There are many acquired and inherited developmental abnormalities that alter the size, shape and number of teeth. Individ-
ually, they are rare but collectively they form a body of knowledge with which all dentists should be familiar. The discus-
sion of each condition is short and to the point. Comprehensive reviews of each may be found in any reasonably new text-
book of oral pathology. For those conditions that are inherited (eg: ectodermal dysplasia, dentinogenesis imperfecta and
others) go to: www3.ncbi.nlm.nih.gov/, it will bookmark as OMIM (Online Mendelian Inheritance in Man).

Supernumerary Teeth
Slide #1 is an example of an extra incisor. When located in the midline between the two permanent central incisors, they
are referred to as mesiodens. Slide #2 depicts an extra molar tooth (a paramolar) and Slide #3 is an example of a super-
numerary bicuspid tooth. These are the most common supernumerary teeth in the order shown.

Hyperodontia and Cleidocranial dysplasia
Count the teeth in Slide #4 — there are more than 50. This patient has cleidocranial dysplasia (CCD). This is inherited
as an autosomal dominant trait, the gene maps to chromosome #6. The gene encodes a protein called Core Binding Fac-
tor Alpha 1 (CBFA1). This protein is essential for the formation of a normal skeleton but its role in tooth formation is
not yet known. The heterozygous state produces the CCD phenotype, the homozygous state is lethal. Main features of
the phenotype include hyperdontia, small or missing clavicles, delayed closure of the cranial fontanelles (soft spots) and
short stature. Slide #5 shows multiple supernumerary teeth removed from a patient with CCD. Gardner syndrome (intes-
tinal polyposis and skeletal osteomas) also features supernumerary teeth but not to the extent seen in CCD. (J. Med.
Genetics 1999;36 p177-182 and Cell 89; 773-779 May 1997)

Slide #6 is a little tricky. The coin shape radiolucent lesion between the mandibular bicuspid teeth was thought to be a cyst.
It was removed and found to be a tooth germ of a developing supernumerary tooth. It was discovered early in develop-
ment before mineralization commenced. Radiographically, it resembles a lateral periodontal cyst.
Hypodontia and Oligodontia

Glossary: Anodontia: failure of teeth to develop (same as agenesis of teeth)
Hypodontia: having less than 6 congenitally missing teeth.
(Oligodontia: having 6 or more congenitally missing teeth.
Hyperdontia: extra teeth, same as supernumerary teeth, may be single or multiple as in CCD.

(*To see how the web site OMIM works, log on and enter hypodontia.)

Congenitally missing teeth (hypodontia and oligodontia) are not rare. Generations of dental students have learned about ectodermal dysplasia, the best know of the “missing teeth” syndromes. With the discovery of the Pax 9 gene, some light has been shed on the molecular genetics of congenitally missing teeth. Pax 9 maps to chromosome #14, it encodes a transcription factor that functions in the development of derivatives of the pharyngeal pouches. Mice with Pax 9 mutations have “craniofacial and limb anomalies and teeth fail to form beyond the bud stage”. A family with a framshift mutation in Pax 9 had normal deciduous teeth but lacked permanent molars in both the maxilla and mandible. It is transmitted as a dominant trait. (Nature Genetics 24;18-19 2000 Jan , also European Journal of Oral Sciences 106; 38-43 1998)

The best known of the missing teeth syndromes is X-linked hypohidrotic ectodermal dysplasia. Ectodermal derivatives such as hair, sweat glands, nails and teeth are involved. Head hair, eye lashes and brows are sparse, nails are dystrophic and there is marked oligodontia, rarely total anodontia. Diminished sweat glands leads to the inability to regulate body temperature, a major disability in warm months in a hot climate. According to your text (Neville’s Oral & Maxillofacial Pathology 2nd ed), there are 150 variants of this syndrome. Dominant, recessive and X-linked inheritance have been reported.

