Congenital defects of dentin and enamel

All clinical units mentioned in this paragraph are caused by mutation of single gene. They are transmitted in typical mendelian way. According to the type of disorder originate in growth stages of histodifferentiation, apposition and calcification.

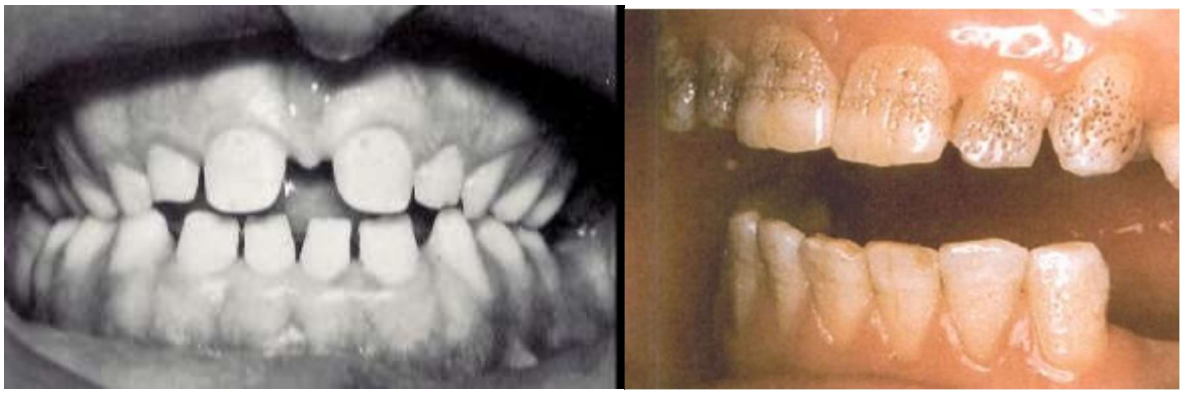
Amelogenesis imperfecta consists of several clinical units with different types of mendelian transmission. All types of such a condition occur in population in a ratio of about 1 : 14 000. The most common type is the autosomal dominantly inherited hypocalcified form, which occurs in ratio of about 1: 20 000.

Enamel formation may be divided into three processes: (1) formation and secretion of an organic matrix (histodifferentiation), (2) mineralization of the matrix (3) maturation of enamel.

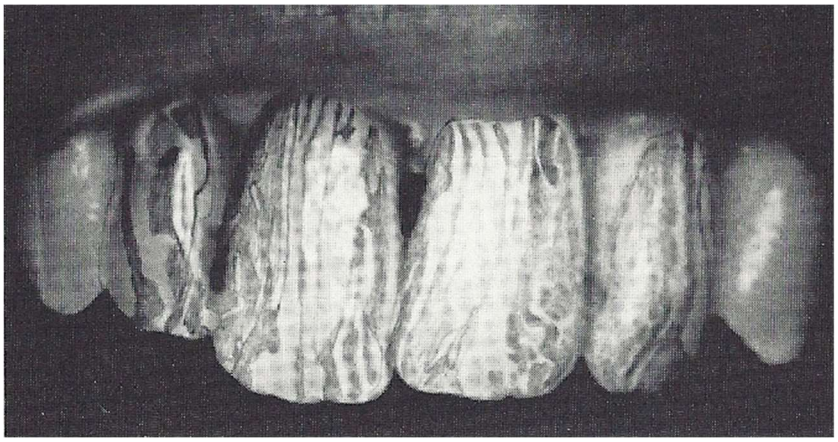
More than 20 genes are, so far, known to be responsible for this condition.

Hypoplastic forms of AI originate in stage of histodifferentiation and are caused by heterozygous mutation in the enamelin gene (*ENAM*) Enamel layer in these forms is reduced, with colour changes. The tooth look like being prepared for crowns. Because the unit is heterogenous, both types (autosomal dominant and recessive) were described.

In the left picture is AR AI, the right one AD.



