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Hereditary Dentine Disorders and Regional Odontodysplasia

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Hereditary Dentine Disorders



Dentinogenesis Imperfecta (DGI)







Clinical Presentation

- Blue to brown discolouration of the dentition
- Enamel frequently separates from underlying dentine
- Variable expressivity
- Primary and/or permanent dentition
  - **Primary teeth more severely affected** *(Malmgren et al 2004)*





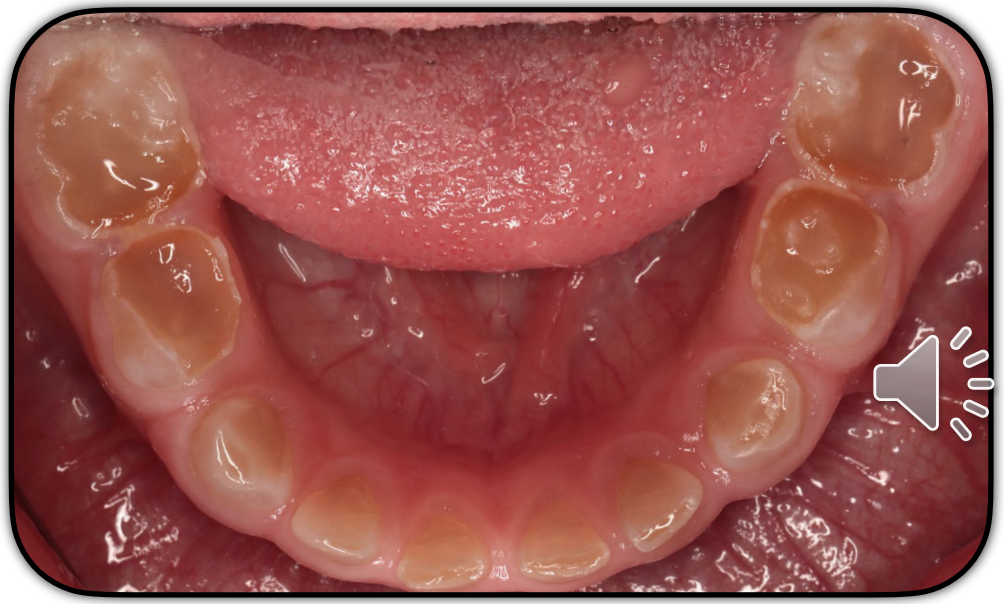
Shields et al 1973

Clinical Presentation

- Accelerated attrition → severely worn dentition
- Spontaneous abscesses
- Pain
- Infection
- Poor aesthetics







Histomorphologic Characteristics

- Dysplastic dentine
  - Amorphous areas void of dentinal tubules
  - Irregular dentinal tubules
  - Embedded cells
  - Interglobular dentine
  - Continuous formation of abnormal circumpulpal dentine
- DEJ
  - Straight with lack of scalloping
- Enamel
  - Normal
  - Some reports of aberrations



Davis et al 2015
Malmgren & Lindskog 2003
Malmgren et al 2004

Prevalence ~ *1 in 8000 children*

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Author	Prevalence	Country
Witkop 1957	0.013 – 0.017 %	United States of America
Gupta et al., 2011	0.09 %	India
Yassin 2016	0.1 %	Saudi Arabia
Cassia et al., 2017	0.057 %	France
Andersson et al., 2018	0.0022 %	Sweden