Slide #7 shows the mildest form of hypodontia, a single congenitally missing tooth, in this case a bicuspid. Notice the deciduous molar has been retained. Slide #8 illustrates the other extreme, a 10 year old child missing virtually all permanent teeth except 1st molars. Slides #9 and #10 are of a child with ectodermal dysplasia. There is profound oligodontia and teeth that are present are cone shaped. Note the sparse scalp hair, brows and lashes. (Remember X-linked disease are milder in females because they enjoy partial protection thanks to lyonization.)

We will exit the subject of hypodontia/oligodontia by looking at Slide #11, an example of a cone-shaped lateral incisor, a peg lateral, a form of microdontia. This may be inherited as a dominant trait. If both parents have “peg laterals”, the homozygous child will have total anodontia of succedaneous teeth. (Am. J. or Med. Genetics 26:431-436 1987)
Taurodontism

A morphologic abnormality of teeth called taurodontism (bull teeth) is seen in slide #12. Slide #13 is a normal cynodont for comparison. Slide #14 is also a taurodont. This condition is most conspicuous in the molar teeth. The distance from the roof of the pulp chamber to the root bifurcation is greatly increased. Variants include hypotaurodontism, mesotaurodontism and hypertaurodontism. Cases illustrated here are hypertaurodents. This conditions may exist as an isolated trait (autosomal dominant) or as part of several syndromes including the trichodentoosseous syndrome (TDO), otodental dysplasia, ectodermal dysplasia, tooth and nail syndrome, amelogenesis imperfecta and others. (Am. J. Med. Genetics 11: 435-442 1982 April). Visit OMIM for more.

Fusion, Gemination, Dilaceration & Concrescence

These conditions are illustrated in Slides #15 through #27 and require little comment.

**Fusion**, the joining of two adjacent tooth germs to form a single large tooth is seen in Slide #15. Notice the lateral incisor is missing, it fused with the central incisor.

**Gemination**, an attempt by a single tooth germ to form two teeth…twinning, seen in Slides #16 and #17.* It is not always readily apparent if a large tooth is an example of fusion or germination. If you have a problem, just count the teeth. If one is missing it is fusion, if one is not missing, it is germination.

Slide #18, **dilaceration**, an abnormal deviation (bend) in the root of a tooth, presumably caused by traumatic displacement during root development.

Slide # 19 is an example of **concrescence**, the fusion of cementum of adjacent teeth, a good reason to have radiographs before extraction of a tooth.
Hypercementosis, screwdriver incisors and mulberry molars:

Hypercementosis is seen in Slides #20 and #21. Note the thick mantle of cementum that makes the root look fat. (a national board pearl: If the jaws are involved in Paget disease of the skeleton (osteitis deformans), the teeth may show hypercementosis. But in practice, Paget disease seldom involves the jaws. Somewhere long ago in a faraway place, a case of osteitis deformans of the jaws with hypercementosis was reported and has endured in dental literature.) Slides #22 and #23 also show dental defects that have endured in dental literature but have virtually disappeared in practice. They illustrate “Screwdriver” incisors and “Mulberry molars”, dental defects seen in congenital syphilis and caused by direct invasion of tooth germs by Treponema organisms (yes, Treponema pallidum can pass through the placenta).*** Slides #22 and #23 are not UMKC slides and are not to be copied or published.

Macrodontia, Dens in Dente, and Enamelomas

Slide #24 is an example of macrodontia, note the unerupted lower 2nd bicuspid teeth are as large or larger than the molar teeth, “molarization of the bicuspsids”. Slides #25 and #26 show a developmental defect called “dens in dente”, dens invaginatus or tooth within a tooth. Invagination of the cingulum has resulted in enamel being reflected into the tooth. This is of no clinical significance except that caries may develop in the invagination and escape detection. The ectopic formation of enamel is depicted in Slide #27. Small droplets of enamel form on the root surface, so-called enamelomas or enamel pearls.
**Attrition, Erosion and Abrasion of teeth.**

**ATTRITION:** Loss of tooth surface due to normal wear. Some wearing is normal (physiologic) but accelerated wear beyond normal is pathologic.