X chromosomal form (*AMELX*) is interesting from genetic point of view. The enamel of heterozygous women consists of vertical bands of normal and hypoplastic or hypomature tissue (Lyonization effect – it means that during development one X chromosome of pair is not active. According to inactivation of normal or mutant one originate strips of hypoplastic and normal thickness enamel). The next picture of X transmitted hypomaturation is from Stewart´s and Prescott´s Oral facial genetics. To see the strips , the special lighting was needed.

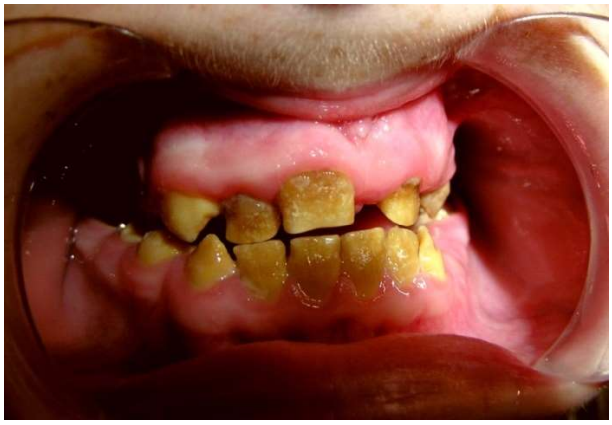


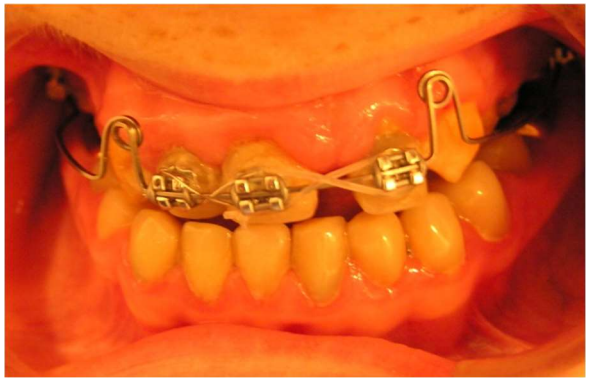
Occasionally could AI occurred as a part of syndromes, for example amelo-onycho-hypohidrotic, tricho- dento- osseous and together with nephrocalcinosis (gene *FAM20A)*.

The most frequent (1: 20 000) and the most serious is AD hypocalcified form.

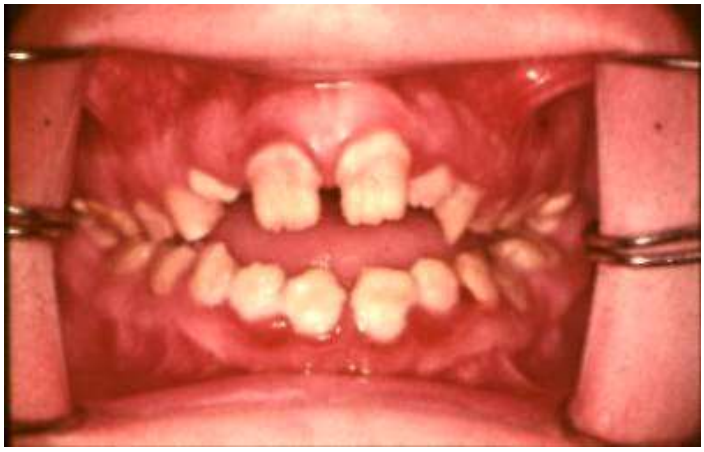
The anomaly is characterized by enamel of normal thickness on newly erupted and unerupted and unresolved teeth. The enamel is soft and may be lost soon after eruption, leaving the crown composed only of dentin. The enamel has a cheesy consistency and can be scraped from the dentin. An anterior open bite has been recorded in over 60% of the cases observed.
This type is caused by heterozygous mutation in the *FAM83H* gene.

In next two pictures is one of my orthodontic patients with hypomineralised AI. The left central incisor was lost because of complications of AI. In the next picture is restoration with crowns. Fortunately these children could be treated prosthetically very early. Permanent irritation of the pulp caused its reduction.