Classification

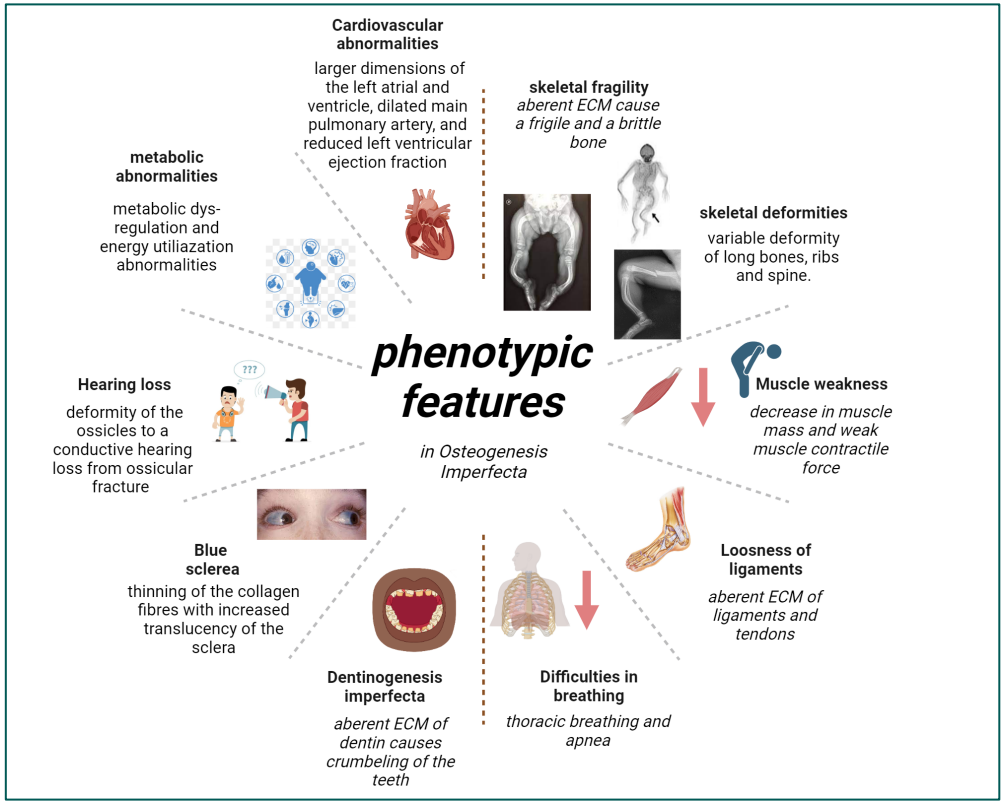
Shields et al 1973:

- Dentinogenesis Imperfecta (DGI) type I, II and III
- Dentin dysplasia (DD) type I and II

- Based on clinical phenotype rather than molecular data
- Does not account for genetic aetiology of hereditary dentine defect

DGI-I

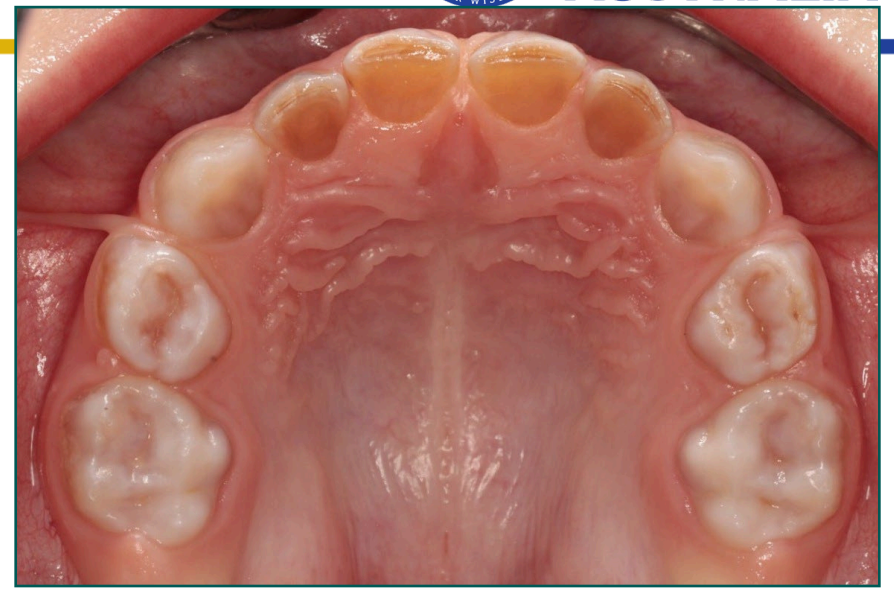
- Syndromic form of DGI associated with, and now classified within, osteogenesis imperfecta
- Discolouration in primary and permanent dentitions
- Radiographically - short, constricted roots, dentine hypertrophy → pulpal obliteration
  - Variable expressivity
- Mild form of OI – risk of misdiagnosis
  - DGI-I may be the only clinically apparent findings