**EROSION:** The chemical dissolution of tooth structure often attributed to regurgitation of gastric acid, excessive intake of acidic food or drink, (eg, two liters of cola/day for years)

Sometimes a cause cannot be identified, it is idiopathic.

**ABRASION:** Wear beyond normal caused by mechanical forces. This sounds like pathologic attrition but the difference is the pattern of wear. Attrition is ordinarily confined to the occlusal and incisal surfaces. Abrasion is ordinarily used when the loss is on a non-occluding surface.

**ABFRACTION:** I have avoided this because I am not sure it exists but it is in the literature and you should be familiar with the term. It is proposed that with each bite, occlusal forces causes the teeth to flex ever so little. Constant flexing causes enamel to break from the crown, usually on the buccal surface. Does this really happen? If it does, why don’t we all have it? ("Abfraction lesions: myth or reality." *Journal of Esthetic & Restorative Dentistry* 15(5):263-71, 2003)

As the following slides will show, it is not always easy to distinguish between attrition, erosion and abrasion, they may coexist. Slides #28 and #29 are of a 21-year-old man who had occlusal wear that had flattened the occlusal surfaces and loss of enamel on the buccal surfaces that cannot be explained by occlusion. Is the occlusal wear just an example of advanced attrition and the buccal lesions caused by erosion or abfraction? We could not identify a reason for erosion and he denied nocturnal bruxing or coarse diet that could account for the wear. Slide #30 shows advanced wear on the occlusal and incisal surfaces presumably due to end to end occlusion coupled with erosion. We could not identify an erosive agent but there is an almost identical picture in your text that identifies it as erosion. (Neville’s 2nd ed. Oral and Maxillofacial Pathology)

Slides #31 and #32 are two different people with what appears to be erosion but I was not able to identify a chemical or dietary habit that could account for it. Slide #33 is a little less mysterious. Notice the loss of enamel on the lingual surfaces caused by long term, daily exposure to gastric acid reflux in a patient with bulimia. And finally Slide #34 is abrasion caused by tooth brushing. Toothbrush abrasion is common but usually not to the extent seen here.
Internal and External Resorption

Slides #35–39 illustrate resorption from within, internal resorption. One or many teeth may be involved and the cause is a total mystery. (*Skeletal bone has a counterpart in which a bone or adjacent bones mysteriously disappear, so-called vanishing bone disease or Gorham’s syndrome.) Internal resorption may mimic dental caries as seen in Slide #36 (although you can’t see it, there was a thin shell of tooth structure covering the lesions). Osteoclasts (dentinoclasts?) arising in the dental pulp inexorably resorb dentin and enamel that can be stopped only by complete removal of all pulp tissue following early recognition and prompt endodontic treatment. Slide #40 shows dentin on the left lined with a row of multinucleated osteoclasts occupying Howship’s lacunae. Inflamed pulp is seen on the right.

External resorption starts on the root surface and progresses inward. Although the cause may be idiopathic, in some cases the cause is apparent. Slide #41 exhibits short roots on lower incisor teeth in a person who had orthodontic treatment, a well known cause of minor external root resorption. Slide #42 exhibits resorption of the roots of a molar tooth caused by a keratocyst (the black hole) and Slide #43 shows resorption of the roots of teeth # 30 & 31 by a tumor, an ossifying fibroma. Slides # 44–46 are examples of idiopathic external resorption. This is mysterious and frustrating to patient and dentist alike.


*Caution: when you see short teeth, it is not always external resorption. Sometimes teeth never form completely. Slide # 47 shows short roots, almost no roots in the maxillary teeth. This patient had radiation treatment for a brain tumor at age 3. The maxillary teeth were in the field of radiation and the formative tissue of the roots were irreparably damaged so root development was terminated. And sometimes roots never form and a cause cannot be found as seen in Slide #48.