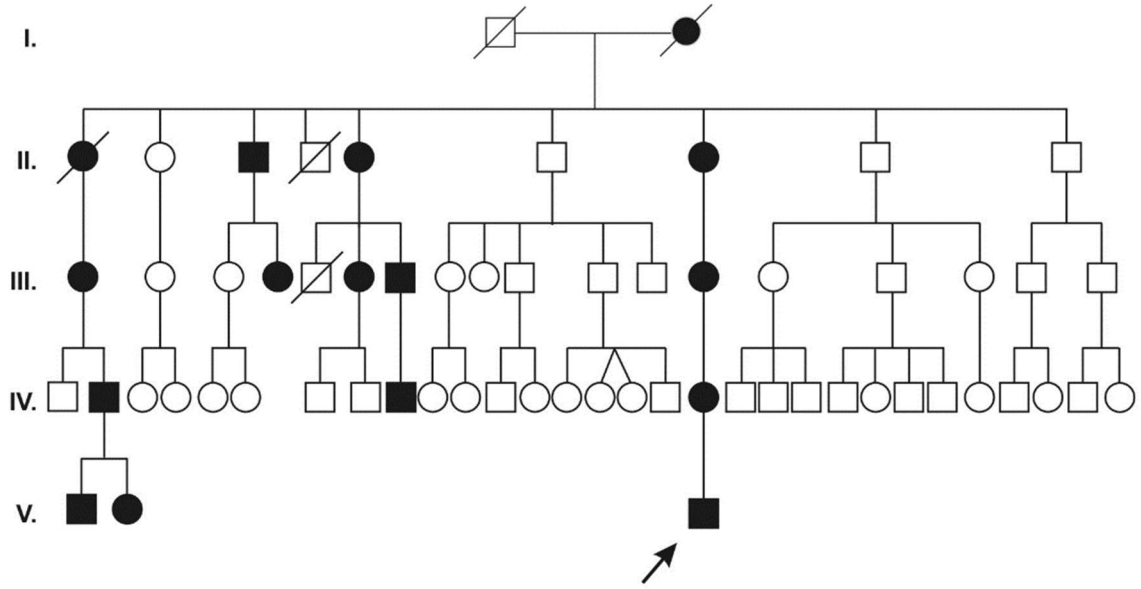




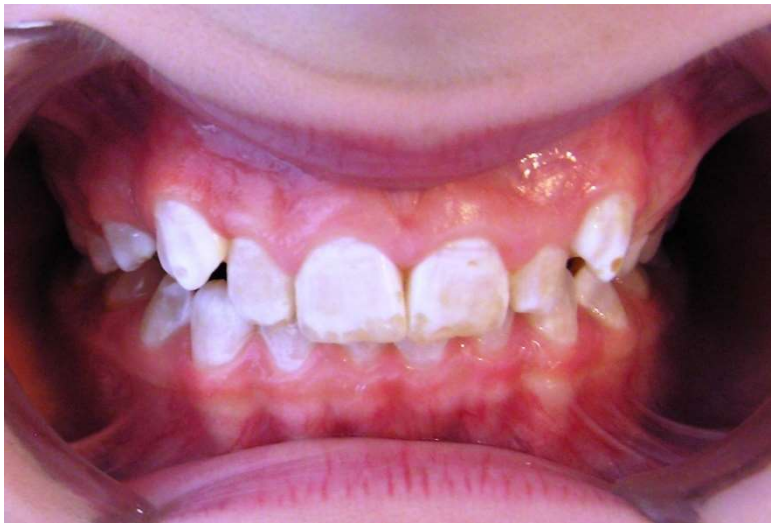
In the next picture is AI patient with typical open bite