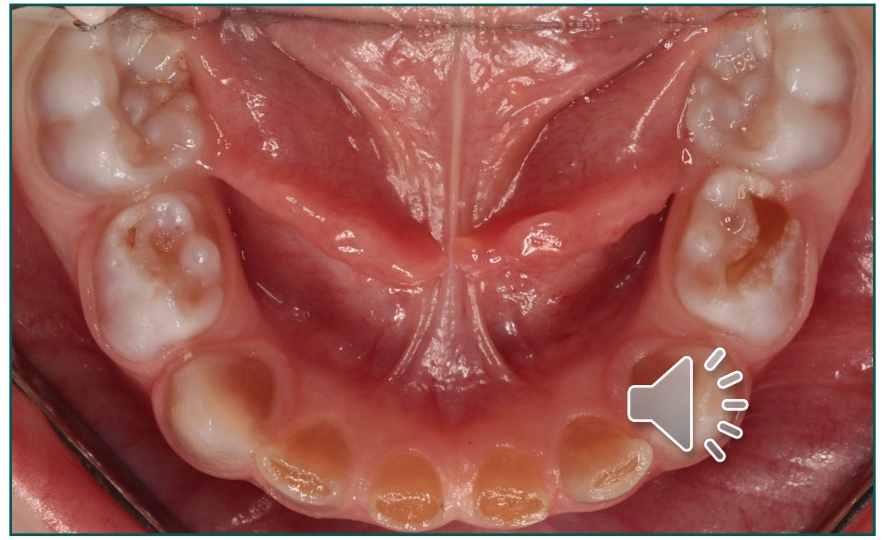


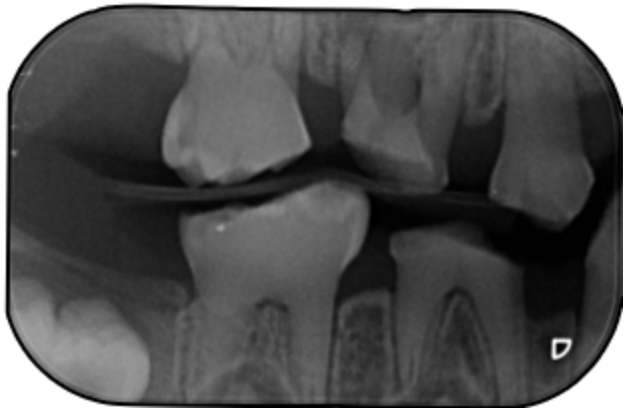
DGI-II

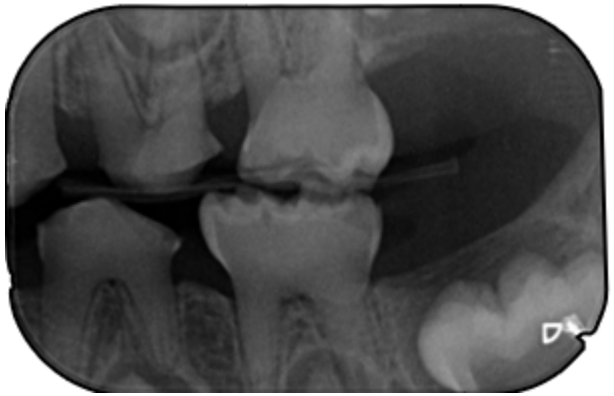
bulbous crowns with marked cervical constriction







