

Inherited disorders result in dental anomalies

Dr. Barta Adrienn

Semmelweis University of Budapest
Clinics of Orthodontics and Paediatric Dentistry

Anomalies

 **inherited (genetic) → mutation**

 **acquired → harms**

 **abnormalities of placenta**

 **radiation**

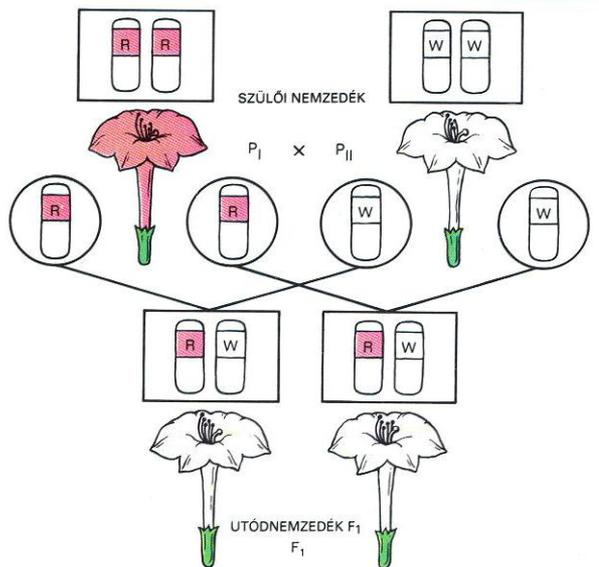
 **medication**

 **virus infections (rubeola, influenza)**

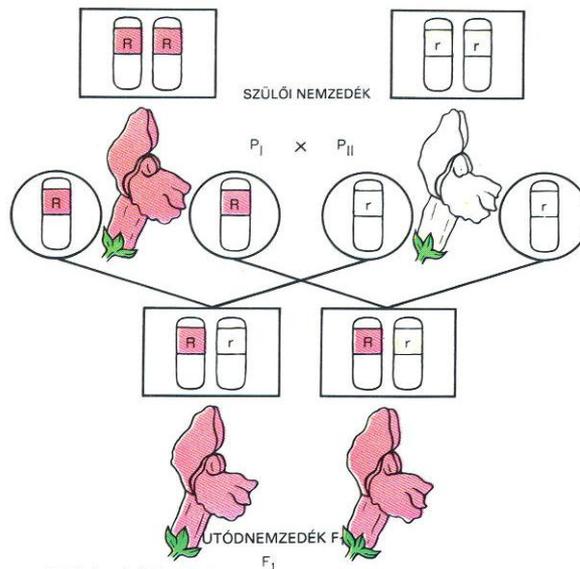
Genetics

Mendel

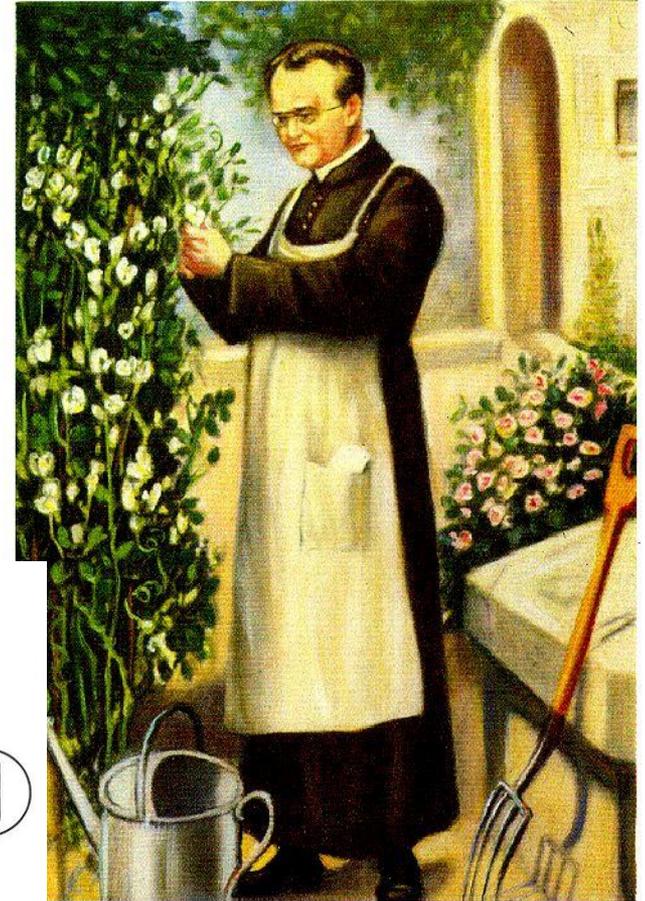
- 📖 forming of the science of genetics
- 📖 1865
- 📖 today: Mendelian laws of heredity
- 📖 he examined 22 different types of pea



intermediér öröklésment
nagy csodátöltésér



domináns öröklésment
oroszlánszáj



Gregor Mendel

Genetic disorders

3 groups:



**anomalies caused by harms of 1 gene
(locus)**

 autosomal (dominant, resessive)

 X-linked (dominant, resessive)



multifactorial inherited anomalies



chromosome aberrations

Autosomal dominant inheritance

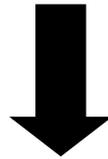
1. **Scheuthauer-Marie-Sainton syndrome** (dysostosis cleidocranialis)
2. **Osteogenesis imperfecta**
3. **Crouzon syndrome**
4. **Apert syndrome**
5. **Treacher Collins-Franceschetti syndrome** (dysostosis mandibulofacialis)
6. **Gardner syndrome**
7. **Waardenburg I. syndrome**
8. **Marfan syndrome**

Autosomal dominant inheritance

1. Scheuthauer-Marie-Sainton syndrome

(Marie and Sainton P, 1898.)

(dysostosis cleidocranialis)



general dysplastic bone formation manifested in typical malformations in skull, pelvis and thoracic region

(abnormal ossification
life)



persistence of sutures for years or for life

prevalence: 1: 1.000.000

Autosomal dominant inheritance

1. Scheuthauer-Marie-Sainton syndrome

(dysostosis cleidocranialis)

General symptoms:

-  short stature
-  ankylosis
-  absent or hypoplastic clavicles
(hypermobility of shoulders)

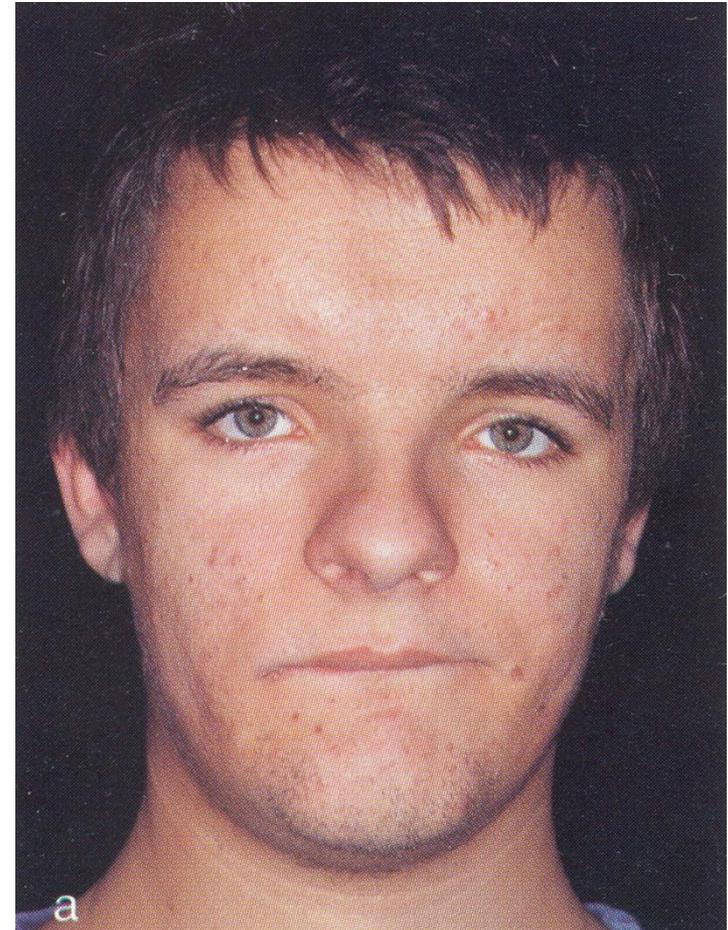
Autosomal dominant inheritance

1. Scheuthauer-Marie-Sainton syndrome

(dysostosis cleidocranialis)

