DOWN SYNDROME

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History

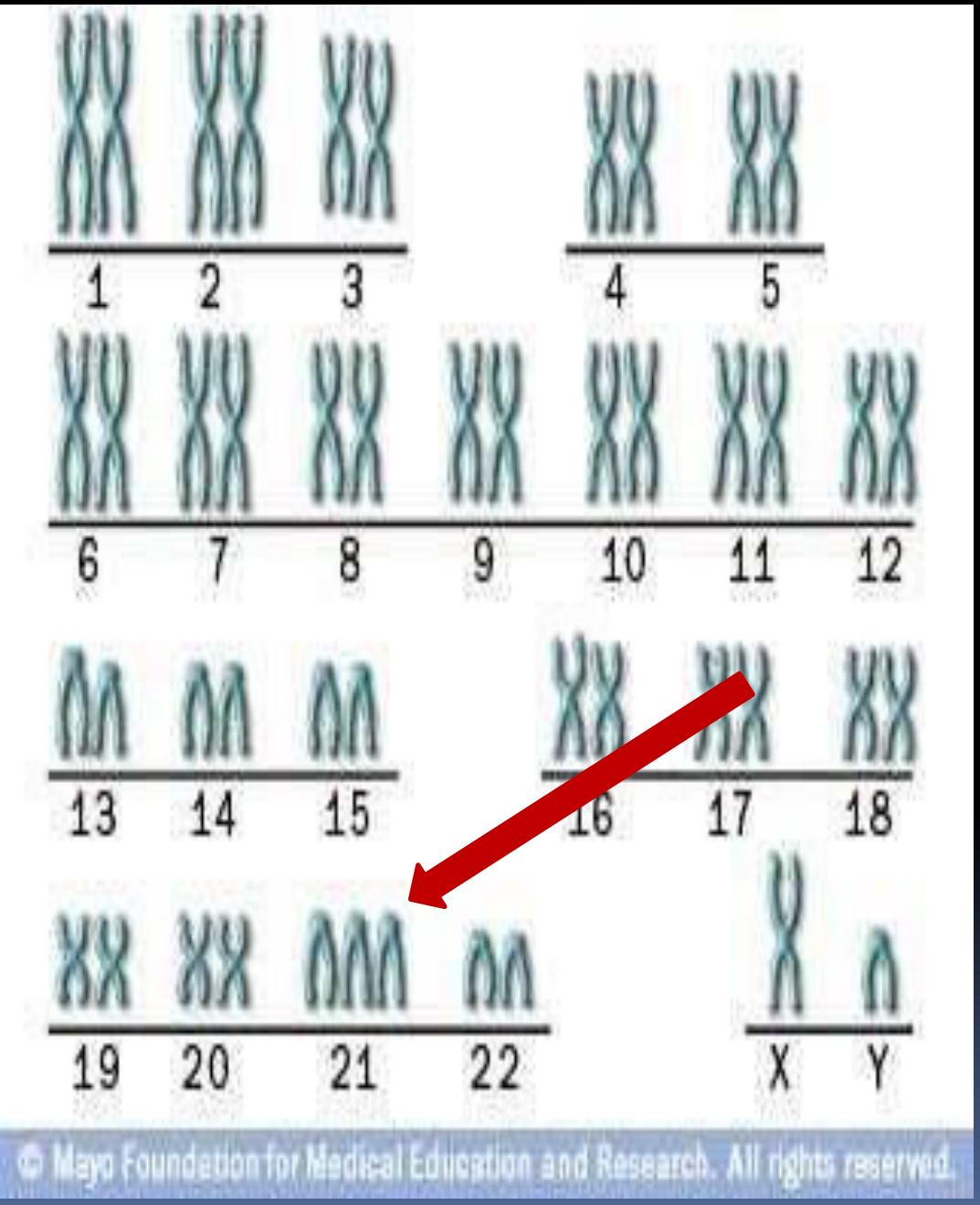
- Named after a physician, “John Langdon Down” in 18th century.
- Described as Mongoloid child of European parentage: “Mongolism”
- In 1959 a French doctor, named “Jerome Lejeune”, discovered it was caused by the inheritance of an extra chromosome 21.
- Also known as “trisomy 21”

Introduction

- Down syndrome is an autosomal disorder because it affects chromosome 21, which is an autosome.
- Down syndrome is neither a dominant nor recessive trait because it is just an error in the “translation” process of chromosome 21.

Genetics of Down Syndrome

- It is believed that the amyloid precursor protein gene (App) is the cause of Down syndrome and it is located on chromosome 21.
- A mutation in this gene usually results in Alzheimer's disease . Similarly three copies of this gene has a huge effect on the brain & other tissues of body.
- Scientists believe that excess App gene is causing the cells to die (apoptosis), because it interfere with the normal cell division (mitosis).
- Therefore people with down syndrome tend to develop the brain with signs of Alzheimer's and abnormalities other parts of the body.