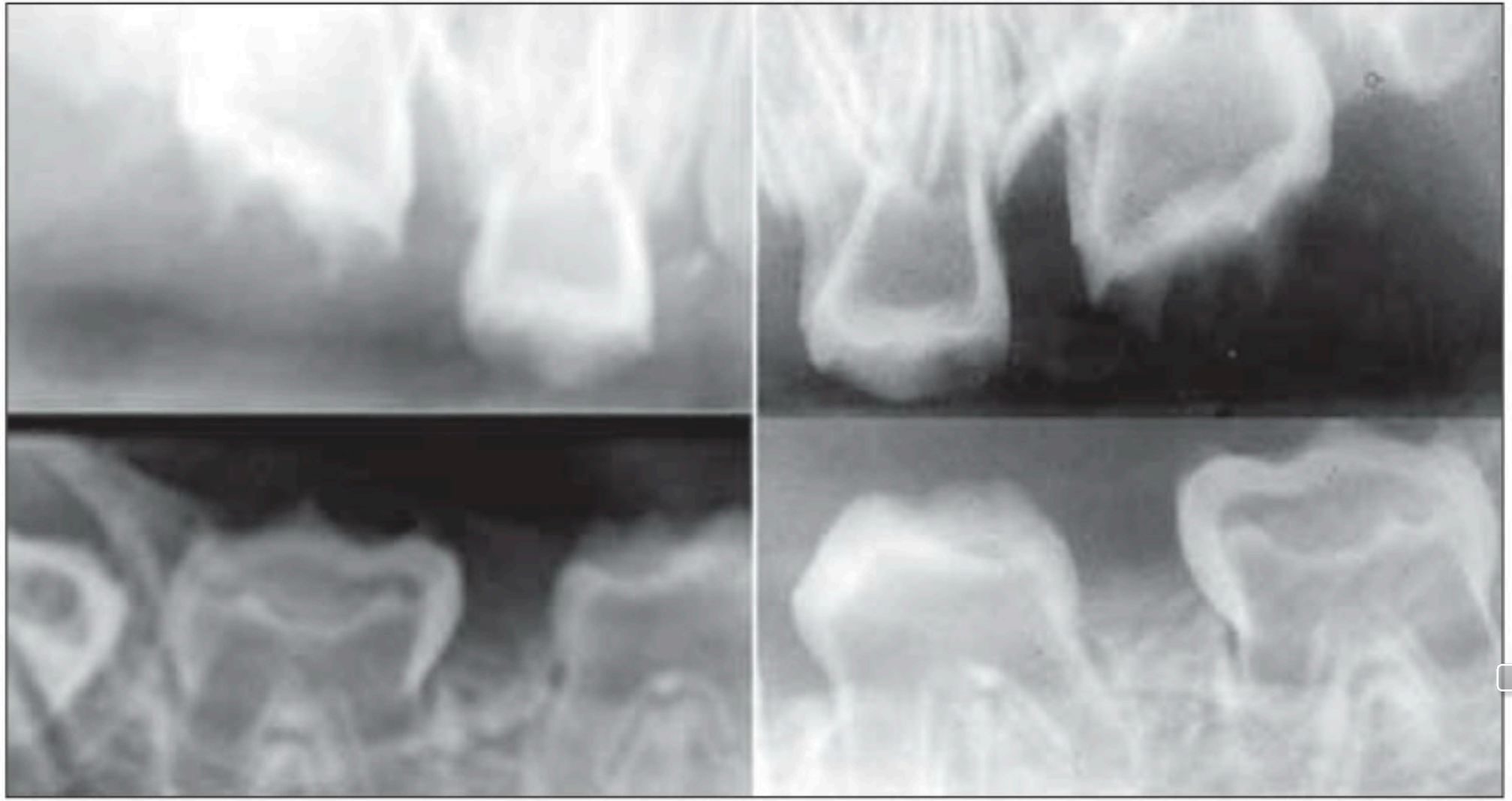




Images courtesy of Dr Ellie Nadian

DGI-III

* Defining feature: shell teeth



Sapir & Shapira et al 1973

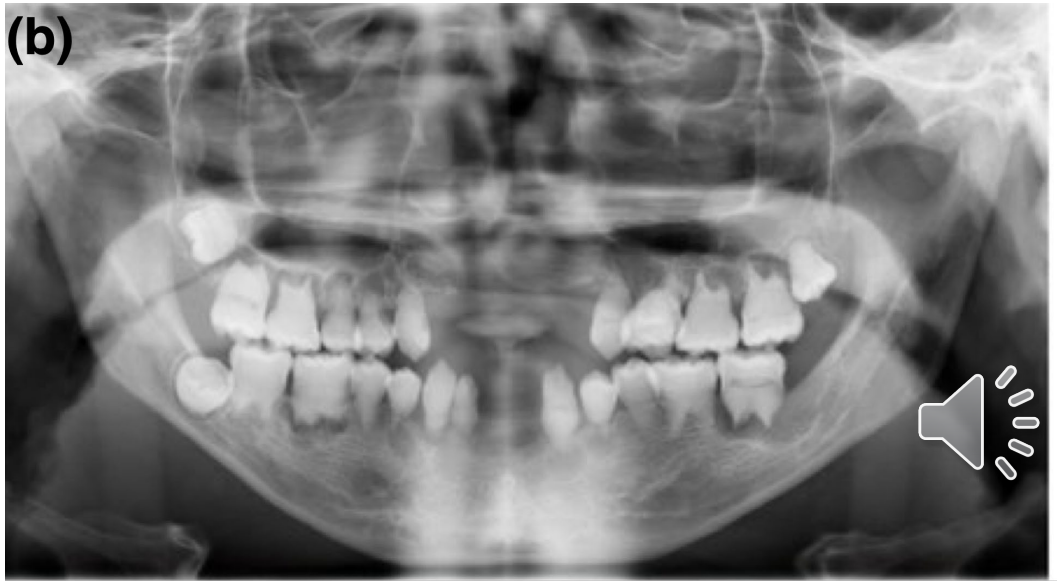
DD-I

- Rarest of the hereditary dentine defects
  - Prevalence of 1:100,000

Witkop et al 1957

- Loss of organization of root dentine
  - Sharp, conical and shortened root lengths
  - Extreme tooth mobility and premature exfoliation





Chen et al 2019

DD-II

- Similar to DGI type II
- Primary teeth affected more severely than permanent teeth
  - Primary teeth in DD-II similar to DGI-I
  - Permanent teeth in DD-II mildly or not affected
- Thistle shaped pulp and pulp stones
  - Thistle shaped pulp = large pulp chamber with thin root canals

Burkes et al 1979

Classification

OMIM	*Shield classification of isolated dentin diseases*	*Proposed classification of isolated dentin diseases*
125400	Dentin dysplasia type I	Radicular dentin dysplasia
		Dentinogenesis imperfecta
125420	Dentin dysplasia type II	Mild form
125490	Dentinogenesis imperfecta type II	Moderate form
125500	Dentinogenesis imperfecta type III	Severe form
166240	*Dentinogenesis imperfecta type I*	*Osteogenesis imperfecta*

de la Dure-Molla et al 2015

Clinical Findings

- Radicular dentin dysplasia (DD type I) - teeth may appear clinically normal. Extreme tooth mobility and early loss of teeth.

- Dentinogenesis imperfecta
  - Mild (DD type-II) – primary teeth more severely affected. Permanent teeth mildly or not affected. Primary teeth discolouration and attrition.
  - Moderate (DGI type-II) - bulbous crowns with marked cervical constrictions. Teeth discolouration and attrition.
  - Severe (DGI type-III) – opalescent primary and permanent teeth, marked attrition, multiple pulp exposures.

