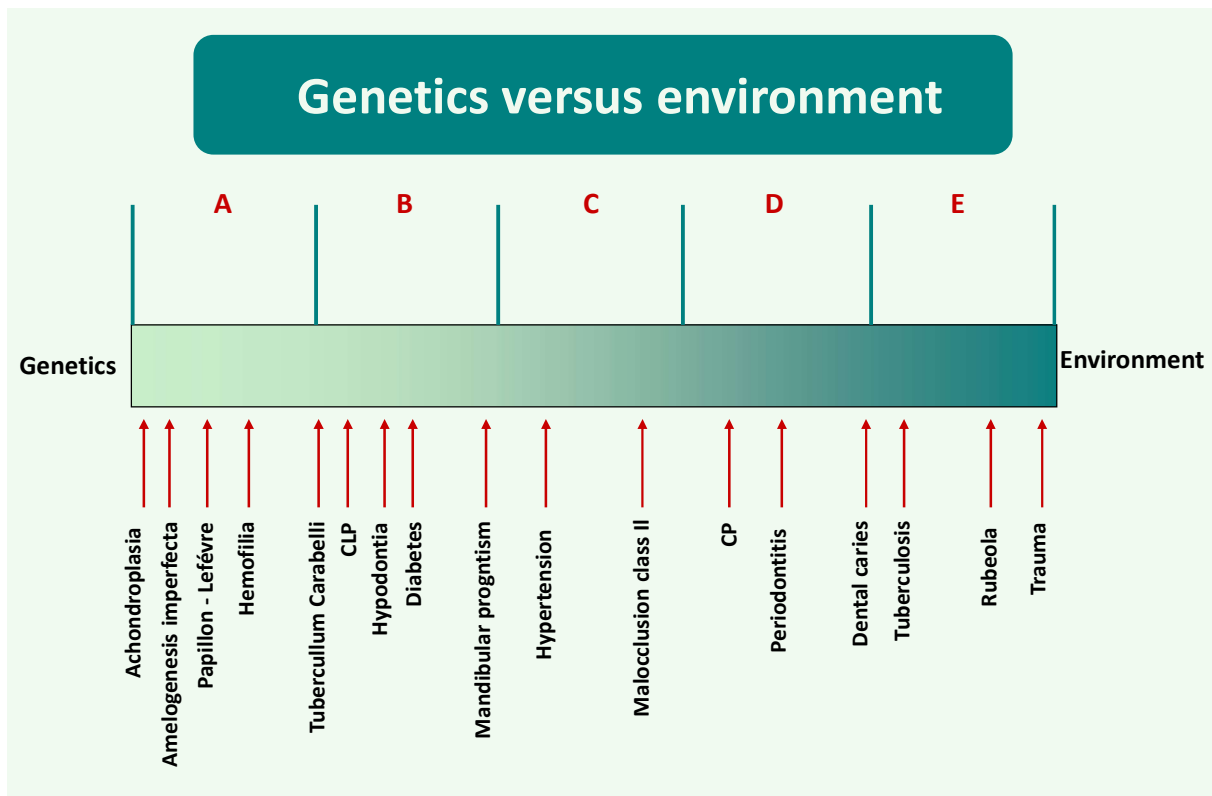


Heredity of orthodontic anomalies

Orthodontic anomalies represent large group of clinical units including number, shape and position of teeth, size, shape and position of jaws, relationship of jaws to each other and relationship of jaws to cranial base.

Genetics and environment



In this figure you see, that orthodontic anomalies are located in centre of the axis genetics x environment. It means, that those units are polygenetically determined, influenced by both genes and environment.

In 2015 celebrated American Orthodontic Association the milestone anniversary - 100 years. One of the leading american orthodontists David S. Carlson elaborated survey to show the changing opinion on this nature versus nurture dilemma.

Evolving concepts of heredity and genetics in orthodontics

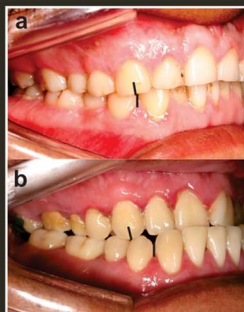
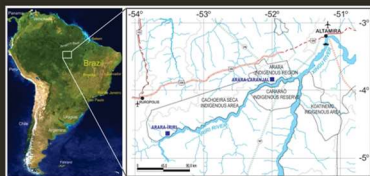


David S. Carlson
Dallas, Tex

During decades had changed substantially the number of articles focused on genetics. The first hiatus of publications in the 1960s suggests that by the middle of the 20th century the pendulum of opinion in the orthodontic community had swung back toward the view that contemporary concepts from genetics had questionable value for the practice of orthodontics. Then there was a second hiatus (1985-1990) during which no substantive articles about heredity and genetics appeared in the *AJO-DO*.