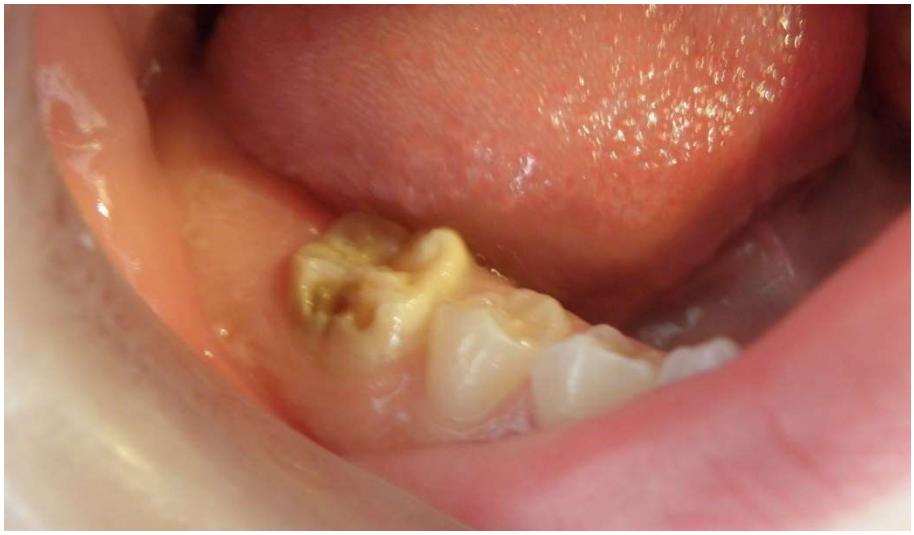
Large pedigree in the next picture demonstrates the AD transmission of this anomaly. Notice, that healthy members have children without AI, the affected members have in average half of children with AI.



Not all hard dental tissue disorders are of genetic origin. Most of them are caused by external influence. The clinical picture of the next patient is more probable due to fluorosis.



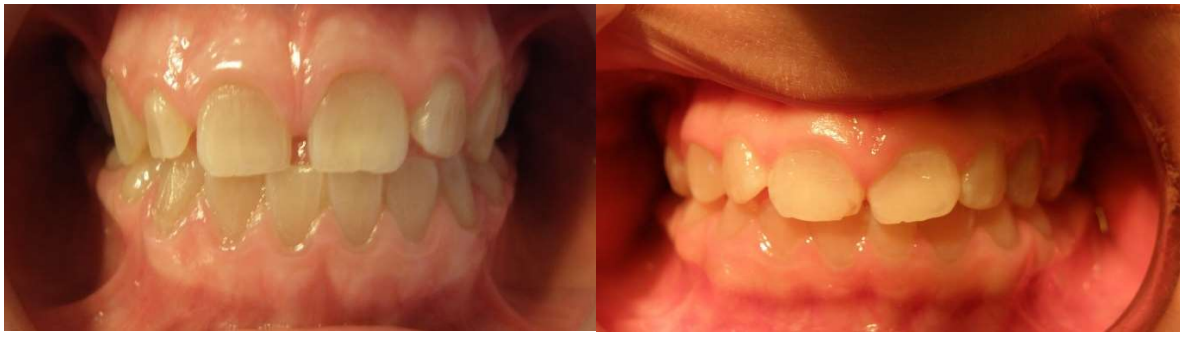
Of recent years, a condition referred to as Molar-Incisor Hypomineralisation (MIH) has become prominent .The aetiology of this condition, which affects one or more of the first permanent molars in a chronologically-reminiscent but unrelated pattern, together with a seemingly random number of permanent anterior teeth, is unknown. The influence of feverish disease during tooth development is mentioned.



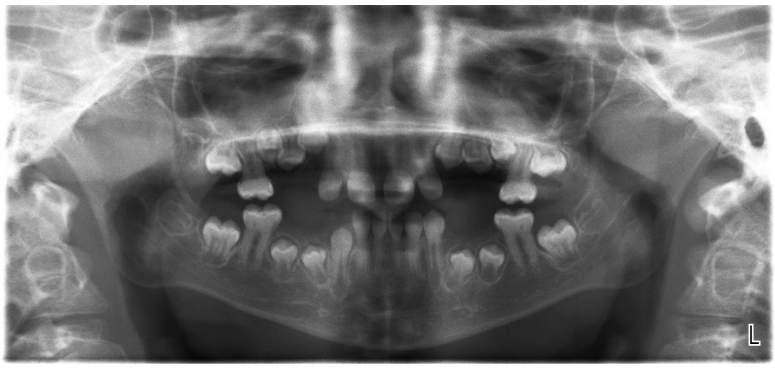
***Dentinogenesis imperfecta* -** despite being relatively rare, this condition is most often among hereditary anomalies of hard dental tissues (1 : 6 000 -8 000). It is touching the development of mesenchymal dental tissues (dentin, pulp, cementum and periodontium). It originates during histodifferentiation.

