Oral pathology in children with dysmorphic features

 Syndromes
Dysmorphic features are developmental disabilities including the maxillofacial area, due to:

- Congenital disorders
- Genetic syndromes
- Birth defects

Electronic data base of syndromes - www.possum.net.au
Dysmorphic features form the group of malformations

More often, they are rare diseases
### Rare diseases

- **Rare diseases**
  - affect fewer than 1 in 2 000 people.

- **In most cases, the reason therefore is**
  - genetic, often chronic and life-threatening. Over 50% of affected are children.

- **Known are over 7000 different types of rare diseases.**
  - According to the World Health Organization in Bulgaria are affected by them a total of about 400,000 people.

- **Diseases are often chronic and severe.**
  - The treatment does not affect the genetic cause and correct the consequences.
A term used in medicine and psychology;

We are talking about a syndrome, when group of symptoms are observed, always together;

Syndrome may be specific disorder, disease, or condition.
The syndrome is:

A combination of clinical signs and symptoms, which are always found together, characterize the disease;

The syndrome is usually congenital or genetic disease.
Syndrome and associated factors

We have to distinguish:

- clinical signs forming syndrome;
- And secondary symptoms, which occurs in large% of patients with a particular disease, but is not mandatory.
The syndromes are:

- multiple defects with common pathogenesis derived from hereditary or congenital reasons.

The most syndromes are:

- recognizable models of various malformations;

but there are syndromes that are not malformations!
In many syndromes a cognitive impairment occur

Cognitive disorders of mind include issues related to:

- memory,
- logical thinking,
- attention, perception, imagination,
- ability to make decisions and others.

Language
Some syndromes are part of oral pathology in childhood

Very often occur in infancy, and according to the severity of the disability children with such diseases do not survive long.
Important for dentist!

The common between these diseases is that oral pathology is part of a diverse systemic pathology that affects both physical and mental development of the child.
Most syndromes with oral manifestation are caused by genetic defects

Genetic defects are most commonly in the child development

- They are chromosomal abnormalities

More often there are defects

- in ectodermic structures and collagen synthesis.
The oral anomalies in the syndromes affect

- Tooth structure,
- Teeth count, The form of the teeth,
- Often combined with periodontal disabilities,
- and other defects associated with different body parts.
Developmental disabilities are divided into three main groups:

- Malformations
- Disintegration
- Deformities
Malformation is a structural defect in the body due to abnormal embryonic or fetal development.
Causes of malformations

- Poorly formed tissues - primary structural abnormalities;

- Poor prognosis for a normal growth in a particular area;

- Incomplete morphogenesis - agenezis, hypoplasia;

- Impaired apoptosis - spina bifida, syndactyly;

- Incomplete migration ectomesenchyme - syndrome Di Georgi
An example of formation of defects in a Di George syndrome

- Impaired migration of ectomesenchym;
- Migration of neural crests (ectomesenchym) stimulate organs and tissues differentiation;
- Maxillofacial growth is part of them.
Specific faces: low set ears, wide distance between the eyes – result of malformations.
Di Georgi’s syndrome

Tetrad of Fallot
- low calcium and high phosphorus in the blood.

Thymus dysfunction
- immune defects, frequent infections.

Hypoparathyroidism

Cleft palate,

Impaired mental and physical development and others.
Deformities

- Unusual forces act on normal tissue during fetal development.

Disintegration

- Destruction of normal tissue caused by vascular, infectious, physical reasons.
The cause of all these developmental disabilities are genetic defects.
Basic philosophy of modern molecular biology say:

The properties of the cells, tissues and organisms are determined by the properties of their proteins!

- Genes control: the structure of proteins; the time of their synthesis; the required amount; RNA is mediator.
Genes are grouped into DNA strands that form chromosomes

Person has 46 chromosomes containing 80,000 genes:
- XX - female sex;
- Y - male sex;
- 44-autosomal;

23 pairs of chromosomes are formed:
- 22 autosomal pairs
- 1 sex pair
Each cell contains a complete and accurate copy of the genomic DNA;

Different cells express different regions of the genomic DNA;

Each cell knows when, who gene to be expressed.
Genetic syndromes

Chromosomal defects may be the cause;

Defects in single gene or multifactorial gene defects may be observed;

Autosomal dominant, autosomal recessive sex-linked inheritance occur;

Secondary gene defects influenced by the environment may occur, also.
Chromosomal mutations may be:

**Numerical chromosomal aberrations**

- **Aneuploidy**
  - trisomy, monosomy, polysomies;

- **Mosaicism**
  - inappropriate segregation of chromosomes during mitosis - the defects are in all tissues;

- **Poliploidy**
  - triplets, tetraploidy
  - incorrect cell division - often leads to pregnancy.
Structural chromosomal aberrations

Cleavage of the chromosome and the change of its normal configuration

Inside chromosomal
- Deletions
- Duplications
- Ring-chromosome
- Inversions and others.