Symptoms:

- 🦷 large, broad and short cranium
(macrocephalus, brachycephalus)
- 🦷 frontal and paritel bossing
- 🦷 broad and depressed nasal bridge
- 🦷 nem légtartó sinusok
- 🦷 hypoplastic midface
- 🦷 relative progenia



Autosomal dominant inheritance

1. Scheuthauer-Marie-Sainton syndrome

Dental symptoms:

-  high and narrow palate
(with or without cleft
palate)
-  supernumerary teeth
-  delayed eruption
-  crowns and roots are in
abnormal localisation
-  teeth in retention are in
ectopic places
-  extremely long-lasting
second dentition

Autosomal dominant inheritance

2. Osteogenesis imperfecta



- complex harm of connective tissue (collagen)
- it is characterized by bones that break easily
- 4 types:

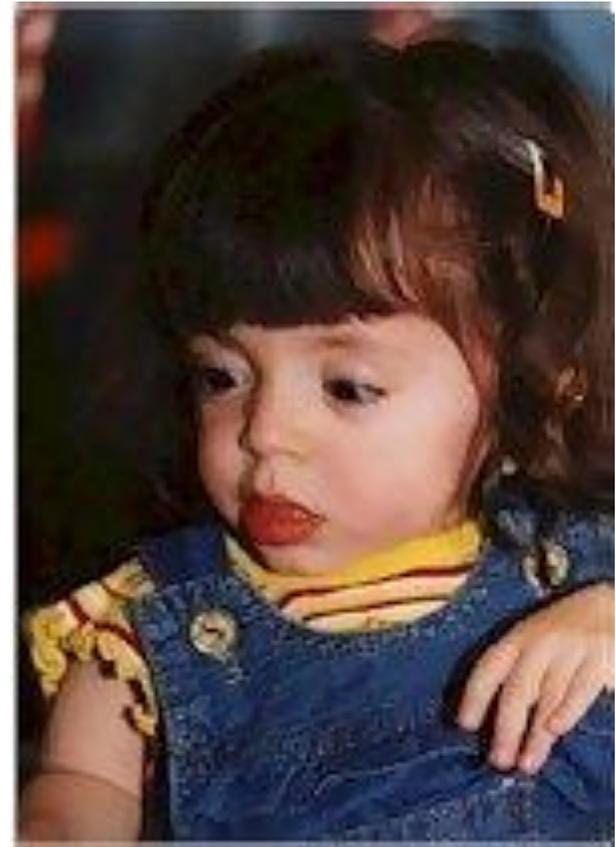
type I – „mild”

type II – lethal

type III

type IV

} - severe



Autosomal dominant inheritance

2. Osteogenesis imperfecta Type I (mild)

Symptoms:

- ⌚ problems with the production of collagen Type I (collagen structure is normal, but the amount is less than normal)
- ⌚ bones break very easily
- ⌚ ossification is quick after fracture, but not normal



Autosomal dominant inheritance

2. Osteogenesis imperfecta

Type I (mild)

Symptoms:

-  normal or near-normal stature
-  blue sclera
-  hearing loss
-  dentinogenesis imperfecta

Autosomal dominant inheritance

3. Crouzon-syndrome

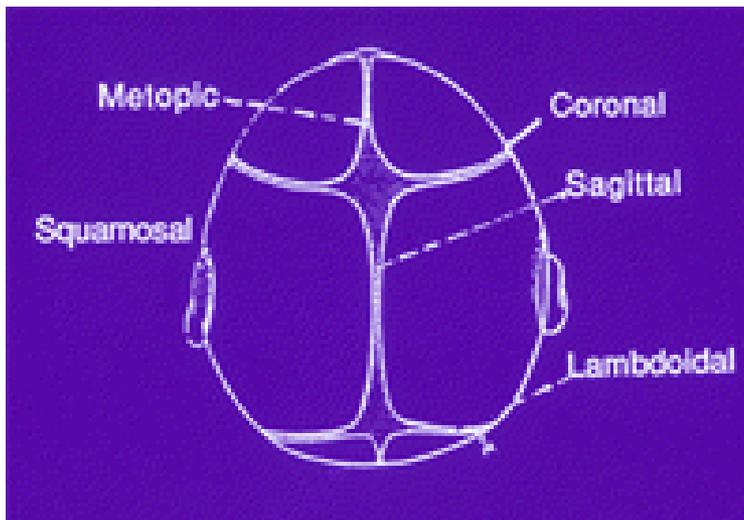
(Octave Crouzon)

(dysostosis craniofacialis)



premature fusion of cranial sutures

(increasing intracranial pressure)



Autosomal dominant inheritance

3. Crouzon syndrome (dysostosis craniofacialis)

Symptoms:

- ☒ cerebral harms
- ☒ mid-face hypoplasia
- ☒ shallow orbits
- ☒ exophthalmus
- ☒ strabismus
- ☒ blindness
- ☒ hearing loss
- ☒ syndactyly



Autosomal dominant inheritance

3. **Crouzon syndrome**

(dysostosis craniofacialis)

Dental symptoms:

- 🦷 maxilla hypoplasia
(narrow arch)
- 🦷 crowding of teeth
- 🦷 progenia
- 🦷 open bite
- 🦷 cross bite (1 or 2 side)



Autosomal dominant inheritance

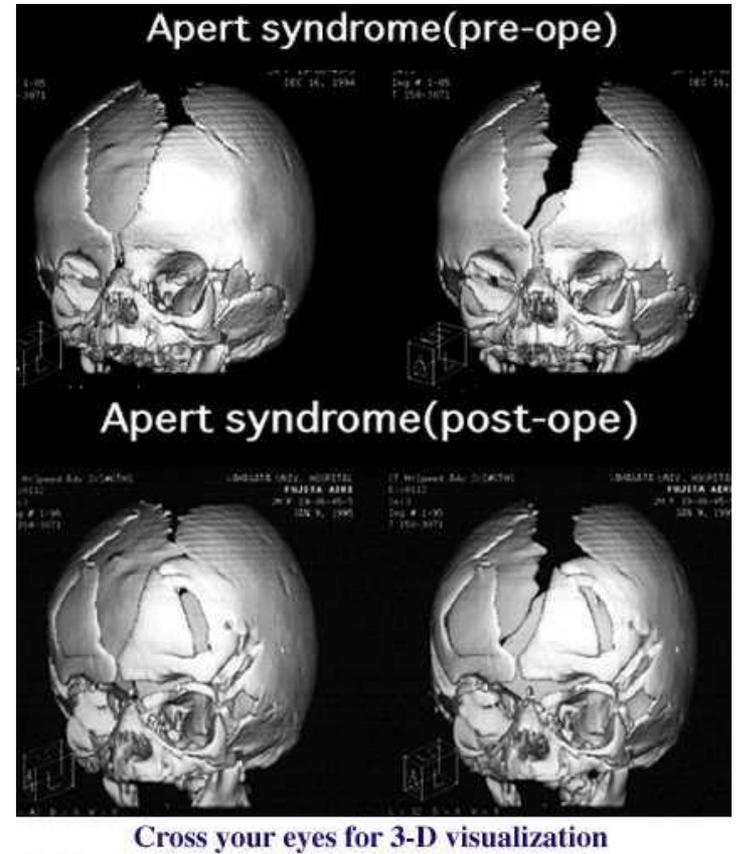
4. Apert syndrome

(E. Apert, 1906)