The reasons behind this second hiatus in research and publication undoubtedly were similar in general to those for the hiatus during the 1960s:

The debate continues to recent days. Very interesting article, published in internet journal PLoS ONE supports genetic origin of malocclusion. The authors compare two indigenous populations living isolated in Amazonia, which has been 90 years ago. In original village most inhabitants had perfect occlusion. In second one, which was founded by pair of people outcasted from original village were many cases with mandibular prognatism. The authors suggest, that this is effect of the founder.



Nomando D, Faber J, Guerreiro JF, Quintao CC. Dental Occlusion in a Split Amazon Indigenous Population: Genetics Prevails over Environment. *PLoS ONE*, 2011, 6(12), e28387.

Our own research also tried to elucidate the role of genetics and environment in orofacial anomalies. Skulls of three cohorts were measured:

1. Old 9. century material from burial ground near Brno
2. 10. – 15. cent. skulls from burial ground Ducové (Slovakia)
3. 14.- 18. cent. from ossuary Mělník

Results from these measurements were compared with head films and cephalometric measurements of recent population (university students from Prague)



We found, that our skulls and faces have changed during centuries. Old populations were dolichocephalic and we are brachycephalic mostly, also our faces were wider than those of historical population.

But, what is surprising, in our wide faces and skulls are narrow and constricted dental arches. Also the amount of serious anomalies as open bite or crossbite is significantly higher in recent population.

What it means? This research emphasises the role of environment, respectively function. It is probable, that food in history was harder and more abrasive than today.

So, the conclusion is that both factors - environment and genetics are responsible for orthodontic anomalies and that multifactorial determination is highly probable.

Genetics of dental anomalies

Dental anomalies are numeric, morphologic or anomalies of eruption. Both dentitions could be affected, however, dental anomalies are more frequent in the permanent one.

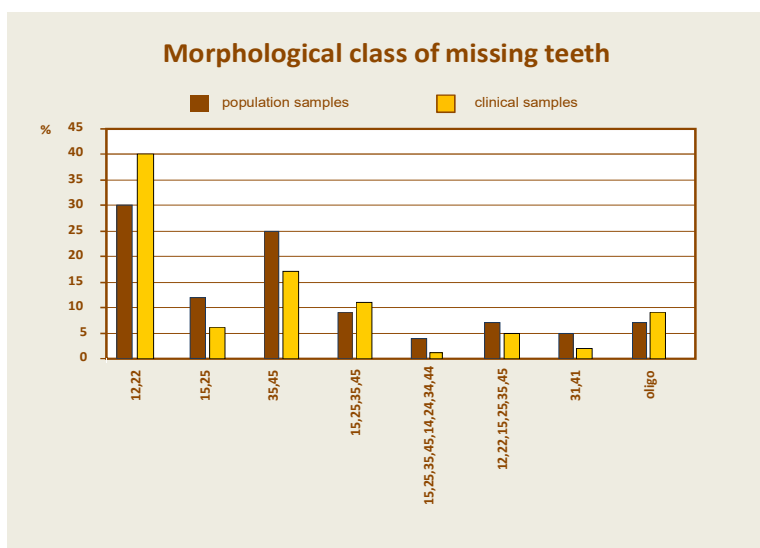
Most common is **hypodontia** – condition, in which one or more teeth are congenitally missing. The term oligodontia implies agenesis of numerous teeth and is commonly associated with systemic abnormalities.

The prevalence of hypodontia of permanent teeth differs in races. For Central European population the agenesis of teeth, other than of the third molar, is about 6 %. (Hypodontia of third molars is more than 20 %). Most common is hypodontia in American Indians, relatively rare in Africa. The girls are affected more often (sex ratio 1: 1,7)

From genetic aspect is hypodontia ideal condition. Is relatively common and easy detectable, so it explains the inflation of studies concerning this topic.