Slide 35: internal resorption
Slide 36: internal resorption
Slide 37: internal resorption
Slide 38: internal resorption (arrows)
Slide 39: internal resorption, unknown cause
Slide 40: internal resorption
Slide 41: external resorption, secondary to orthodontic treatment
Slide 42: external resorption secondary to cyst
Slide 43: external resorption secondary to tumor
Slide 44: idiopathic external resorption
Slide 45: idiopathic external resorption
Slide 46: idiopathic external resorption
Slide 47: failure to form secondary to radiation injury
Slide 48: failure to form, no known cause
Dentinogenesis Imperfecta (DI), also known as opalescent dentin

This is an autosomal dominant condition affecting both deciduous and permanent teeth. Affected teeth are gray to yellow-brown and have broad crowns with constriction of the cervical area resulting in a “tulip” shape. Radiographically, the teeth appear solid, lacking pulp chambers and root canals. Enamel is easily broken leading to exposure of dentin that undergoes accelerated attrition. Slides #49–52 are examples of DI. The gene maps to chromosome #4. It encodes a protein called dentin sialophosphoprotein (DSPP). This protein constitutes about 50% of the noncollagenous component of dentin matrix. It is not known how the mutant protein causes near obliteration of the pulp. A clinically and radiographically indistinguishable dental condition is seen sometimes in patients with osteogenesis imperfecta. The following classification has been proposed.

A. Dentinogenesis imperfecta type I, with osteogenesis imperfecta
B. Dentinogenesis imperfecta type II, without osteogenesis imperfecta
C. Dentinogenesis imperfecta type III (see below)

DI type III is even more rare and paradoxically characterized by too little rather than too much dentin resulting in “shell teeth”, Slide #53. Type III DI may be an allelic variant of type II DI, (a different mutation in the same gene) both genes map to the same region on chromosome #4. (Slide #53 was loaned to us by another pathologist and is not to be copied or published. It is exclusively for the use of students at UMKC.)
Dentin Dysplasia type I & type II (DD I & DD II)
Dentin dysplasia type I is a rare dominantly inherited dental developmental abnormality that maps to the same site on chromosome #4 as does dentinogenesis imperfecta type II & type III. It too may be an allelic variant. The phenotype varies considerably from solid teeth with no roots and periapical radiolucent lesions (Slide #54) to teeth with nearly normal root length but with partial obliteration of the pulp and large pulp stones (Slide #55). The periapical abscesses are due to extension of the pulp horns nearly to the surface. Minimal occlusal wear exposes the pulp to oral bacteria.

Dentin dysplasia type II (coronal DD) is characterized by teeth of nearly normal length but as the pulp ascends to the crown, it flares into a flame shape or thistle tube shape as seen in Slides #56 and #57. (See: "Dentin Dysplasia type I, five cases in one family." Oral Surg Oral Med. Oral Path. 86:175-178 1998)

Regional Odontodysplasia (ROD, ghost teeth)
This is a sporadic (not inherited) developmental defect involving only a few teeth in a small REGION of the jaw. Slide # 58 illustrates ROD involving a lower incisor, cuspid and 1st bicuspid teeth. The dentin and enamel are thin and the pulp huge producing a ghost like appearance. Slide #59 is an example of ROD in the mandible of a 7-year-old boy. It involves five teeth: two incisors, the cuspid and both bicuspids. Slide #60 is histology of ROD, notice the huge pulp and thin rim of tooth structure, mostly dentin.
**Segmental Odontomaxillary Dysplasia (SOD or SOMD)**

Segmental Odontomaxillary Dysplasia (SOD or SOMD) is illustrated in Slides # 61 and #62, the patients are age 13 and 4 respectively. The right maxilla is involved in both cases. Unlike regional odontodysplasia which involves only teeth, segmental odontomaxillary dysplasia involves teeth and bone. The segment of the jaw that is involved is expanded due to enlargement of the bone and hyperplasia of overlying gingiva. Teeth in the involved are malformed and some may be congenitally missing. The condition is not progressive, i.e. it has a limited growth potential. Radiographs show increased density of the affected bone with a granular pattern, the maxillary sinus may be smaller than normal. Those who are not familiar with this condition may interpret the enlarged bone to be evidence of a tumor or fibrous dysplasia of bone, both of which are progressive diseases.