(acrocephalosyndactyly)



- it is characterized by the **premature closure of the seams** between the skull bones, which results in a peaked head and an unusual facial appearance
- it has many different types in combinations of clinical symptoms and severity
- 1:160.000-200.000



Autosomal dominant inheritance

4. Apert syndrome

(acrocephalosyndactyly)

Symptoms:

-  unusual facial appearance
-  skeletal (limb) abnormalities
-  full-length webbing or fusion between the fingers and toes
-  retarded intellectual development
-  prominent or bulging eyes

Autosomal dominant inheritance

4. **Apert syndrome** (acrocephalosyndactyly)

Dental symptoms:

-  **maxilla hypoplasia**
-  **progenia**
-  **high and narrow palate**
-  **cleft palate**
-  **late dentition**
-  **crowded teeth**

Autosomal dominant inheritance

5. **Treacher Collins-Franceschetti syndrome** (dysostosis mandibulofacialis)



- a defect of the 5q 32-33 locus
- causes mainly facial defects

Autosomal dominant inheritance

5. Treacher Collins-Franceschetti syndrome

(dysostosis mandibulofacialis)

Symptoms:

-  abnormal to almost completely absent external ears
-  hearing loss
-  defect in the lower eyelid (coloboma)
-  narrow face

Autosomal dominant inheritance

5. Treacher Collins syndrome (dysostosis mandibulofacialis)

Symptoms:

-  very small jaw (micrognathia)
-  sinus maxillaris hypoplasia
-  parotis aplasia
-  macrostomia
-  macroglossia
-  cleft palate
-  crowded teeth



Autosomal dominant inheritance

6. Gardner syndrome

Symptoms:

-  multiple osteomas
(particularly of the
jaws and facial
bones)
-  multiple polyps of
the large intestine
-  epidermoid cysts
and fibromas of the
skin
-  polyps have a
tendency to rapid
malignant change
-  **supernumerary
teeth**

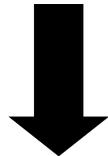
Autosomal dominant inheritance

7. Waardenburg syndrome

(Petrus Johannes Waardenburg: Dutch eye doctor, who first noticed it in 1951.)

there are at least 4 types

WS1, WS2A, WS2B, WS3 (WS4 – alternative)



WS1 (classic form)

- characterized by varying degrees of **hearing loss** and **changes in skin and hair pigmentation**

Autosomal dominant inheritance

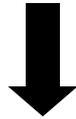
7. Waardenburg I. syndrome

Symptoms:

- 🦷 hearing loss (20%)
- 🦷 disorder of eyelid
- 🦷 differently colored eyes (one eye is usually blue and the other is brown)
- 🦷 distinctive hair coloring
(patch of white hair)
- 🦷 broad nasal bridge
- 🦷 **maxilla and mandibula hypoplasia**
- 🦷 associated symptoms also:
 - 🦷 abnormal development
of heart and limbs

Autosomal dominant inheritance

8. Marfan syndrome



disorder of connective tissue, that affects:

-  skeleton
-  lungs
-  eyes
-  heart
-  blood vessels

defect: a protein called
fibrillin is deficient or
Abnormal
(the fibrillin gene
on chromosome 15)

Autosomal dominant inheritance

8. Marfan syndrome

Symptoms:

- ☒ cardio-vascular system
 - ☒ wide and more fragile aorta
 - ☒ aortic aneurysms
 - ☒ a mitral and/or aortic valve prolapse
- ☒ lungs
 - ☒ spontaneous collapse of the lung (pneumothorax)
- ☒ eyes
 - ☒ near-sighting
 - ☒ dislocation of the ocular lens
 - ☒ retinal detachment

Autosomal dominant inheritance

8. Marfan syndrome

Symptoms:



skeleton:



tall stature



hyperasthenia



**arms, legs and
fingers are long**



scoliosis



pectus deformity



long face

Autosomal dominant inheritance

8. Marfan syndrome

Dental symptoms:

-  high palate
-  malocclusion

Harms of 1 locus



autosomal



dominant



recessive



X-linked



dominant



recessive

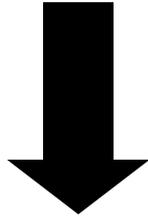
Autosomal recessive inheritance

1. **Dubowitz syndrome**
2. **Ellis van Creveld syndrome**
3. **Pierre Robin syndrome**
4. **Johanson-Blizzard syndrome**
5. **Morquio syndrome**
6. **Sanfilippo syndrome**
7. **Papillon LeFevre syndrome (hyperkeratosis palmoplantaris et periodontoclasia)**
8. **Goldscheider syndrome (epidermolysis bullosa dystrophica)**

Autosomal recessive inheritance

1. Dubowitz syndrome

(1965)



3 main symptoms:

- dwarfism
- eczema
- peculiar face

the phenotype varies from
mild to severe aberrations

about 200 affected patients

Autosomal recessive inheritance

1. Dubowitz syndrome

Characteristics:

-  growth retardation (short stature)
-  primary microcephaly
-  characteristic facial appearance:
 -  small face
 -  micrognathia
 -  broad nasal bridge
 -  telecanthus
 -  broad and high forehead
-  mild mental retardation
-  eczematous skin eruption (min. 50 %)

-  photosensitivity
-  sparse body and scalp hair
-  anaemia
-  anaemia
-  jug-ear
-  short neck
-  poly- or syndactyly

Autosomal recessive inheritance

1. Dubowitz-syndrome

Characteristics:

-  large or small mouth
-  high and narrow palate
-  cleft palate
-  micrognathia
-  dental symptoms:
 -  delayed eruption
 -  crowded teeth
 -  micro- or macrodontia
 -  malocclusion
 -  diastema
 -  oligodontia
 -  fused teeth
 -  doubled teeth