But there are also serious associations. Mutation of genes responsible for tooth development were detected in families with colorectal carcinoma. Hypodontia is often in group of patients with ovarian carcinoma.

Frequency of hypodontia depends on type of the tooth. Most common is hypodontia of second lower premolars, then upper lateral incisors and upper second premolars. On the other hand very rare is hypodontia of canines or central incisors. Hypodontia of one central incisor could special case- the mild manifestation of holoprosencephalic spectrum . One form is caused by Sonic hedgehog mutation.



This figure shows frequency in Czech population according to morphological class and comparison between population and clinical sample. The high number of lateral incisor hypodontia is influenced by esthetic reasons. More people (and especially girls) ask for treatment.

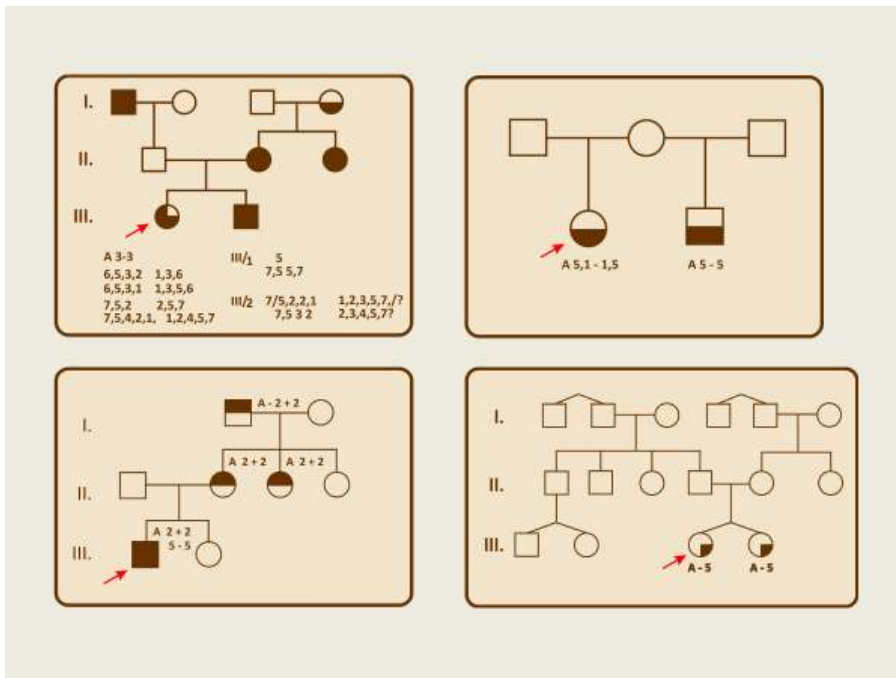
The hereditary component in the determination of such an anomaly is undoubtedly multifactorial. The model with several major genes and environmental background is suggested.

The multifactorial determination is supported by results of twin studies : 55% cocordance in MZ twins and 10 % in DZ twins.

With the advent of genomic research, a lot of studies have appeared. As candidates genes were proved, for example, MSX1, PAX 9,EDA and others. The PAX9 mutation is connected with a relatively rare hypodontia of molar region.

Recently also genes IRF6, BMP4, WNT10A and AXIN2 are mentioned.


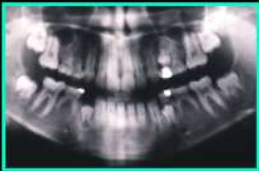
In the following picture you can see several pedigrees from our hypodontia research project.

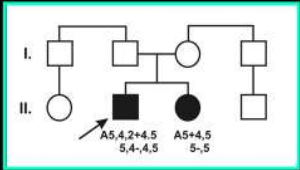


In the next figure is family with hypodontia. The first diagnosed person was brother a later was proved suspicion of hypodontia of his sister.