The teeth are blue-grey or amber brown and opalescent. On dental radiographs, the teeth have bulbous crowns, mushroom shape, roots that are narrower than normal, and pulp chambers and root canals that are smaller than normally or completely obliterated.

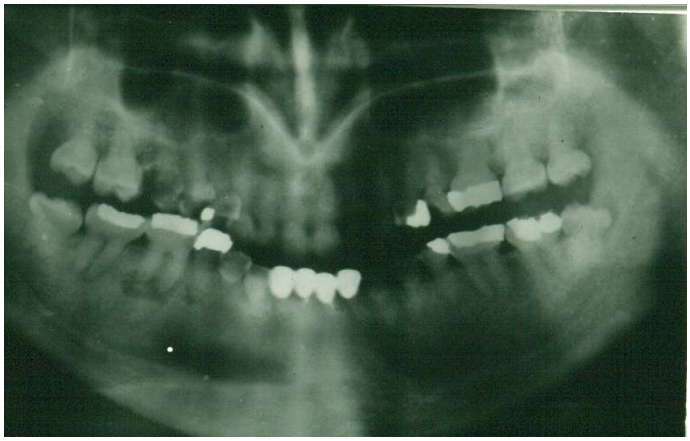
In the next picture are dizygotic twins boy and girl with dentinogenesis imperfecta . Their mother was also affected.



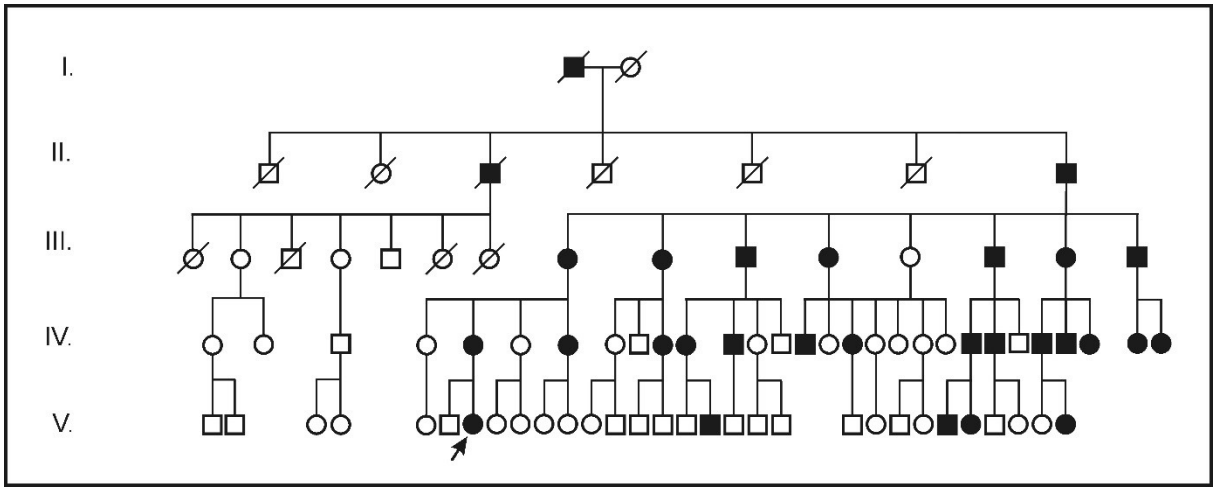
Typical X-ray changes (mushroom shape, pulp obliteration) are visible in the next picture (10 years child)



And adult patient with complication (restorations, granuloma…)



The AD transmission was proved. The condition is caused by mutation in the *DSPP* gene encoding dentin phosphoprotein and dentin sialoprotein. The attached pedigree collected by Racek and Sottner clearly demonstrated this type of transmission.



Dentinogenesis imperfecta also accompanies some cases of more severe disorder - osteogenesis imperfecta. Caused mainly by mutation of *COL1A1, COL1A2* genes, characterised by bone fragility and blue sclerae.