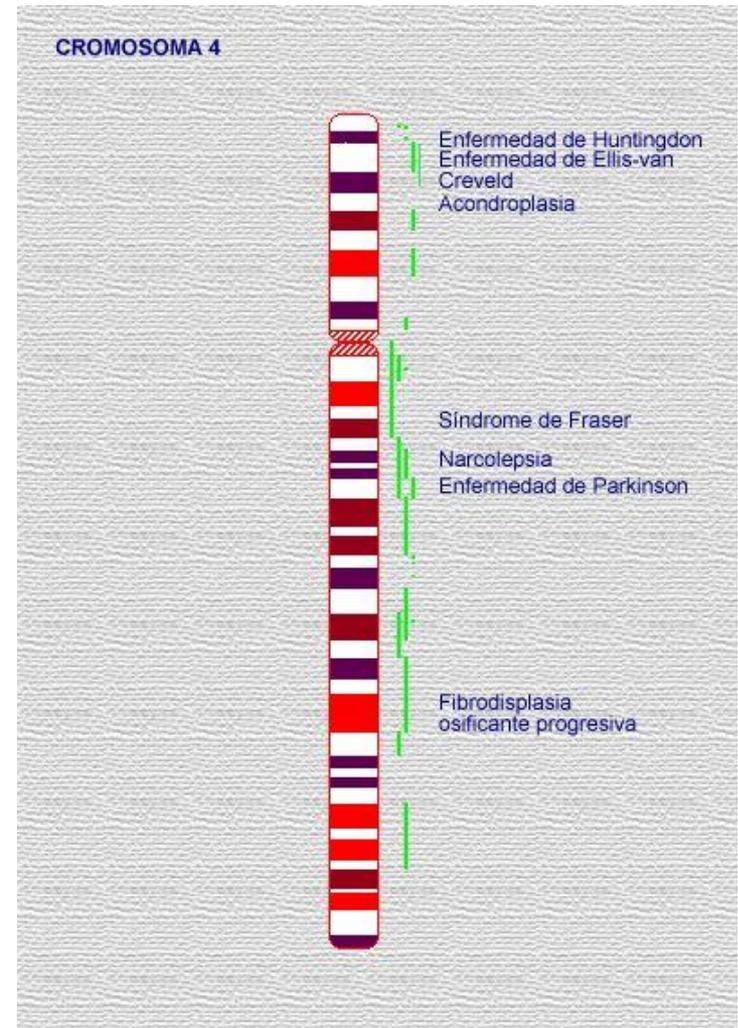
Autosomal recessive inheritance

2. Ellis van Creveld syndrome



chondroectodermal dysplasia
with oral symptoms

- the gene has been mapped to the short arm of chromosome 4 („**EVC-gene**”)
- the function of a healthy EVC gene is not known
- this syndrome is often seen in a little isolated group of people in Pennsylvania



Autosomal recessive inheritance

2. Ellis van Creveld szindróma

Symptoms:

-  malformations of the bones
 -  short limbs
 -  short stature
-  dystrophy of the fingernails and hair
-  mental retardation
-  congenital heart defects (50%)
-  polydactyly (additional digit next to the 5th finger)

Prognosis

Autosomal recessive inheritance

2. Ellis van Creveld syndrome

Symptoms:

-  oral problems:
 -  prenatal eruption of the teeth
 -  mainly lower incisors are affected
 -  hypodontia
 -  abnormal frenulae
 -  missing or peg-shaped teeth
 -  enamel hypoplasia
 -  tendency to get cavities (early dental care!)

Autosomal recessive inheritance

3. Pierre Robin syndrome

It was first described in 1891.



Prevalence: 1 per 8500

3 pathophysiological theories exist to explain the occurrence, the most accepted:

The **mechanical theory**: The mandibular hypoplasia occurs between the 7th and 11th week of gestation. This keeps the tongue high in the oral cavity, causing a cleft in the palate by preventing the closure of the palatal shelves.

Autosomal recessive inheritance

3. Pierre Robin syndrome

Symptoms:

-  respiratoric distress syndrome in the newborn
-  heart defects
-  syndactyly, polydactyly (in 70-80%)
-  clubfeet
-  otitis media
-  hearing loss
-  gastroesophageal reflux
-  esophagitis

Autosomal recessive inheritance

3. Pierre Robin syndrome

Symptoms:

-  micrognathia (91.7%)
-  glossoptosis (70-85%)
-  macroglossia and
ankyloglossia
-  cleft palate

Autosomal recessive inheritance

4. Johanson-Blizzard syndrome

Extremely rare inherited disorder!

Symptoms:

-  congenital absence or underdevelopment of the nostrils: „beak shaped” nose
-  insufficient intestinal absorption (malabsorption)
-  pancreatic insufficiency
-  short stature
-  hypothyroidism
-  weakness of mental and physical activity (psychomotor retardation)
-  congenital hearing loss

Autosomal recessive inheritance

4. Johanson-Blizzard syndrome

Symptoms:

- 🦷 dental symptoms:
 - 🦷 abnormally small, malformed primary teeth
 - 🦷 problems with the development of permanent teeth
 - 🦷 misshapen or absent permanent teeth



Autosomal recessive inheritance

5. Morquio syndrome



2 types:

mucopolysaccharidosis type IVA (galactoseamine-6-sulfatase enzyme deficiency)

mucopolysaccharidosis type IVB (β -galactosidase enzyme deficiency)

Consequence:

accumulation in the body and brain of abnormally large amounts of mucopolysaccharide

Autosomal recessive inheritance

5. Morquio syndrome

Symptoms:

-  short stature
-  coarse facial features
-  abnormal development of many bones including the spine
-  hypermobile joints
-  enamel defects
-  life expectancy: may exceed 20 years

Symptoms are delayed until after the 1st year!

Autosomal recessive inheritance

6. Sanfilippo syndrome

(Dr. Sylvester Sanfilippo, 1963)



- mucopolysaccharidosis
- absence of one of several enzymes
- large amounts of heparan-sulfate (mucopolysaccharide) are excreted in the urine
- 4 main types:

Sanfilippo A	Heparan-N-sulphatase
Sanfilippo B	α -N-Acetylglucosaminidase
Sanfilippo C	AcetylCoA:N-acetyltransferase
Sanfilippo D	N-Acetylglucosamine 6-sulphatase

Autosomal recessive inheritance

6. Sanfilippo syndrome

Symptoms:

-  short stature
-  coarse facial features
-  mental retardation
-  gait disturbances
-  short, massive bones
-  stiff joints
-  liver and spleen enlargement
-  speech disturbances
-  full lips
-  bulky eyebrow
-  gargoyle-face
-  macroglossia

Autosomal recessive inheritance

6. Sanfilippo syndrome

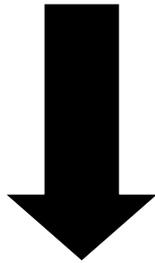
Characteristics:

-  symptoms are delayed until after the 1st year!
bulky eyebrow
-  life expectancy: may exceed 20 years
-  1: 66.000

Autosomal recessive inheritance

7. Papillion LeFevre syndrome

(hyperkeratosis palmoplantaris et periodontoclasia)



main characteristics:

- palmoplantar hyperkeratosis
- juvenile parodontitis



Autosomal recessive inheritance

7. Papillion LeFevre syndrome

Symptoms:

- ☞ right after the eruption of primary teeth:
 - ☞ chr. gingivitis
 - ☞ fast destruction of the alveolar bone
 - ☞ primary teeth are eliminated from the granulated tissue 1-2 years after the eruption
- mucous membrane is intact in the toothless oral cavity
- after the eruption of permanent teeth:
 - parodontitis again
 - fast bone destruction
 - 1st molars are missing before the eruption of incisors
- ☞ individual becomes totally toothless for 14-16 years