Inter chromosomal

Chromosome fragility;

With the absence of chromosomal material
Different gene mutations may be observed

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
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<tbody>
<tr>
<td>Duplications</td>
<td>More than one copy of a chromosome in the genome;</td>
</tr>
<tr>
<td>Deletions</td>
<td>Loss of chromosome segment;</td>
</tr>
<tr>
<td>Inversions</td>
<td>Flip the beginning and end;</td>
</tr>
<tr>
<td>Translocations</td>
<td>Detached segment is attached to another chromosome.</td>
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</tbody>
</table>
Besides hereditary and congenital mutations, there are environment factors causing mutations in the life

- **Physical** - X-rays and radiation;
- **Chemical** - pesticides, heavy metals, anesthetics and the like.
- **Biological** - oncogenic viruses
Besides the development, mutations can also affect physiology

Mutations affecting the structural proteins:
- Collagen - $\frac{1}{4}$ of the proteins
- Family of collagen genes - 35 gene

On the enzymes regulating processes in the body;

Accumulation of metabolic products.

Reduced activity of the receptor proteins.
Dysmorphic features - malformations
Wolf-Hirschhorn Syndrome

- 4chr. deletion
- Common features:
  - Microcephaly
  - Broad nose
  - Ocular anomalies
- Oral manifestations:
  - Cleft palate - 40%
  - Cleft lip and palate - 10%
  - Micrognathia
  - short philtrum
  - lips downward angle
Cri-du-Chat Syndrome

5 chr. deletion

Common feature:
- like a cat scream
- microcephaly
- round face
- low ear shells
- severe psychomotor problems
- mental retardation
- hypotension
- hypertelorism
- early mortality

Frequency - 1: 20000

It occurs in 1% of children in institutions
Cri du chat syndrome – oral symptoms

- Macrognathia;
- Malocclusion – overjet
- Cleft lips and palate
Patau Syndrome

Trisomy 13

Common feature
- Microcephaly
- Microphthalmia
- Polydactyly
- Mortality - 86% to 1d.

Oral symptoms
- Micrognathia - 45%
- Cleft lip
- Agenesis of the frontal alveolar upper jaw
Prader-Willi Syndrome

15chr. deletion

General characteristics:
- Hypertension, obesity, small limbs; hyperpigmentation; diabetes;

Oral symptoms
- dental caries
- enamel hypoplasia
- malocclusion, calculus,
- decreased and viscous saliva,
- gingivitis;
- microdontia
- high palate
Prader-Willi syndrome (PWS)

1 : 12,000 to 15,000

Oral manifestations, also:

- Reduced salivation;
- Low pH, buf. capacity;
- Emotional and behavioral problems;
- Oral mucosal lesions;
- Common candidiasis;
- Edema of lower lip.
Angelman Syndrome or „Happy Puppet Syndrome“

15chr. deletion

Oral symptoms:
• macroglossia;
• drooling;
• prognathia;
• macrostomia
• tremas
Angelman syndrome

Happy puppet syndrome - often because of causeless laughter and uncontrollable movements of the arms, against the background of mental problems and ataxia.
Edwards Syndrome

Trisomy 18

General characteristics
- Convexity of the occipital part;
- Short palpebral fissures;
- Characteristic thumb;

Oral symptoms
- Micrognathia;
- High palate;
Syndrome of fragile X chromosome

- The most common cause of mental retardation;
- The most common cause of autism;
- 1: 4000 boys; 1: 8000 girls;
  - mutation of the FMR1 gene;
  - convex forehead
  - big ears;
  - apart eyes.
Syndrome of fragile X chromosome

Oral symptoms

- high and narrow palate;
- progeny;
- crossbite,
Klinefelter syndrome

- 47 chromosomes observed;
- The 47th is XX;
- It may be inherited, but may be innate also;
- Recognizable features not detected at birth;
- It affects only men and no mental deviation occur;
  - osteoporosis; wide pelvis; testicular atrophy are observed.

