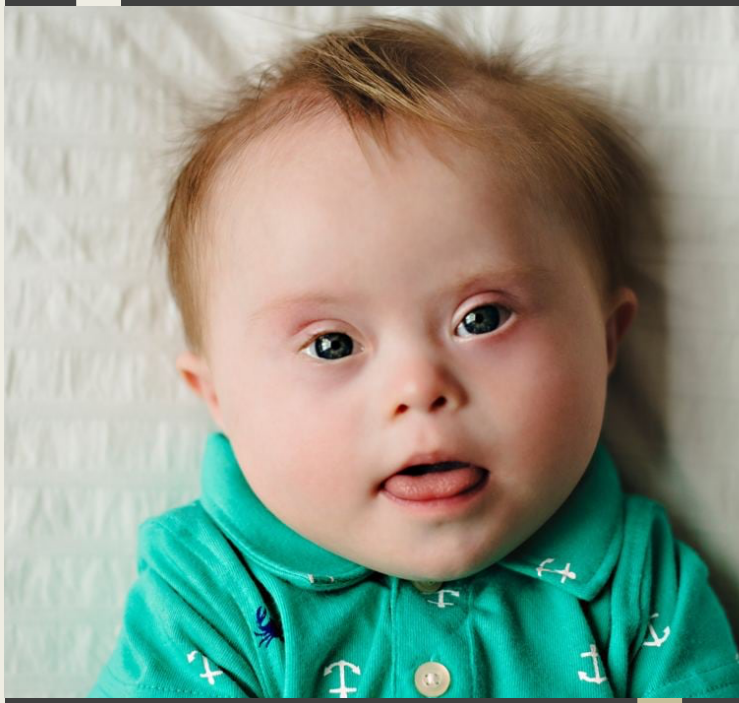
DOWN AND PATAU SYNDROME

Fredrik Christopher Gihle Karlsen
Frederik Glinz
Andrea Femenia Mahiques

INTRODUCTION

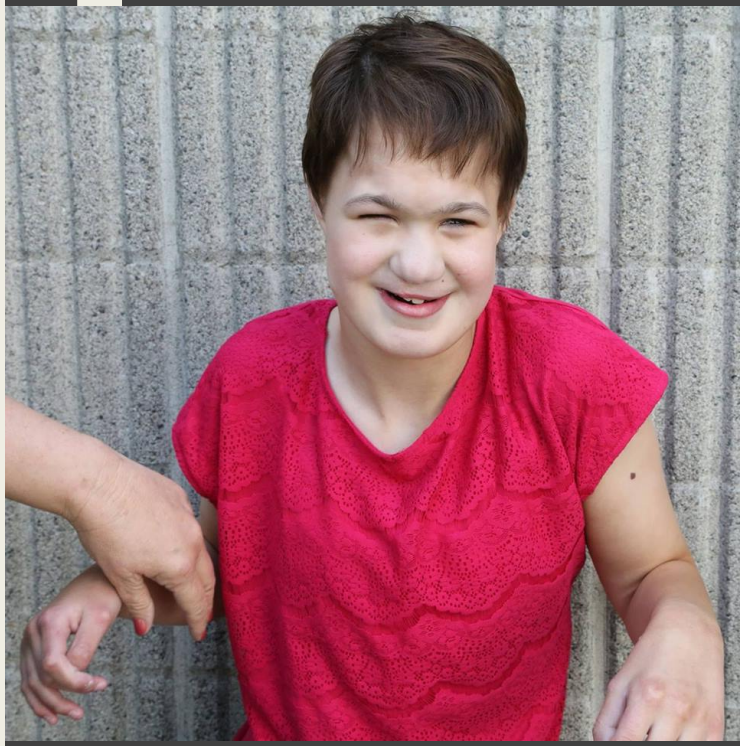
DOWN SYNDROME

- Is a genetic disorder caused by the presence of all or part of **a third copy** of **chromosome 21.**
- It is associated with
  - **Physical growth delays**
  - **Mild to moderate intelectual disability** (IQ= 50, mental ability of a 8-9 year old).
  - **Characteristic facial features**: a small chin, slanted eyes, poor muscle tone, a flat nasal bridge, a single crease of the palm, and a protruding tongue…
- They also have an **increased risk** of a number of other **health problems**, including congenital heart defect, epilepsy, leukemia, thyroid diseases, and mental disorders.