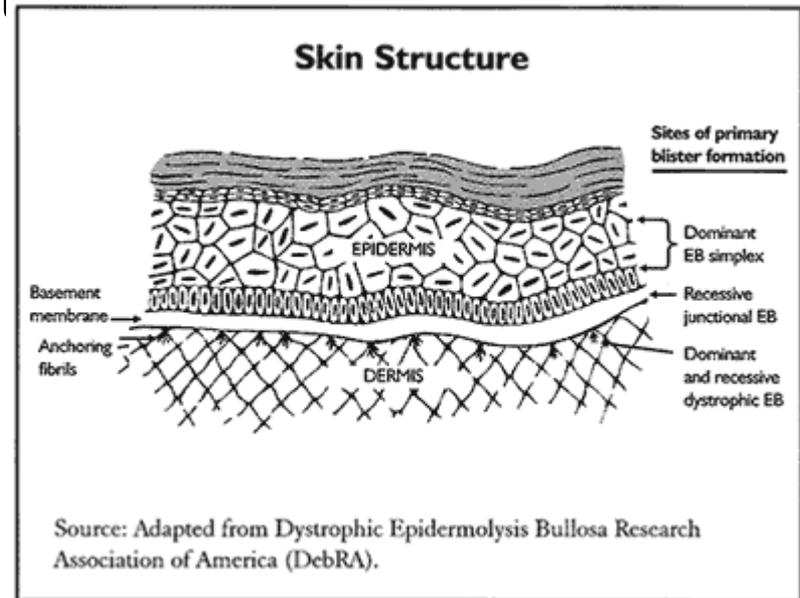
Autosomal recessive inheritance

8. Goldscheider syndrome

(epidermolysis bullosa dystrophica)

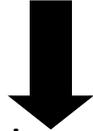
Epidermolysis bullosa (skin blisters develop in response to minor trauma):

- 2-4: 100.000
- more, than 10 genes
- 4 main types:
 - epidermolysis bullosa simplex
 - epidermolysis bullosa junctionale
 - epidermolysis bullosa dystrophica
 - epidermolysis bullosa hemidesmosomale



Autosomal recessive inheritance

8. **Goldscheider syndrome** (epidermolysis bullosa dystrophica)



-  fluid-filled blisters that develop in response to minor trauma or temperature change (or spontaneous)
-  chafing of the skin, rubbing or even increased room temperature may cause blisters to form
-  scarring after blister may cause deformities
-  there are 4 main types and many different subtypes
-  varies in severity from minor blistering of the skin to a lethal form involving other organs

Autosomal recessive inheritance

8. **Goldscheider syndrome**

(epidermolysis bullosa dystrophica)

Symptoms:

-  blistering of the skin
-  nail loss or deformed nails
-  alopecia (hair loss)
-  hoarse cry and cough
-  other respiratory difficulties
-  blistering in or around the mouth and throat
-  **enamel hypoplasia**
-  **tooth decay**

Autosomal recessive inheritance

8. **Goldscheider-syndrome**

(epidermolysis bullosa dystrophica)

Prevention:

- 🕒 keeping rooms at an even temperature
- 🕒 applying lubricants
- 🕒 using simple, soft clothing that requires minimal handling when dressing a child
- 🕒 wearing mittens at bedtime
- 🕒 **regular control in dental office**



Genetic disorders

3 groups:



anomalies caused by harms of 1 gene (locus)



multifactorial inherited anomalies



chromosome aberrations

Harms of 1 locus

 autosomal

 dominant

 recessive

 **X-linked**

 **dominant**

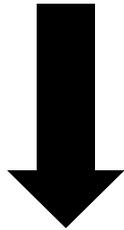
 **recessive**

X-linked inheritance

- 1. Aarskog syndrome**
- 2. Albright-Buttler-Bloomberg syndrome**
- 3. Goltz (Goltz-Gorlin) syndrome**
- 4. Rapp-Hodgkin syndrome (ectodermalis dysplasia)**

X-linked inheritance

1. Aarskog syndrome



X-linked recessive genetic disorder



main symptoms:



short stature



musculoskeletal and genital anomalies



facial abnormalities

X-linked inheritance

1. Aarskog syndrome

Symptoms:

-  short stature, which not be obvious until the child is between 1 and 3
-  delayed sexual maturation
-  mild to moderate mental retardation
-  macrocephalia
-  underdeveloped mid-portion of the face
-  rounded face
-  wide set eyes with droopy eyelids
-  small nose
-  **hypoplasia of the upper and lower jaws**
-  **delayed eruption of teeth**

X-linked inheritance

2. Albright-Buttler-Bloomberg syndrome

Fuller Albright

Allan Macy Butler

Esther Bloomberg



X-linked, dominant genetic disorder

X-linked inheritance

2. Albright-Buttler-Bloomberg syndrome

Symptoms:

-  dwarfism
-  rachitis
-  osteomalatia
-  hypophosphataemia
-  hyperphosphaturia
-  the amount of Ca in the serum is normal!
-  bone anomalies do not respond to vitamin D treatment
-  **enamel hypoplasia**

X-linked inheritance

3. **Goltz (Goltz-Gorlin) syndrome** (focal dermal dysplasia)

(RW Goltz, 1962)



- X-linked, dominant disorder; de novo mutation
- gene: short arm of the X chrom.
- women (1998: 150 females and 11 males)
- it is characterized by malformed skin (appearance, function); hair, nails, teeth also

X-linked inheritance

3. Goltz (Goltz-Gorlin) symptoms

Symptoms:

Skin:

🦷 epidermis: deformed or absent

🦷 dermis: abnormal formation of IV type collagen

Hair:

🦷 missing in patches or completely absent

Teeth and nails:

🦷 unusual appearance

} The skin is completely absent, or discolored, itchy, or blistered.

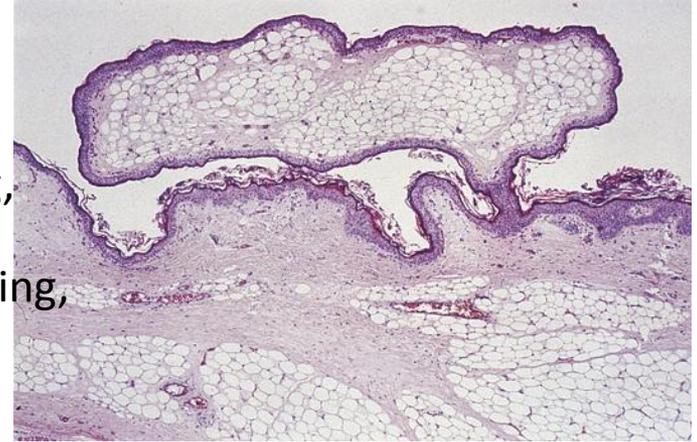
X-linked inheritance

3. Goltz (Goltz-Gorlin) syndrome

Symptoms:

Skin lesions:

- depigmentation
- inflammed, irritated areas (itching, blistering, reddening, swelling, crusting, bleeding)
- papillomas (gums, lips, tongue, vaginal opening, anus)
- excessive skin growth: hyperhidrosis
- hypohidrosis



Skeleton:

- fingers and toes (webbing, fusion or polydactyly)
- curvature of the spine
- underdevelopment or protrusion of the lower jaw
- fused vertebrae