Oral manifestations:
- progenia; large tooth crowns; taurodontizm;
- Taller than average height
- Reduced facial hair
- Reduced body hair
- Breast development (gynaecomastia)
- Osteoporosis
- Feminine fat distribution
- Small testes (testicular atrophy)
Trizomiya 21 - Down Syndrome

Oral characteristics:

- Enamel hypoplasia - temporary teeth;
- Periodontal pathology - 90% and 30% ANUG
- Hypoplastic sinuses;
- Hypodontia – of 8, 7, 2th teeth;
- calculus, tooth decay, reduced salivation;
- Taurodontism;
- Malocclusion - cross and open bite;
- Macroglossia and fissured tongue;
- Dry and chapped lips – lips fissures
Down syndrome - symptoms

Flat occipital region;

Brachydactyly - short fingers,

Clinodactyly - curved fifth finger.
Down syndrome

- Mental retardation;
- White spots around the iris;
- Multiple immune defects;
- Cheilitis;
- Blepharitis.
Down syndrome

Hypoplasia of the teeth;

Often aggressive periodontitis.
Syndromes due to defects in single gene
Neurofibromatosis (NF-I)

Common symptoms:
- Six or more cafe-au-lait spots at birth and axillary freckling in a child
- Lisch nodules in iris
- Multiple neurofibromas about back
- Hypertrophy of leg and fingers, pseudarthrosis of tibia
- Unilateral neurofibromatosis

Multiple spots - "cafe-au-lait"

Multiple neurofibromas

Bone turnover.
Neurofibromatosis (NF-I) - Oral symptoms

- Expanding fungiform papillae;
- In oral fibromatosis – macroglossia
- Hypoplasia of the oral soft tissues;
- Bone hypoplasia;
- Malposition of teeth;
- Wide and low situated mandibular canal.
Neurofibromatosis (NF-I)

1 fig.
- Wide bone defect in the left side of the upper jaw;
- Wide mandibular canal in the same area.

2 Fig.
- 10 year child;
- Bone defect on the left shoulder of the lower alveolar jaw.
Tuberous Sclerosis

Common features
- Angiofibromas
- Fibromas under the nails
- Convulsions, epilepsy
- Mental retardation

Oral symptoms:
- Gingival fibromatosis – epulis
- Enamel hypoplasia

Multiple white macules on back of infant

Angiofibromas about nasolabial folds

Shagreen patch lumbosacral area

Gingival fibromas

Lytic lesions of hand bones

Subungual fibromas about toes and fingers

CT scan with periventricular calcifications
Tuberous Sclerosis

Enamel hypoplasia

Growth in different organs;

Epilepsy, learning disabilities, autistic spectrum disorders;

and kidney problems may occur.
Apert syndrome – disease with acrocephalo-syndactyly

Congenital malformation of the skull, face, hands and feet;

This is of the group of defects in brachial arcs - of pharyngeal arch (lower, upper alveolar jaws);

At the extremities - impaired cellular apoptosis, which releases the fingers - linked fingers.
Apert syndrome - oral symptoms

- High palate with lateral swellings
- Heavy malocclusion - open bite;
- Hypoplasia of teeth in half persons;
- Progenia;
- Decreased nazofaringeal distance.
Stickler Syndrome

Oral symptoms
- Hypoplasia of the face
- Long palate filtrum;
- Mobile soft palate.

General symptoms
- Myopathy - retinal detachment;
- Hypermobility joints;
- Flat face.
Stickler Syndrome
Rett Syndrome

It is combined with autistic-like symptoms in girls;

Start at 6 m 3 years, to this age - normal development occurs;

Stereotyped movements - such as cleaning, applause, etc;

Hands in his mouth;

Respiratory dysfunction - apnea;

Aerofagia, hyperventilation.

Bruxism; dystonia;

Scoliosis, kyphosis
Smith-Magenis syndrome - It is not genetic but congenital disease

Frequency - 1: 25000,
Medium intellectual level;

Delayed speech and speech problems;
Typical vision;

Behavioral problems;
Disturbed sleep from infancy;

Aggression, impulsivity; anxiety; self-harm.
Smith-Magenis syndrom

Taurodontism;

Wide pulp chamber downloaded apically;

Defective development of the Hertvig Brun vagina.
Turner Syndrome

General symptoms

- Lymph edema of the hands and feet;
- It is not associated with delayed mental development;
- Hollow nails;
- Convex chest;
- It affects the girls;
Turner Syndrome

Oral manifestations

- Macrognatia
- Early eruption of the 1st molars;
- High palate;
- Malocclusion.
Coffin-Lowry Syndrome