92% -94% Trisomy 21
- nondisjunction during fertilisation

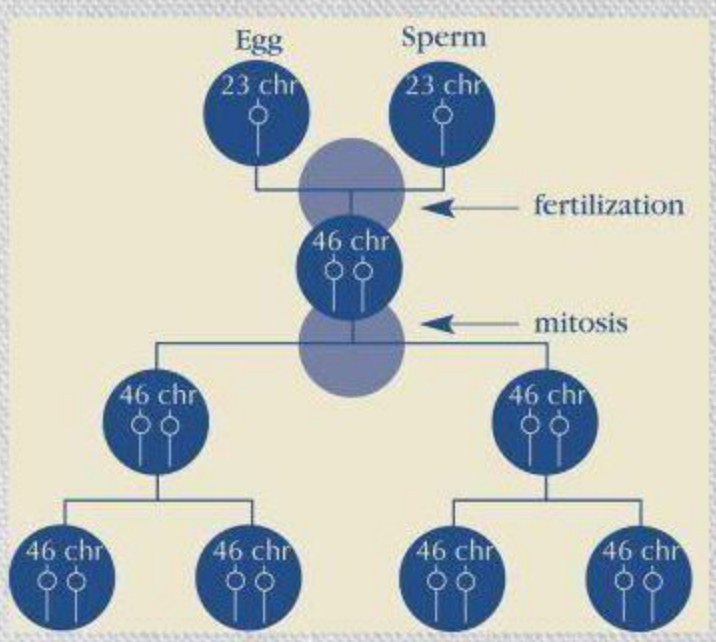
2-4% Mosaicism
- error in cell division after fertilisation

3-4% Translocation of chromosome 21
- breaking and attaching to other chromosomes (14) during cell division

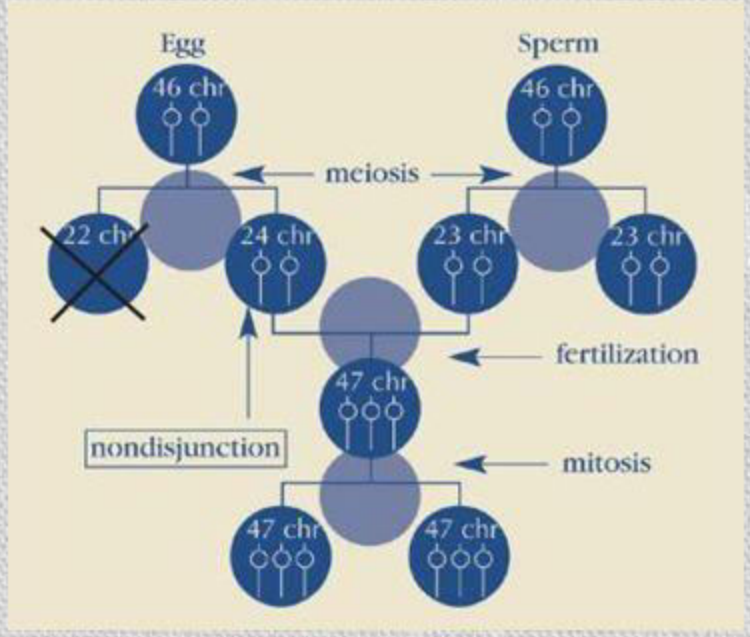
Trisomy 21 (nondisjunction)

- Down syndrome is usually caused by an error in cell division called "nondisjunction." Nondisjunction results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This type of Down syndrome, which accounts for 95% of cases, is called trisomy 21.

TYPICAL CELL DIVISION



TRISOMY 21 (NONDISJUNCTION) CELL DIVISION



Risk factors

- Advancing maternal age – usually women of age 35 and above
- Mothers who already have one child with Down syndrome – increased risk for subsequent pregnancies
- Parents who are carriers of the genetic translocation for Down syndrome

Down Syndrome & Maternal Age

A study done in Mysore, India - paternal age and maternal grandmother"s age influences Down Syndrome in neonates.

Age	Incidence of Down Syndrome
< 30	Less than 1 in 1000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12

Clinical features

Life expectancy : 55 **years** (National Down Syndrome Society)

Physical appearances

- flat facial profile and an upward slant to the eye
- short neck
- abnormally shaped ears
- white spots on the iris of the eye (called Brushfield spots)
- single, deep transverse crease on the palm of the hand.

-relatively late development of deciduous and permanent teeth as compared with other children

- Teeth could appear in a different sequence and positions
- Teeth are often are rounded, pointed or cone-shaped. Smaller with gaps
- Fewer teeth.
- Maxilla is narrow, the tongue appears too big for the mouth and the teeth may be pushed out of place, as the child grows older.