Facial asymmetry

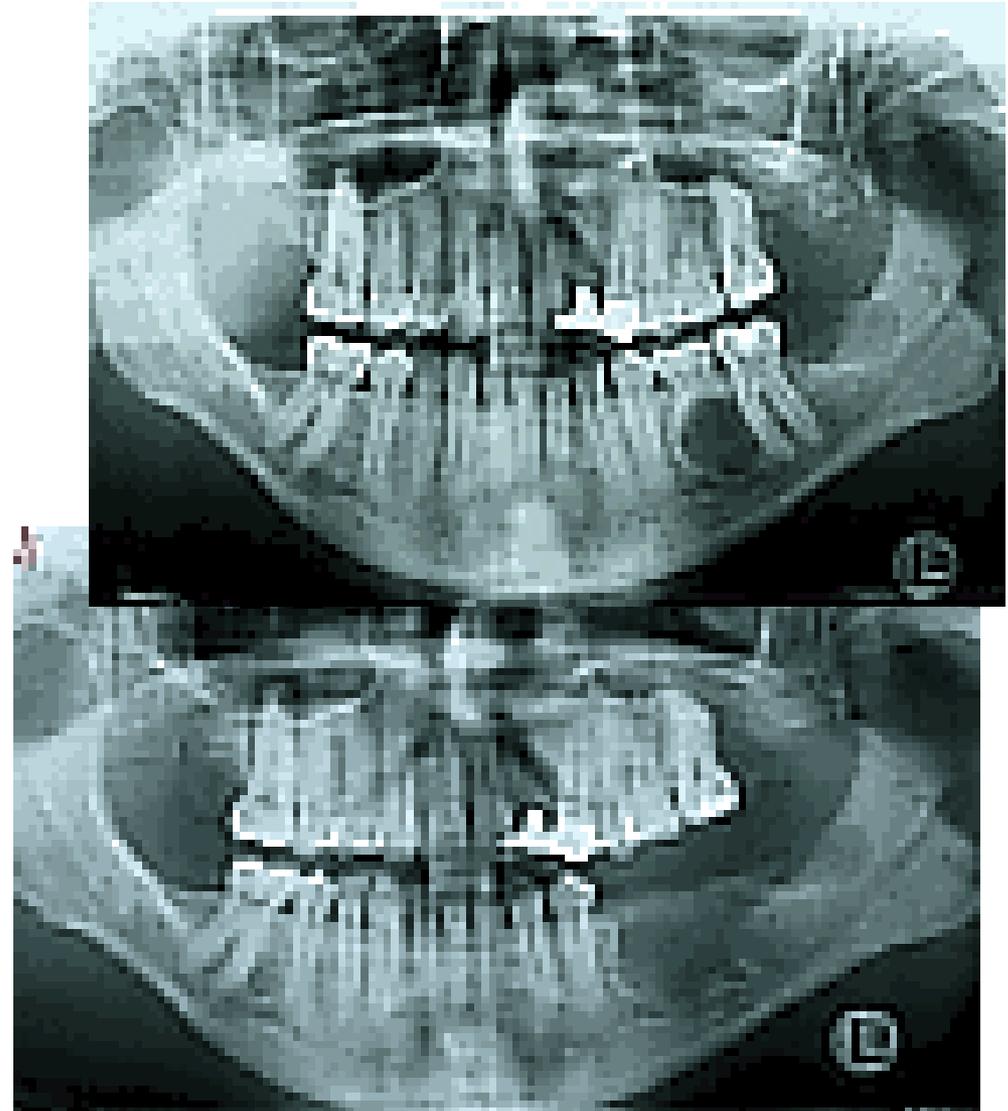
- eyes, ears, chin

X-linked inheritance

3. Goltz (Goltz-Gorlin) syndrome

Dental symptoms:

- ❏ incomplete tooth enamel
- ❏ frequent cavities
- ❏ teeth
 - ❏ malformed
 - ❏ mispositioned
 - ❏ absent



X-linked inheritance

4. Rapp-Hodgkin syndrome (anhidrotic ectodermal dysplasia)



- it has many different types
- X-linked, dominant inherited anomaly is the most common
- it is characterized by abnormal development of the skin and associated structures (hair, nails and teeth, sweat glands)

X-linked inheritance

4. Rapp-Hodgkin syndrome

(anhidrotic ectodermal dysplasia)

Symptoms:

-  absent or decreased sweat and tear glands
-  temperature control is a problem (heat intolerance, extremely high fevers)
-  chronic nasal infections
 -  foul-smelling discharge
-  thin skin
-  decreased pigment
-  photophobia
-  absent or scanty body hair
-  alopecia
-  small and abnormal nails

X-linked inheritance

4. Rapp-Hodgkin syndrome

(anhidroticus ectodermalis dysplasia)

oral structures:

-  **small mouth**
-  **cleft palate**
-  **hypodontia**
-  **enamel hypoplasia**
-  **delayed or absent tooth formation**

Genetic disorders

3 groups:



harms of one locus



**multifactorial inherited
anomalies**



chromosome aberrations

Multifactorial inherited disorders

there are many gene variations in a population (set of genes, environmental effects)

many gene pairs make individual being susceptible to disorders caused by the environment

accumulation can be in a family

risk is higher among near relatives (shared genes)

when the parent is healthy the anomaly will recur in offspring in 1-15%

distribution of normal and abnormal genes (limit)

the limit can be different in males and females

disorders having quantity character:

- hypertony
- ischemic heart disease
- psychiatric diseases
- diabetes mellitus

disorders having quality character:

- cleft palate and lip
- spina bifida

Genetic disorders

3 groups:



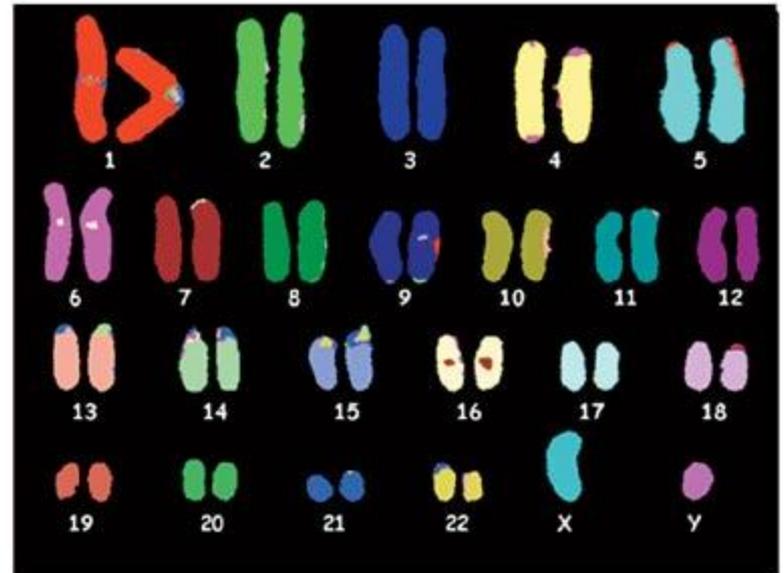
harms of one locus



multifactorial inherited anomalies



**chromosome
aberrations**



Chromosome aberrations



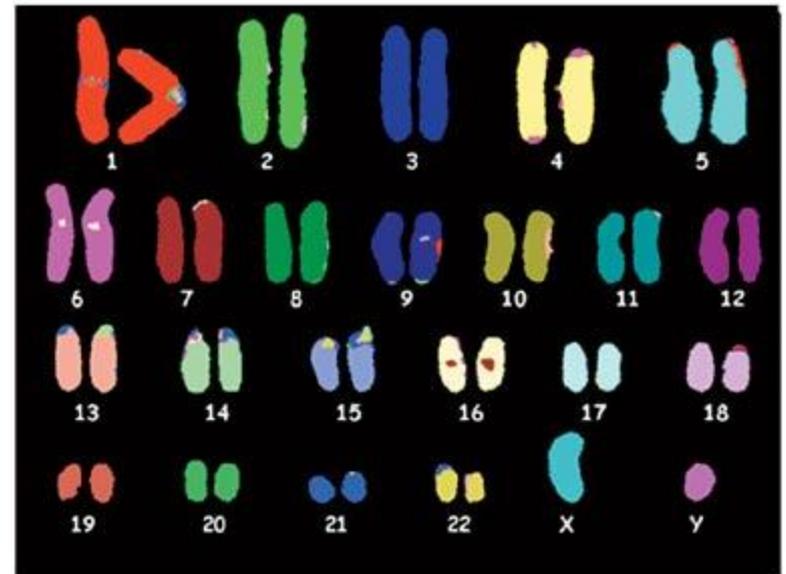
numerical



structural (in shape) – one or more chromosomes have changed morphology (size, pattern)