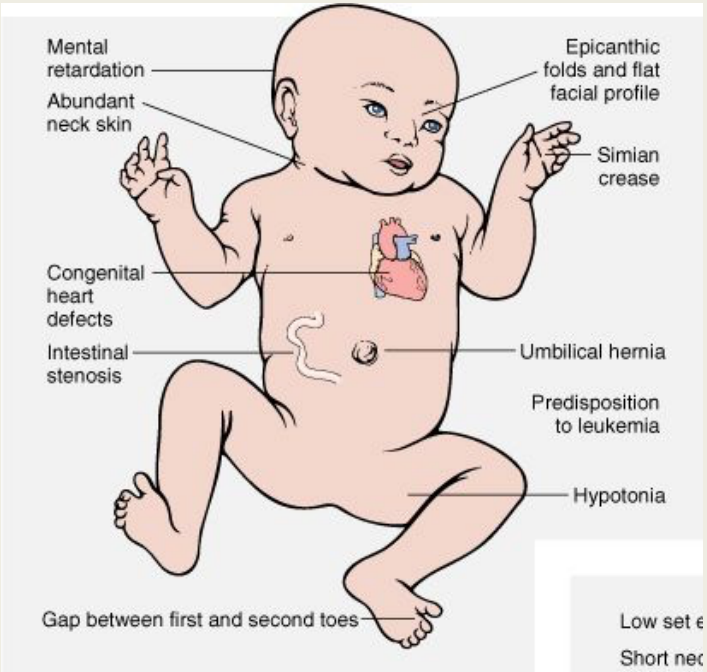


PATAU SYNDROME

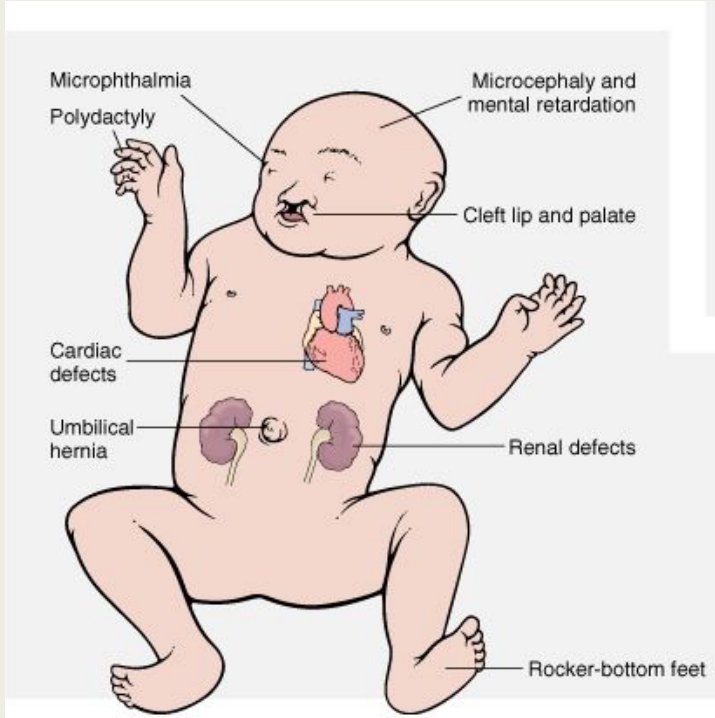
- Is a syndrome caused by a chromosomal abnormality, in which some or all of the cells of the body contain **extra genetic material** from **chromosome 13**.
- This disrupts the normal development leading **to organ defects**.
- **Approximately 90% of infants with Patau syndrome die within the first year of life.** Those who survive, are typically **severely disabled** with:
  - Intelectual disability, seizures, psychomotor issues, physical characteristic features (ex. microcephaly, polydactyly, low-set ears, cleft palate…) abnormal urogenital system, heart defects …



DOWN SYNDROME



PATAU SYNDROME



Causes of Down syndrome

- The vast majority of cases (95%) are caused by trisomy 21, and the incidence increases with maternal age
- Maternal meiotic nondisjunction is the usual cause
- Translocation leads to a familial form of Down syndrome, with significant risk of the syndrome in subsequent children
- From 3% to 5% of cases result from translocation between chromosome 21 and another chromosome. The fertilized ovum has three chromosomes bearing the chromosome 21 material, the functional equivalent of trisomy 21

Characteristics of Down syndrome

- Mental retardation is always present but is highly variable in degree
- Large forehead, broad nasal bridge, wide-spaced eyes, epicanthal fold, large protruding tongue, and small low-set ears
- Brushfield spots, small white spots on the periphery of the iris
- Short, broad hands with curvature of the fifth finger; simian crease, a single palmar crease; and an unusually wide space between the first and second toes

Complications of Down syndrome

- Congenital heart disease, especially defects of the endocardial cushion, including atrioventricular valve malformations and atrial and ventricular septal defects
- Acute leukemia (20-fold increase), most often lymphoblastic
- Increased susceptibility to infection
- In patients surviving into middle age, morphologic changes in the brain similar to those of Alzheimer disease

Maternal screening for Down syndrome

- Alpha-Fetoprotein – low
- Human chorionic gonadotropin (hCG) – high
- Unconjugated estriol – low
- The foregoing assays are referred to as the „triple screen“. When assay for elevated serum inhibin A is also performed, it is referred to as the „quadruple screen“. Expansion of this panel to include other substances such as pregnancy associated plasma protein-A (reduced in DS) and ultrasound studies for physical defects such as increased nuchal fold has shown promise.

PATAU SYNDROME

Causes of Patau syndrome

- Autosomal trisomy of chromosome 13
- Cytogenic types: Meiotic nondisjunction in one parent, Trisomy 13 mosaicism due to mitotic nondisjunction, translocation of chromosome 13.
- Risk Factors: Advanced maternal age, Family history.

Signs and symptoms

- Microcephaly, neural tube defects, deafness
- Microphthalmia, cyclopia.
- Formation of single forebrain with one ventricle (Holoprosencephaly).
- Cleft palat/lip, omphalocele, cutis aplasia.

Diagnose and treatment

- Prenatal ultrasound (week 11-14)
- Prenatal diagnosis (amniocentesis/chorionic villus sampling, beta-hCG, uE3,AFP, quadruple screen)
- Postnatal diagnosis: Fluourscence (FISH) karyotyping
- Treatment: Surgical repair of cardiac defect, cleft palate/lip. Pallative, supportive.