---

**Amelogenesis Imperfecta (AI)**

There is a large group of inherited developmental defects in enamel collectively referred to as amelogenesis imperfecta. At least 14 phenotypes have been identified and autosomal dominant, recessive and X linked inheritance have been reported. The conditions are rare, only about 1 in 14,000 have it. Establishing a pattern of inheritance requires constructing a pedigree (family tree) of several generations, identifying those with and without the condition. This is no minor task. Furthermore there is subtle overlaps in the phenotypes so that you may not be able to identify a specific type of AI when only a single or few patients are available for study. Much of what is known about AI comes from northern Sweden where the condition is more common. Three general categories of AI are recognized:

1. **Hypoplastic type**: Inadequate formation of enamel matrix; both pitting and smooth types exist. Enamel may be reduced in quantity but is of normal hardness.

2. **Hypomaturation type**: A defect in the crystal structure of enamel leads to a mottled enamel with white to brown to yellow colors.

3. **Hypocalcified type**: A defect not in the quantity but in the quality of enamel. It is poorly mineralized, soft and chips and wears easily.

Slides # 63 and #64 are examples of the hypocalcified type of AI. Slide # 65 is the smooth hypoplastic type. Slide # 66 shows the mottled appearance of the hypomaturation type.

*** Acquired (not inherited) defects in enamel may cause color and pitting defects, see your text for more information.

The matrix of enamel is comprised mainly of a protein called amelogenin. The gene for this protein is on the short (petit) arm of the X chromosome (Xp22.1). A number of mutations have been described including deletions, missense and nonsense mutations.

Autosomal dominant AI has been traced to a gene on chromosome #4 near the site as the gene for dentinogenesis imperfecta and dental dysplasia. (It appears that a region on #4 chromosome is a hot spot for dental developmental abnormalities). The AI gene at this site codes for another enamel protein, enamelin. Enamelin is thought to serve as a “nucleation site for hydroxyapatite crystals.”

See: (1) "Detection of a novel mutation in X-linked amelogenesis imperfecta"


(2.) "An amelogenin gene defect associated with human x-linked AI"


(3) "Localization of a gene for autosomal dominant AI to chromosome 4q."

*Human Molecular Genetics* 3:1621-25 1994

---
SELF-EXAM CASES

#1 What is your diagnosis of the condition in Slide #67?

Slide 67

#2 Slides # 68 and #69. Does this child have dentinogenesis imperfecta? Neither parent had discolored teeth.

Slide 68

Slide 69

#3 Slide #70 Several children in the family had mottled teeth, the parents did not. Is this an example of recessively inherited hypomaturation amelogenesis imperfecta? Note the marked similarity to HAI in Slide # 66.

Slide 70

Answers

#1 The answer is taurodontism and hypoplastic (smooth) amelogenesis imperfecta. Note the large, tall pulp chamber and the thin enamel cap on the teeth.

#2 The answer is no. This child has mottled teeth caused by taking tetracycline during the years the teeth were forming. This antibiotic is deposited in teeth and bone and darkens both. Slide # 69 is an undemineralized, thin section of a tooth viewed with fluorescent light. Each yellow band of fluorescence in the dentin marks an episode of treatment with tetracycline.

#3 Slide #70 is an example of dental fluorosis. The family moved to a farm in the Texas panhandle and drank water from a well with a fluoride content of greater than four ppm.

Charles Dunlap, DDS
September 2004