DOWN SYNDROME
Diagnosis Management Prevention

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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
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, which are strands of DNA that code genetic information.
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
CHROMOSOMAL – ANEUPLOIDIES
NUMERICAL

Karyotype from a female with Down syndrome (47,XX,+21)
STRUCTURAL REARRANGEMENTS

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

DOWN SYNDROME
FEDERATION OF INDIA
What will be the genotype?

NON-DYSJUNCTION OR TRANSLOCATION OR MOSAICISM ??
NON-DYSJUNCTION (> 95%)
Nondisjunction

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

What should happen

Nondisjunction
TRANSLOCATION (3-4%)
MOSAICISM (1%)
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.
RECURRENCE RISK

- After 1st child with trisomy 21: <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
CHILD WITH DOWN SYNDROME

Pure Trisomy

Karyotype

Translocation

Rec. Low

Indirect / Direct

If parent is carrier – Rec high
If parent is normal – Rec. Low

Parental KT
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 ???
CENTRAL NERVOUS SYSTEM

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

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

CUTANEOUS

- Hyperkeratosis
- Seborrhea
- Xerosis
- Perigenital folliculitis

INFECTIONS AND VACCINATIONS
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GROWTH CHARTS

Growth Charts for Children with Down Syndrome
2 to 20 years: Girls
Height-for-age percentiles

Growth Charts for Children with Down Syndrome
Birth to 36 months: Boys
Weight-for-age percentiles
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
STRENGTHS OF DOWN SYNDROME CHILDREN

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