Oral symptoms

- Openbit with a relaxed lower lip;
- Malocclusion - progenia;
- Hypodontia (lower permanent teeth);
- Periodontal disease - more frequently;
- Dental hypoplasia.
Coffin-Lowry Syndrome

General symptoms

- Square forehead
- Protruding ears
- Severe mental disabilities.
Lesch-Nyhan Syndrome

- Self-harm;
- Impaired purine metabolism;
- Accumulation of uric acid;
- Spasticity;
- Delayed mental development;
- Arthritis, kidney problems; Decreased muscles tonus.
Lesch-Nyhan Syndrome

Self Injure lips and nails

Varying degrees of self-mutilation of lips

Self-mutilation of fingers

Tophi on pinna
Syndromes with defective ectodermal formation

Obligatory for the exam 67-84sl.!!
Ectodermal dysplasias

- Hair, teeth, nails, and salivary glands;
- They are 10 different species.

Hereditary dysplasia of ectodermal structures –

According to the affected genes –
- Hay-Wells syndrome, Rapp-Hodgkin syndrome and EEC syndrome - gene TP63.[3]
- Hypohidrotic ectodermal dysplasia ED – гени A, EDAR, EDARADD
- Margarita Island ectodermal dysplasia - gene PVRL1
- Ectodermal dysplasia with skin fragility - gene PKP1
- Clouston's hidrotic ectodermal dysplasia - gene GJB6
- Naegeli syndrome/Dermatopothy pigmentosa reticularis - gene KRT14
- Pachyonychia congenita - gene of keratine
- Focal dermal hypoplasia - gene PORCN
- Ellis–van Creveld syndrome - gene EVC
- Palmoplantar ectodermal dysplasia – affect the hands and the feets.
Ectodermal dysplasia

Transmitted through sex chromosomes;

Characteristics:

• Hypohidrosis - reduced secretion of sweat;
• Hypotrichosis - sparse hair, thin hair;
• Hypodontia;
• Normal psychological development.
Ectodermal dysplasia
hypodontia; conical teeth
Hypohidrotic Ectodermal Dysplasia

It can be seen during the first year after birth with temperatures of unknown origin, mainly in the summer.

with a teeth eruption or lack of temporary teeth;

Teeth with a conical shape.
Hypohidrotic Ectodermal Dysplasia

- Xerostomia due to hypoplasia of the salivary glands may be rarely observed;
- Reduced to a lack of perspiration (anhidrosis), due to a severe reduction in sweat glands;
- Dry skin, thin hair, decreased nasopharyngeal and tear secretion;
- Changes in voice due to atrophy of the laryngeal mucosa.
Ectodermal dysplasia

Typical faces
Ectodermal dysplasia
Ectodermal dysplasia

Severe hypodontia;
Retention of temporary canines;
Lack of permanent teeth.
Ectodermal dysplasia

Teeth anomaly
Behavior of dentists of children with ectodermal dysplasia

- Esthetic restorations of the conical teeth;
- If missing teeth occur - conventional dentures may be used, that may be changing periodically.
Orthodontic treatment have to accompanied the teeth eruption

Retainers may be used
Ectodermal dysplasia

In the beginning:
- Specific prevention program orthodontic control

In the future:
- Crowns, bridges, implants, after completion of the growth of the jaws
Ellis-van Crefeld Syndrome – Chondroectodermal dysplasia (cartilage disorder)

- Autosomal recessive inheritance.
- Skeleton, mouth, hair and nails, heart, eyes, nervous system are Impacted;
- Bilateral polydactyly over numburing fingers, short, thick limbs, elongated thorax;
- Chondrodysplasia (cartilage disorder) in the field of long bones;
- Thin hair, dystrophic nails;
- Frequently congenital heart defects and genital abnormalities occur.
Ellis-van Crefeld syndrome

Oral manifestations:
- Fusion of the lips and attached gingiva in the frontal area;
- Lack of teeth;
- Small conical teeth;
- Presence of teeth at birth occur.
Goltz- Gorlin syndrome - focal dermal hypoplasia

Defects in the number of fingers; microphthalmia;
Hypodontia; Enamel hypoplasia;
Linear skin defects.
Goltz- Gorlin syndrome
Kleydocranial dysplasia

Autosomal dominant inheritance;

6 chromosome 21 - ST FA1 gene;

Aplasia of the clavicles

The fontanelle Ossification;

Convex front;

