Oral and craniofacial challenges in osteogenesis imperfecta – a clinical overview

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Osteogenesis imperfecta (OI)

OI type	DGI*	Clinical features	Inheritance**
IA	-	Normal or short stature Little or no bone deformity Blue sclerae Hearing loss common	AD
IB	+		
II	?	Severe osseous fragility, perinatally lethal	AD (de novo mutations) AR (rare)
III	+/-	Very short stature Progressively deforming bones Scleral hue varies Hearing loss less common than type I	AD AR (uncommon)
IVA	-	Variable short stature Mild to moderate bone deformity Normal sclerae Hearing loss less common than type I	AD
IVB	+		

Modified from Sillence 1979
*AD = autosomal dominant; AR = autosomal recessive

Dentin

- Under enamel
- Supports the enamel
- Protecting the pulp
- Hydroxyapatite
- Collagen type I
- Similarities to bone tissue

Dentin formation

- Dental papilla
- Collagen type I trimers, III, GA,GP and PG
- Odontoblast differentiation
- Predentin
- Collagen type I increases

Bone vs. dentin

Bone
- 70 % hydroxyapatite
- 20 % organic
- Remodellation
- Response to hormones

Dentin
- 70 % hydroxyapatite
- 20 % organic
- No remodellation

Assessment of DGI

- Clinical assessment
  - -Color
  - -Attrition?
  - -Fractures?
- Radiographic assessment
- Histological assessment?
- More common when qualitative defects
- Both dentitions affected more often when qualitative defects

Levin et al. 1980
Lund et al. 1998
O'Connell and Marini 1999
Rauch et al. 2010
Lindahl et al. 2015
Andersson et al. 2017

Radiological assessment of DGI

- Cervical constriction
- Obliteration
- Short roots
- Pulp stones have also been reported

Shields et al., 1973
Lukinmaa et al., 1987
Malmgren and Norgren, 2002

Assessment of dental variables in OI

- May be of importance for OI diagnosis
- Initially based on clinical findings
- Radiographic and histologic examination may be necessary
- Tooth agenesis, taurodontism, second molar retention and aberrant craniofacial development

Lukinmaa et al., 1987
Malmgren and Norgren, 2002
Malmgren et al., 2016
Andersson et al., 2017

Tooth agenesis in OI

- Congenital absence of one or more teeth
- Agenesis common in OI (17-22%)
- Hypodontia 11%
- Oligodontia 6%

Lukinmaa et al., 1987
Malmgren and Norgren, 2002
Malmgren et al., 2016

Taurodontism

- Lack of cervical constriction at the level of the cemento-enamel junction
- Enlarged pulp chamber
- Apical displacement of the pulpal floor
- 0.3-2.5% general population
- Common in OI (6-42%)

Lukinmaa et al., 1987
Malmgren and Norgren, 2002
Bäckman and Wahlin, 2001
Gupta et al., 2011
Malmgren et al., 2016
Andersson et al., 2017

Retention of permanent second molars

- Frequency 31-37%
- More common in individuals with qualitatively defect collagen type I, 50% vs. 16% (p=0.003)
- 2.3±1.2 molars
- No effect of Pamidronate treatment

Bondemark and Tsiopa, 2007
O'Connell and Marini, 1999
Malmgren and Norgren 2002
Andersson et al. 2017

Malocclusion

- Abnormal craniofacial development
- Class III malocclusion
- Crossbites
- Open bites
- Inhibition of maxillary growth
- Maxillary hypoplasia
- Mandibular protrusion

Jensen and Lund (1997)
Malmgren and Norgren (2002)
Waltimo-Sirén (2005)
Chang et al. (2007)
Rizkallah et al. (2013)

Clinical challenges in OI

- Loss of occlusion
  - -DGI
  - -Agenesis
  - -Abnormal craniofacial development
  - -Infections
- What do we know about dental implants in individuals with OI?
- Can we be safe with oral surgery in OI?
- Esthetic concerns
  - -What can we do?


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