- Habit of breathing through the mouth
- Mental retardation varied from mild to moderate – some even have special abilities after training and early interventions

Neonatal features

- Flat facial profile
- Poor Moro reflex
- Excessive skin at the nape of neck
- Slanted palpebral fissures
- Hypotonia
- Hyper flexibility of joints
- Dysplasia of pelvis
- Anomalous ears
- Dysplasia of midphalanx of fifth finger
- Transverse palmer crease(simian)


flattened
nose and face,
upward slanting
eyes,
single palmer
crease, short
fifth finger that
curves inward
widely separated
first and second
toes and increased
skin creases
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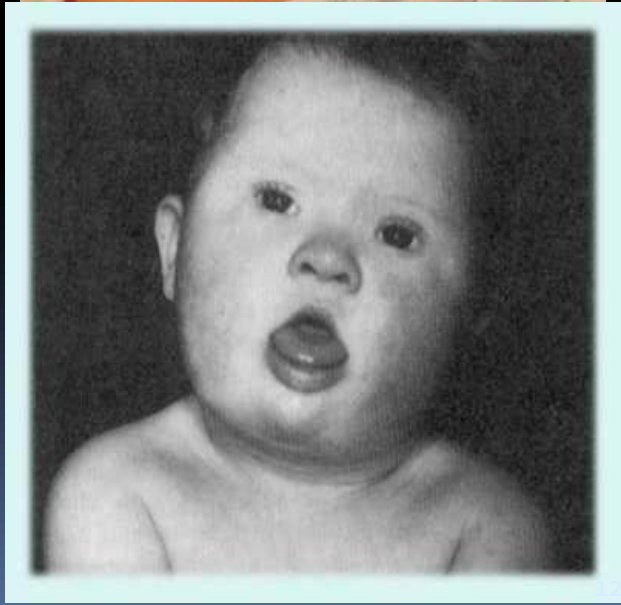


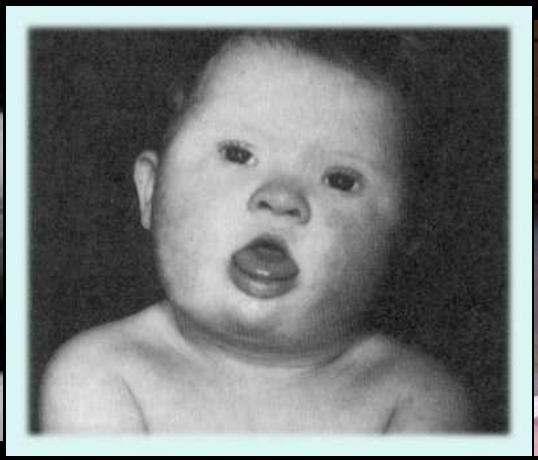
- Other Health-related problems
  - Cardiovascular problems
    - ventricular spetal defect, atrial septal defect, patent ductus arteriosus
  - Endocrine problems
    - thyroid problems, diabetes mellitus
  - Gastrointestinal problems
  - duodenal, esophageal and anal atresia,Hirschprung‘s disease
  - Haematological problems
    - Acute leukemia, transient myeproliferative disease
  - Neurological problems
    - Epilepsy, severe behavioral problems, Alzheimer‘s, memory problems

- Sleep problems
  - Sleep apnoea, other sleep disturbance
- Skeletal problems
  - Flat foot, atlantoaxial subluxation
- Visual problems
  - Refractive disorder, squint, nystagmus
- Hearing problems
  - Hearing loss, conductive hearing loss, chronic otitis media

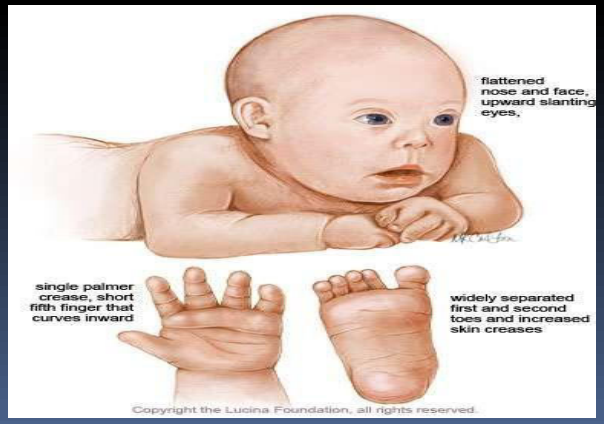
Obesity and nutrient deficiency

Malabsorption (probably linked with celiac disease) due to intestinal damage

- Some has lack of vitamin B12, folic acid and zinc
- Need for antioxidants i.e. vitamin E



Thank you


flattened nose and face, upward slanting eyes,
single palmer crease, short fifth finger that curves inward
widely separated first and second toes and increased skin creases
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Simian crease
Normal palm creases
ADAM.