Hypertelorism and maxillary hypoplasia.
Hyperodontia
Kleydocranial dysplasia

- **Hyperdontia**
- Conical teeth;
- Late teeth eruption;
- Dento-alveolar cysts;
- Absence or impaired cellular cement;

**Treatment:**
- Extraction of unresorbed primary teeth.
- Teeth extraction of hyperdontia;
- Orthodontic treatment.
“Papillon – Lefevre” syndrome

- Autosomal recessive genetic disease;
- Rarely occur 1: 3,000,000;
- Palmar-plantar hyperkeratosis;
- Impaired neutrophil chemotaxis;
- Defective immune function;
- Prepubertalen aggressive periodontitis;
- MO: A. actinomycetemcomitans; P. gingivalis, F. nucleatum, E. corrodens.

Obligatory for the exam
“Papillon – Lefevre” syndrome

The defective gene is localized to 11q, 14 - 21 chromosomes;

There are genes encoding MMPs, whose disfunction explains hyperkeratosis, acanthosis and proliferation of cells in the basal epithelial layer;

Defects in MMPs are responsible for aggressive parodontitis.

Mutation of the gene encoding the enzyme Cathepsin - inflammatory protease,

breaks down protein and cellular matrix components in inflammation.
Papillon-Lefevre syndrome is an autosomal recessive genetic disorder caused by a deficiency in cathepsin C.
Palmoplantar keratoderma
Melkerson Rosenthal Syndrome

- Neurological disease (genetic);
- Facial paralysis;
- Facial edema;
- Edema of the lips;
- Fisural glossitis
- Associated with orofacial granulomatosis and Crohn's disease.
The syndrome is characterized by: cheilitis granulomatosa, facial paralysis, and Lingua plicata.
Melkerson Rosenthal Syndrome

Sag palate in the midline.
Sotos syndrome

- Cerebral gigantism;
- Rapid growth in 2-3 years;
- Hypertelorism (big between eyes distance);
- Genetic disorder;
- Mental retardation, muscular hypotonia;
Sotos syndrome

4 year old child with Sotos syndrome;

Advanced bone maturation of early childhood;

Mental retardation;

Unusual craniofacial form.
SOTOS-syndrome

The same boy of 17 years;

Macrognatia

Strong growth of the mandible;

High palate;

Premature tooth eruption.
Sotos syndrome

High palate of the same patient.
Sturge-Weber syndrome

Angioma is along the trigeminal nerve;

Accompanied by edema and hypertrophy.

Embriopatia in external and mesodermal development.

Neuro-ectodermal disease

angioma;
The same child

- Side if the angioma has a slightly palatal swelling.
Trichlo - rhino – pharyngeal syndrome

Langer-Giedion syndrome

- Autosomal-dominant genetic disorder;
- Thin hair;
- Bullous nose;
- Growth retardation;
- Mild hypoplasia;
- Outstretched ears and hypodontia.
The same child

Typical signs of tricho-rhino-pharyngeal syndrome.
The same child

A child of five years;

Hypodontia;

Missing two upper second temporary incisors.
Congenital defects in connective tissue

Osteogenesis imperfecta
Marfan’s Syndrome
Ehlers – Danlos syndrome
others

Obligatory for the exam 107-121sl.!!
Osteogenesis imperfecta

A genetic disease that is characterized by easy bone fragility (often broken or cracked for no apparent reason);

Osteogenesis imperfecta is caused by a genetic defect that affects the production of collagen (the most common type 1) in the body;

In the human body it is produced in an amount of less than normal, or in a worse quality.
There are four main types of osteogenesis imperfecta:

- Type I - mild
- Type II - severe
- Type III - very severe
- Type IV - indefinite
Osteogenesis imperfecta

- Disease associated with a defect in collagen-type one;
- Bring to thin and brittle bones worst injuries;
- Progressive hearing loss
- Dentinogenesis imperfecta occur
Dentinogenesis in children with osteogenesis imperfecta

- Teeth with amber transparency;
- Quickly abrasion occur;
- Pulp obliteration;
- Scalloped enamel-dentin border;
  Slim mantel dentin;
- Irregular, atubularen dentin;
  Large interglobular dentin.
Dentinogenesis imperfecta

Temporary dentition is affected

The permanent teeth is often unaffected.
Osteogenesis imperfecta may be combined with “Odontodysplasia” in 2 year old children; Teeth shadows, typical defect in this disease.
Osteopetrosis - disease of Albers-Schonberg

- Genetic bone disease
- The balance between bone resorption and apposition is disturbed;
- Autosomal dominant and recessive inheritance;
- Delayed skeletal development, ossification of the joints occur;
- Reduced bone marrow spaces:
  - Mielopoetic pancytopenia;
  - Anemia;
  - Hemorrhagic diathese;
- Risk of infection (<Leu);
- Early mortality.