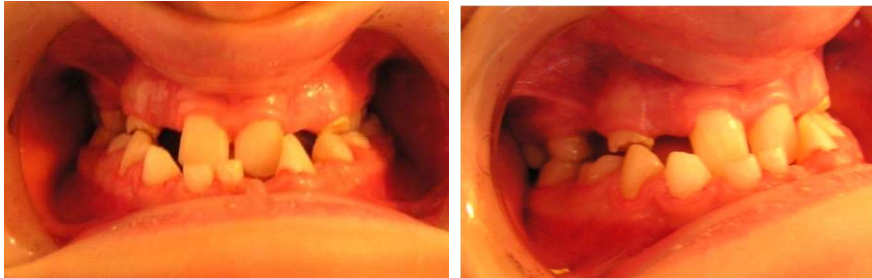
Hypodontia

- MSX1 -premolars + lateral incisors
- PAX9 -oligodontia, lower incisors + molars

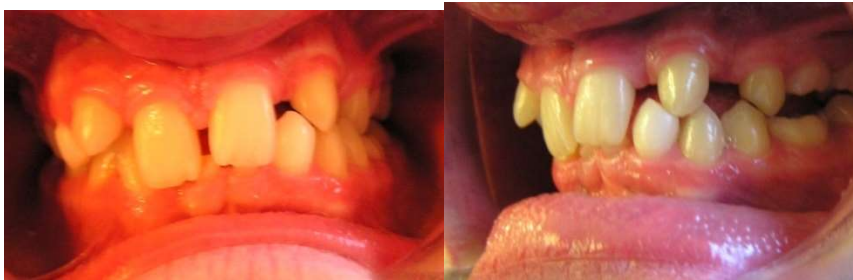





And clinical picture of oligodontia girl (only central incisors and molars are permanent)



Another case of oligodontia



Hyperodontia and morphological changes of teeth (microdontia and pegshaped teeth) occurs often in one family or in one person together as a part of DAP (dental anomaly pattern).

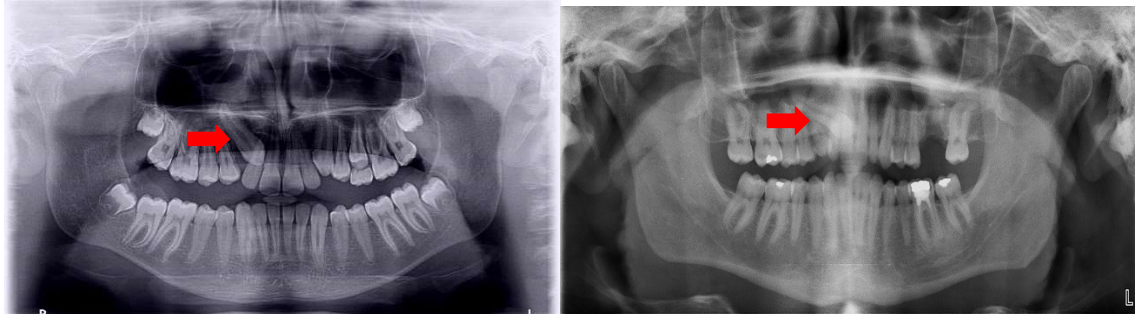
From eruption disorders are clinically important impacted canines. In most cases it is **palatally displaced upper canine (PDC)**. This anomaly is not so frequent as hypodontia - cca 2% - 3% of population.

Boys are affected slightly more frequent than girls, the heritability is from 65 to 71%.

In next pictures are impacted canines in two brothers.

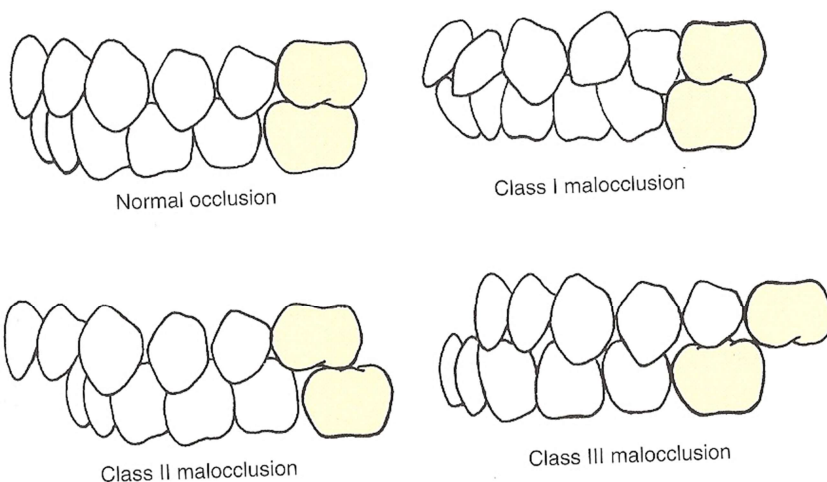


In the next picture is father and son with PDC



Genetics of malocclusion

To understand this chapter, the knowledge of basic orthodontic classification – classes according Angle- is needed.



