Anomalies and features of oral facial region could be divided into two groups from genetic point of view. There are monogenic and polygenic, resp. multifactorial, units determined. Expression polygenic is not used absolutely correct. Being researched on molecular level, many polygenic anomalies desintegrate into several units, which are determined monogenic.

**Monogenic determined** are firstly very rare syndromes with oral facial manifestation, secondly some anomalies of enamel and dentin. Responsible gene mutation have been detected in many cases.

From hundreds of such **syndromes** I mention two only. Crouson sy – dysostosis craniofacialis, AD transmission, premature obliteration of cranial sutures, midface hypoplasia. Pierre-Robin sy – mandibular hypoplasia, there is possible respiratory airways obstruction after delivery. Families with affected siblings were described.

**Dentin anomalies** – dentinogenesis imperfecta – typ II (dentinogenesis imperfecta type I is connected with osteogenesis imperfecta, where the whole organism is affected). Defect of dentinogenesis, dentin is soft, brown and yellow discolored. Teeth with hypoplastic roots, mushroom shaped, pulp chamber is obliterated- no endodontic treatment is possible. Ostitic focal frequent. Frequency 1: 8 000.

**Enamel anomalies** – amelogenesis imperfecta, 1: 14000, several types with different heredity. Most frequent type is AD with hypomineralisation. Enamel matrix is normally created, but the mineralisation is insufficient. The enamel is soft, peels off. Teeth are yellow-brown. Open bite is common. Reduction of pulp chamber occurs relatively early, so the reconstruction with crowns could be provided before the age of 15.

Condition, where the process of mineralization is affected only partly, is named the form with hypomaturation.

Types with hypoplasia are characterised with insufficient production of the enamel matrix. The enamel is hard enough, but very thin. XD type with lyonising efect is most interesting. Enamel of hemizygotic male is very thin and yellow-brown. Heterozygotic female has different strips of affected and unaffected enamel – it depends which of X chromosomes (with normal or with affected allele) was inactivated during development.

Van der Woude syndrome (lower lip pits + cleft palate) is another example of AD heredity.
Polygenic, resp. multifactorial heredity

Into this group are counted all nonsyndromic cases of cleft lip +/- or cleft palate uni- or bilateral. Several genes from this group has been known (OFC1 – OFC6, MSX1, IRFG/).

There is different genetic determination of the isolated cleft palate and the cleft lip with or without cleft palate

Disorders of the number and the eruption of teeth – there is probably correlation between these entities. Most frequent is hypodontia (congenitaly missing teeth). The expression anodontia could be used for one missing tooth. The oligodontia means condition, where more teeth from several morphological classes are absent.

With hypodontia is affected 5 – 6 % of our population. Women are affected more frequently. Most frequently are missing second lower premolars and upper lateral incisors, than second upper premolars and lower central incisors. There are often combinations of missing teeth of different morphological classes [in one person or in family].

The type of the missing tooth is possible explain with help of Butler s theory of developmental fields. The dentition is divided into several fields – for each morphological class one. In the centre of each field is the anchor tooth, which is very stable and resistend to developmental changes (central incisor, canine, first premolar, first molar). The probability of numerical and morphological changes increases with distance from the anchor tooth.

Hypodontia attracted considerable attention of genetists. This is anomaly of clinical importance, but main reason is, that hypodontia is ideal genetic model, because of its relatively high frequency and easy detectability. Hypodontia is probably determined by several strong genes. Up to now several of them have been detected (PAX 9, MSX 1).

Severe oligodontia is part of ectodermal anhidrotic dysplasia, together with aplasia of sweat glands + hypotrichosis, most often XD. Hypodontia or oligodontia could be component of many others syndromes.

Hyperodontia - supernumerary teeth - is in some families connected with hypodontia.

Familiar with hypodontia and hyperodontia is retention – affected tooth is not able to erupt in correct time. Most frequent is retention of third molars, than upper canines (about 3% of population). There is no sex difference. The model of polygenic transmission is suitable for this anomaly. The heritability is 71%.

Morphological anomalies

Microdontia and peg shaped teeth occur most frequently at upper lateral incisors region, they could be connected with hypodontia and they are supposed to be microforms of hypodontia. The polygenic transmission is presumptiv. One of morphological anomalies is dens invaginatus. Endodontic treatment in these patients could be very difficult, in some cases impossible

Supernumerary tubercle (tub. Carabelli) and shovel-shaped central upper incisors are more frequent in populations with high frequency of hypodontia (e.g. American Indians). It is supposed that it could be developmental compensation. Heredity is not clear, but some familial cases were described.
Anomalies of jaws and occlusion


Mandibular progeny – excessive grow of lower jaw, sometimes small upper jaw. 3 – 9 % of population affected. Families with AD transmission were described. Most popular example is Hapsburg family. Explanation for AD transmission of this anomaly could be frequent consanguineous marriages which have kept polygenes together. Population screening discovered a lot of families with solitary cases of hypodontia. This fact supports theory of polygenic transmission of mandibular progeny.

The knowledge of genetic determination of orthodontic anomaly could influence our therapeutic approach. Anomalies with strong genetic background could be difficult to treat.