Hyper cementosis and loss of periodontal fibres
CHERUBISM - like fibrous dysplasia:

- SH3BP2 gene mutation of the fourth chromosome;
- Bilaterally asymmetric expansion of the upper jaw and directing the regard up at the sky.
- Clinically established about 5 years.
- The facies similar to the "angelic face" (painted in the Middle Ages);
- The disease is self-limiting, it can regress.

Lack of dental germs or displaced teeth

multinuclear giant cells in fibrous stroma
FLORID OSSEOUS DYSPLASIA (FOD) - "Chronic diffuse sclerotic osteomyelitis"

- Multiple sclerosis cementiod endostosis of bone tissue in the jaw, also called: "Giant cementomas,"

- Around the roots of cement-like tissue are formed.
FIBROUS DYSPLASIA - may be associated with dentinogenesis imperfecta

The latest genetic discovery - a mutation of a gene encoding a G-protein that involves in cell signalisation;

Fibroblast proliferation substitute bone marrow;

osteoblast cells metaplasia occur;

Tumor-like growths is observed.
“Fibrose dentinal dysplasia”

- Autosomal dominant inheritance;
- No tendency to abrasion; Unchanged form and color of dentin;
- Obliteration of the pulp chambers;
- Fibrous pulp dystrophy;
- Irregular dentin;
- Large bundles of collagen, calcifications.
Fibrose dentinal dysplasia

Visible obliteration of dental pulp;

It is not complete root formation;
Hurler syndrome or mucopolysaharoidosis

- Deficiency of an enzyme that breaks down mucopolysaccharides in cell lysosomes;
- Coarse facial features;
- Saddle nose;
- Macroglossia;
- Mental retardation;
- Heart problems.
Hurler Syndrome
Mucopolysaccharidosis I-H

Autosomal recessive inheritance;

Oral symptoms:
• Tremas, macroglossia, conical teeth;
• Open bite; ectopic molars;
• Retarded teeth eruption;
• Tooth cysts;

General symptoms
• Apnea;
• Macrocephaly;
• Hepatosplenomegaly;
• Deformity of the chest and spine.
Herler syndrome
Maroteaux-Lamy syndrome - accumulation of acid mucopolysaccharides in the tissues
Hypophosphatasia - mineralization defects

Hereditary (autosomal - recessive) enzyme deficiency in serum and tissue alkaline phosphatase. Rare disease than adults have neonatal and child (2-3g.) form.
Oral manifestation in Hypophosphatasia

- Periodontal lysis (localized aggressive prepubertal periodontitis);
- Loss of temporary, and permanent teeth;
- X-ray observed alveolar destruction;
- Large pulp chambers and wide roots;
- Skeletal abnormalities, eye defects, increased intracranial pressure, impaired development.
Do not differ from the peritubular and inter tubular dentin;

Hypoplastic canines;

Thin dentin;

lack of cement.
Gardner syndrome – of the group of familial colon - rectal polyposis syndromes

Autosomal dominant genetic transmitted diseases;

Oral manifestations:
- multiple osteomas, exostoses;
- fibroid tumors of the oral mucosa;

Skin and bone abnormalities:
- epidermoid cysts, fibroids and pigmentated, osteomas;
- Multiple polypus of the colon and rectum with a tendency to malignancy.
Syndrom Bloch –Sulzberger (Incontinentia Pigmenti)

- Dominant transmission, X-chromosomal genetic disorder.
- Neurological and skeletal problems;
- Pigmentations;
- Dental anomalies:
  - 90% oligodontia, retarded teeth eruption,
  - Enamel dysplasia, conical teeth;
  - Affecting both temporary and permanent teeth.
Ehlers-Danlos syndrome

Autosomal dominant / recessive genetic disorder,

affecting collagen joints, skin, mucous membranes;

Fragile skin and mucosa, bleeding lesions;

The VIII type is associated with aggressive periodontitis with early onset.
Marfan’s syndrome

Connective tissue genetic disorder;

Typical hands;

Hypermotility of joints;

Cardiac abnormalities;

Oral symptoms:
  • Caries
  • Enamel and dentin dysplasia
  • root anomalies
The end