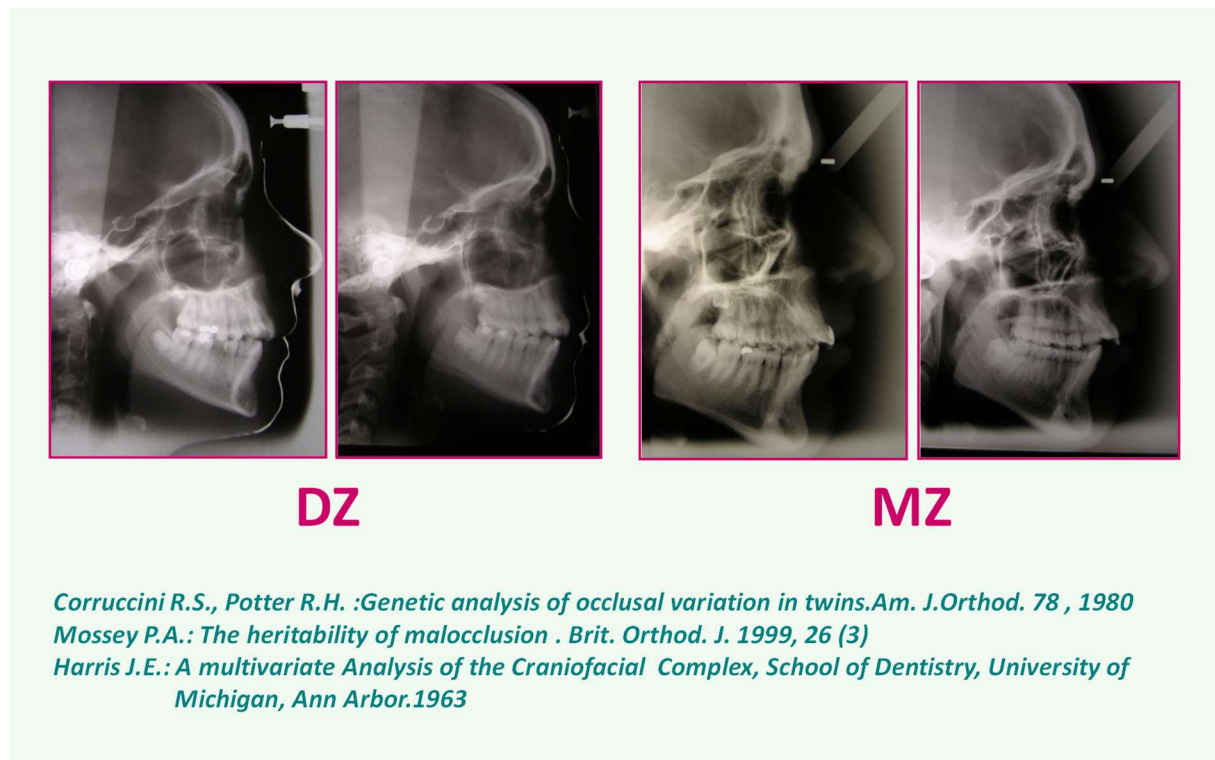
Substantial is relationship of first molars, respectively mesiobuccal tuberculum. Class I is harmonic relationship of both jaws, but can be connected with crooked teeth. Class II means distocclusion – distal position of lower jaw or smaller lower jaw. Class III – mesioocclusion is mainly bigger mandibula , sometimes smaller maxilla.

Basic features in face , especially the central part formation are given genetically. It is obvious, that substantial part of cases is multifactorially determined.

Class II malocclusion

Clinical features are protrusive upper teeth a smaller lower jaw. This malocclusion is extremely frequent in our population more than 20%.

Family and twin studies has proved hereditary determination, part of cases is caused by malhabits as mouth breathing or thumb sucking. In the next picture are rentgenograms



In the next figure is comparison of affected members of families with class II and affected members from population

	Population	Class II
Affected fathers	4,1%	18,8%
Affected mothers	16,5%	29,9%
Affected parents	11,5%	25,3%
Affectwd siblings	13,6%	48,1%

Markova M,1985