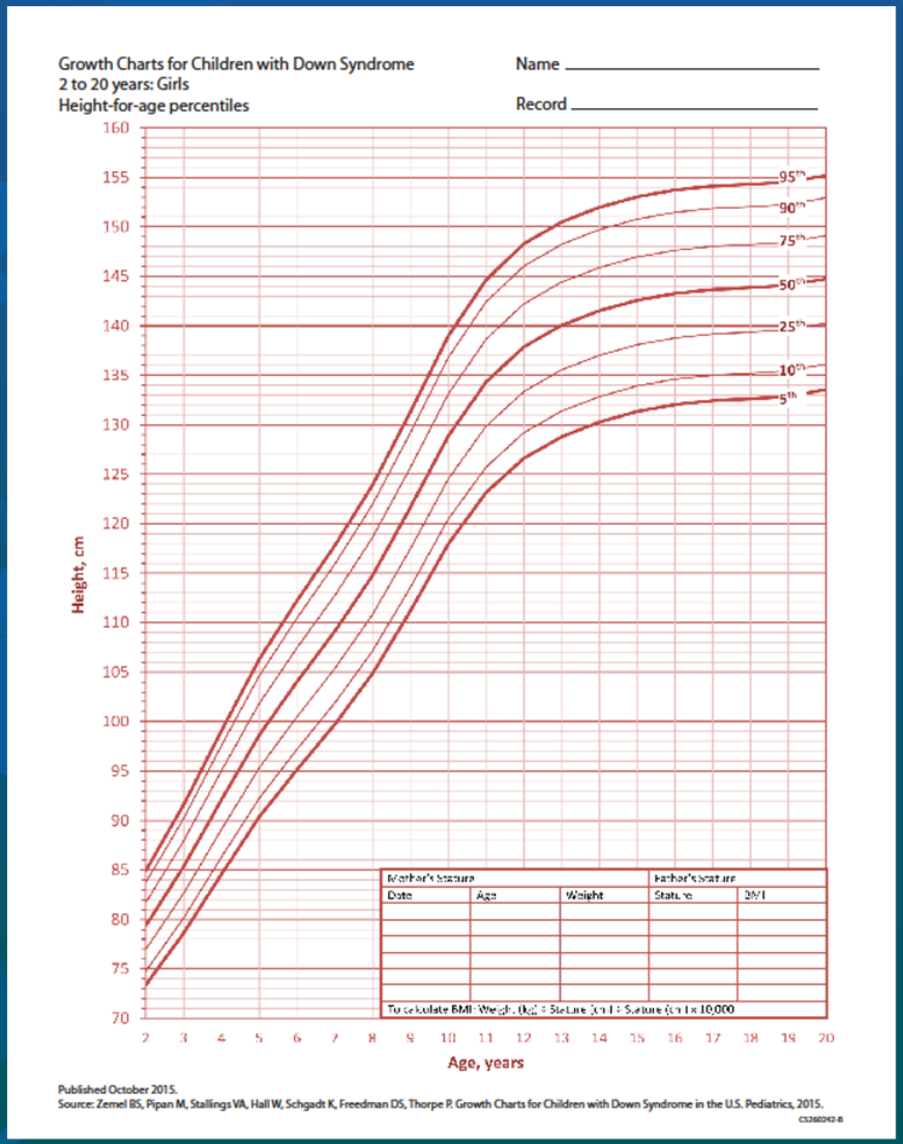
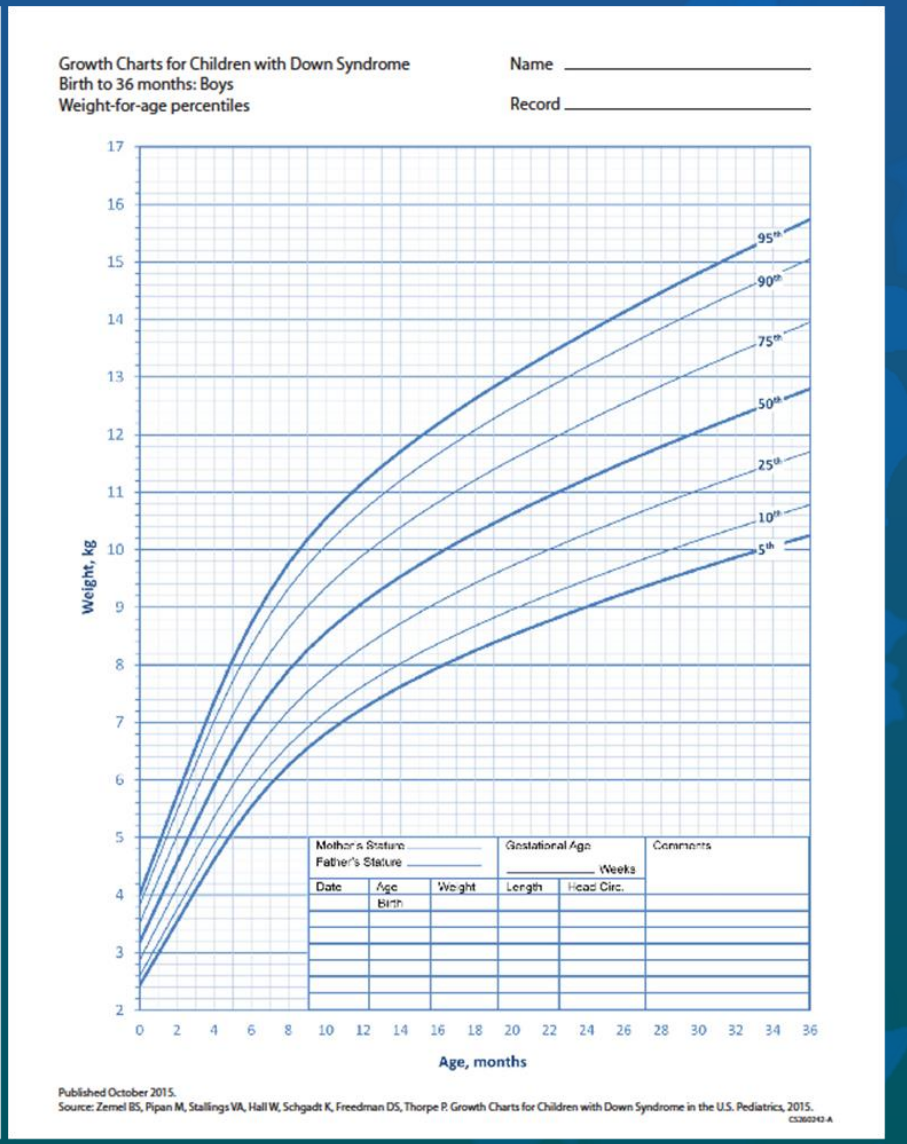
GROWTH CHARTS





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INTERESTING FACTS

- Emotional problems – 13 %
- Atlantoaxial instability 12-20% - asymptomatic
- Life expectancy – 55-60 years
- 25% lives beyond 62 years
- By 60 years of life, 50-70% have Dementia
- Early Mortality due to CVS, Lower respiratory infections

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STRENGTHS OF DOWN SYNDROME CHILDREN

- Friendly
- Music lovers
- Swim fast
- Dance gracefully
- Wont betray
- Shows unconditional love
- Real “God’s children !!!”

THANK YOU !