combined



Chromosome aberrations

Numerical changes

1. Changes in the number of autosomal chromosomes



Down syndrome



Edwards syndrome



Patau syndrome

2. Changes in the number of sex chromosomes



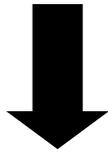
Turner syndrome

Chromosome aberrations

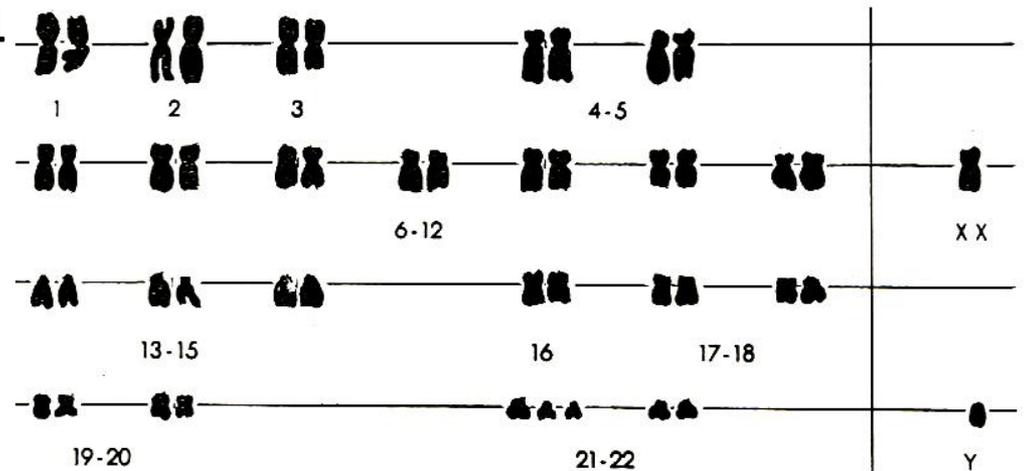
Numerical changes:

(autosomal)

Down syndrome



- the commonest of all malformation, affecting up to 1 in 600 births
- extra 21st chromosome (trisomy 21) in 95%
- it caused by translocation in about 3-4% of people with Down syndrome



Chromosome aberrations

Down syndrome

Symptoms:

-  severe mental retardation
-  short stature
-  microcephalia
-  decreased muscle tone
-  extremely flexible joints
-  characteristic facial appearance
-  mongoloid eye: epicanthal folds over the eye
-  small nose with flattened nasal bridge
-  abnormal outer ear
-  short and wide hand
-  single palmar crease

Chromosome aberrations

Down syndrome

Symptoms:

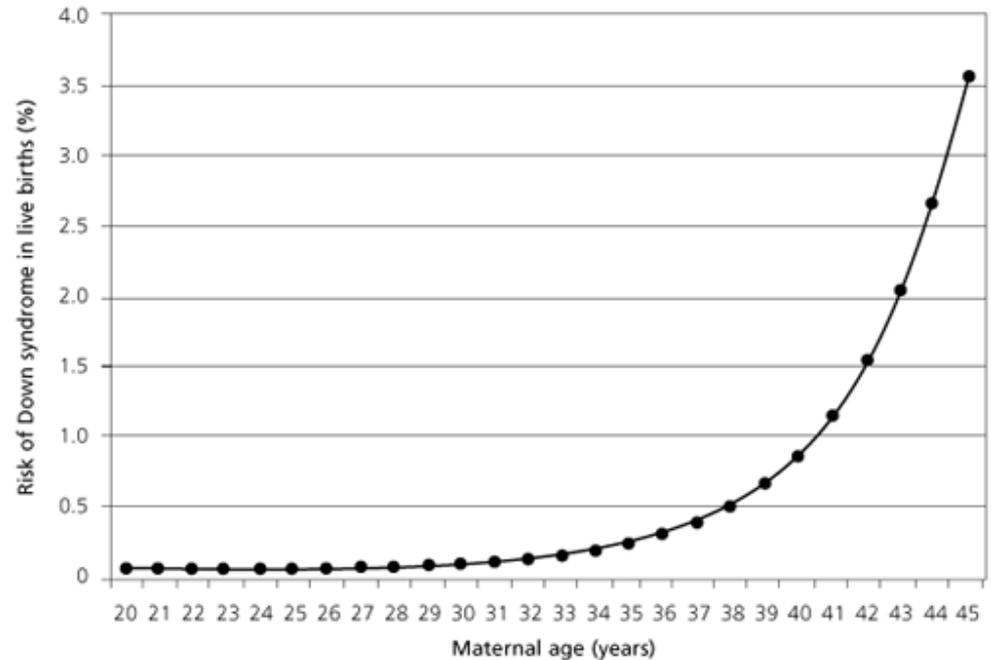
-  macroglossia
-  microdontia
-  hypodontia
-  dysgnathia
-  delayed eruption of teeth
-  cross bite
-  open bite
-  parodontitis
 -  after the eruption of primary teeth
 -  mild gingivitis but deep sulcus
 -  it can not be explained with the bad oral hygiene

Chromosome aberrations

Down syndrome

Symptoms:

- 🕒 frequent congenital heart defects
- 🕒 susceptibility to infections
- 🕒 life expectancy is good
- 🕒 the risk of Down syndrome increases with age after 40 years
- 🕒 prenatal diagnosis (amniocentesis) is recommended for all pregnant women age 35 or older



Chromosome aberrations

Down syndrome

Diagnosis:

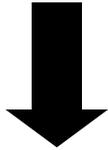
-  screening tests
-  blood tests
-  ultrasound scan
-  chorionic villus testing
-  amniocentesis

Chromosome aberrations

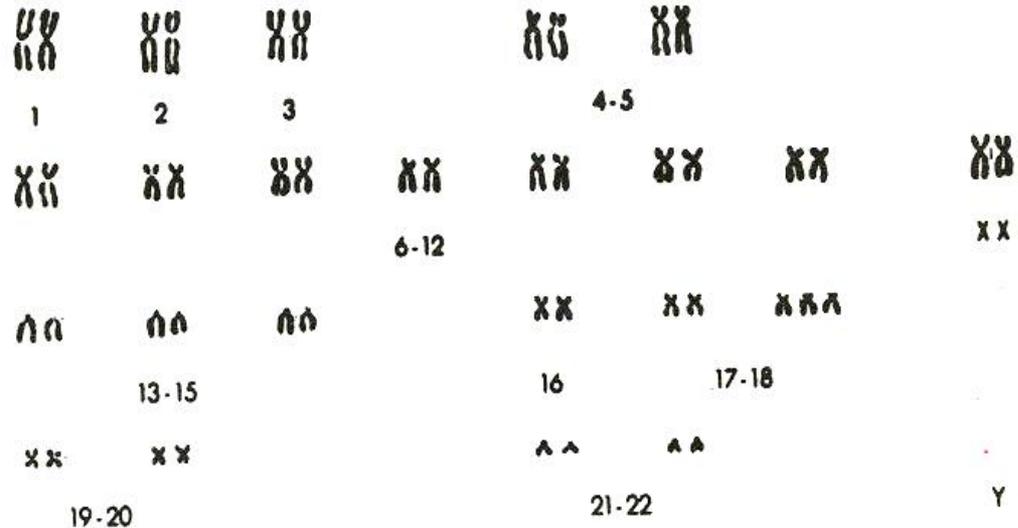
Numerical changes:

(autosomal)

Edwards syndrome



- trisomy 18
- 1 out of 3000 live births
- the multiple abnormalities make hard for infants to live longer than a few months



Chromosome aberrations

Edwards syndrome

Symptoms:

-  severe mental deficiency
-  abnormal development:
 -  heart
 -  kidney
 -  eye
-  clenched hands and underdeveloped fingernails
-  microcephalia
-  flat forehead
-  protruded occiput
-  abnormal ears
-  **cleft palate and lip**

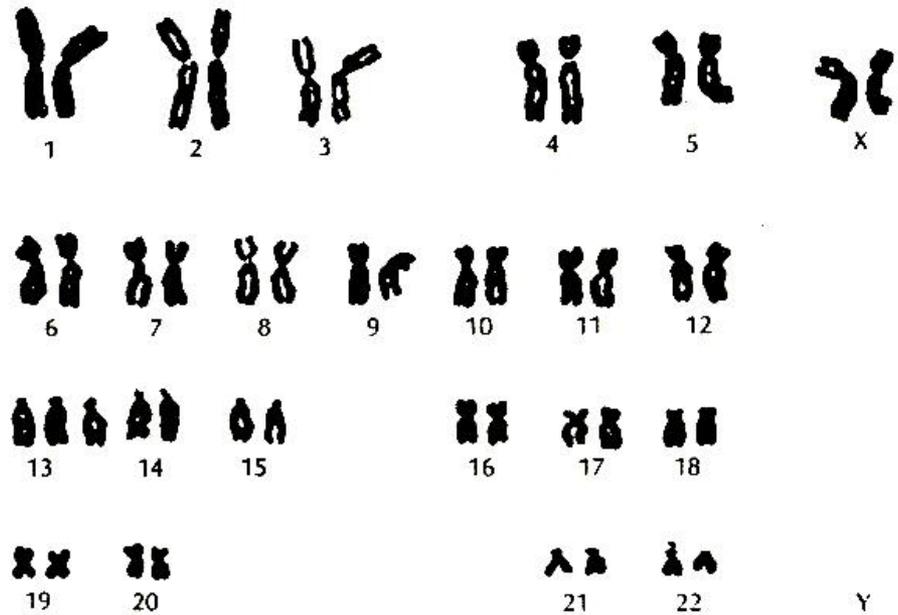
Chromosome aberrations

Numerical changes:

(autosomal)

Patau syndrome

- trisomy 13
- frequency: 1 out of every 5000 live birth
- the multiple abnormalities often are not compatible with life (80% of children die in the 1st month)



Chromosome aberrations

Patau syndrome

Symptoms:

-  multiple congenital aberrations
-  severe mental retardation
-  congenital heart diseases
-  poly- or hexadactyly
-  microcephalia
-  wide nasal bridge and face
-  small eyes (microphthalmia)
and eyes close set
-  **cleft palate and lip**

Chromosome aberrations

Numerical changes

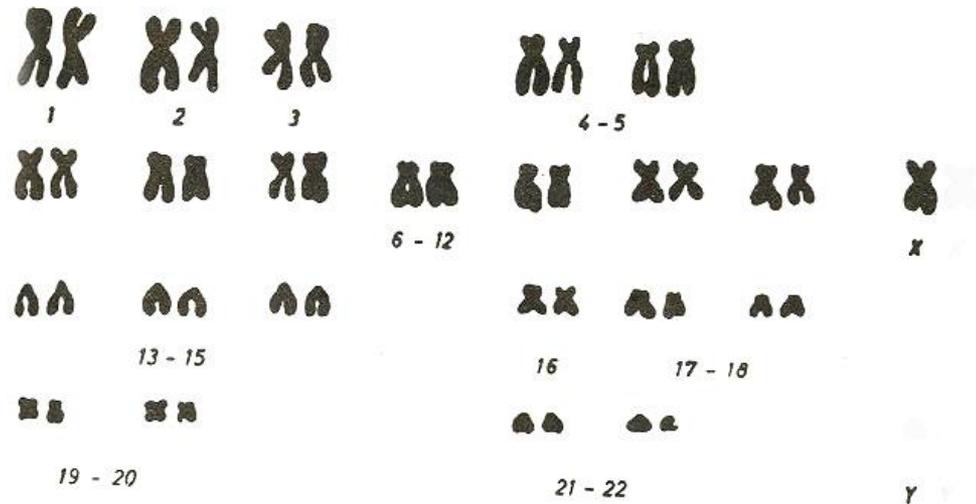
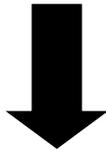
1. Changes in the number of autosomal chromosomes
 -  Down syndrome
 -  Edwards syndrome
 -  Patau syndrome
2. **Changes in the number of sex chromosomes**
 -  **Turner syndrome**

Chromosome aberrations

Numerical changes:

(gonadal)

Turner syndrome



- monosomy X
- frequency: 1 out of 2000 live birth

Chromosome aberrations

Turner syndrome

Symptoms:

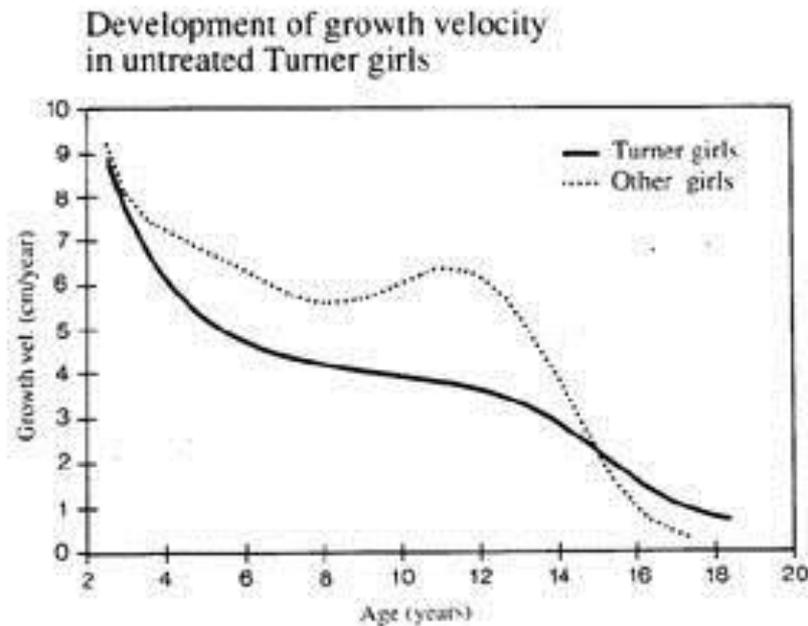
-  it occurs only in females
-  sexual infantilism or gonadal dysgenesis
-  mental deficiency
-  short height
-  webbed neck
-  low hairline
-  underdeveloped body hair
-  „shield-shaped” chest
-  heart and kidney abnormalities

Chromosome aberrations

Turner syndrome

Dental symptoms:

- 🦷 mandibula hypoplasia
- 🦷 narrow and high palate
- 🦷 crowded teeth



Inherited disorders result in dental anomalies:



early diagnosis



preventive treatment (required time and required methods)



special knowledge



thoroughness



more often control as usually

Thank you!