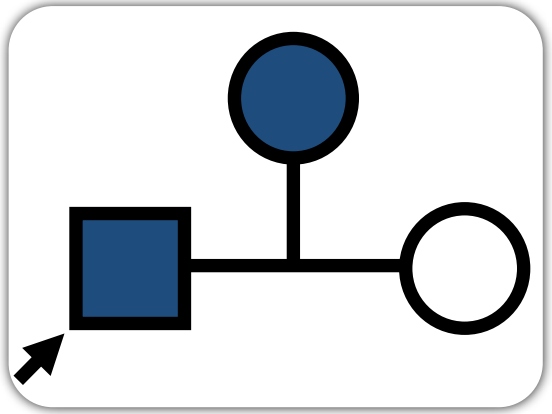
- Osteogenesis imperfecta with DGI (DGI type-I) - teeth discolouration and attrition.

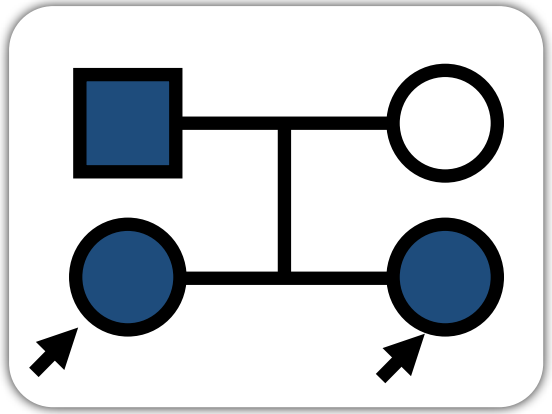
Radiographic Findings

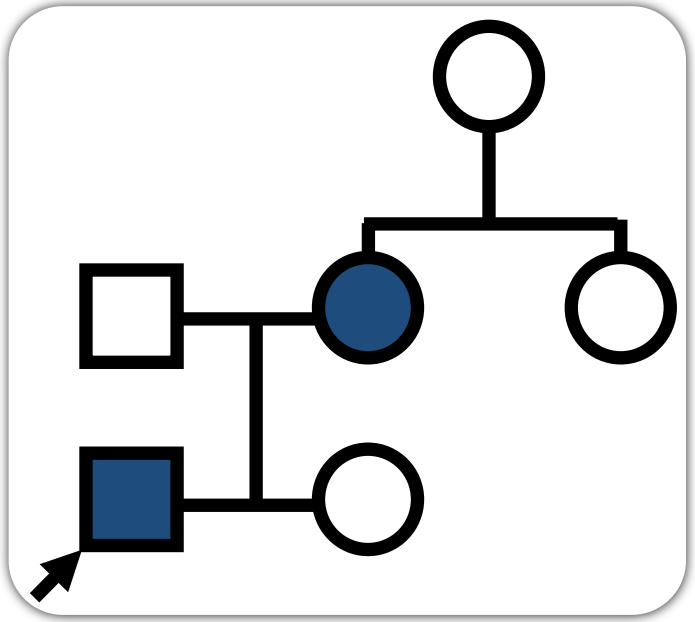
- Radicular dentin dysplasia (DD type I)
  - Sharp conical and shortened roots. short, constricted roots
- Dentinogenesis imperfecta
  - Mild (DD type-II) – thistle shaped pulp (large pulp chamber with thin root canals). Permanent teeth mildly or not affected.
  - Moderate (DGI type-II) - early pulpal obliteration.
  - Severe (DGI type-III) – normal thickness enamel in association with extremely thin dentine and dramatically enlarged pulps aka “shell teeth”.
- Osteogenesis imperfecta with DGI (DGI type-I)
  - Short, constricted roots
  - Pulpal obliteration.

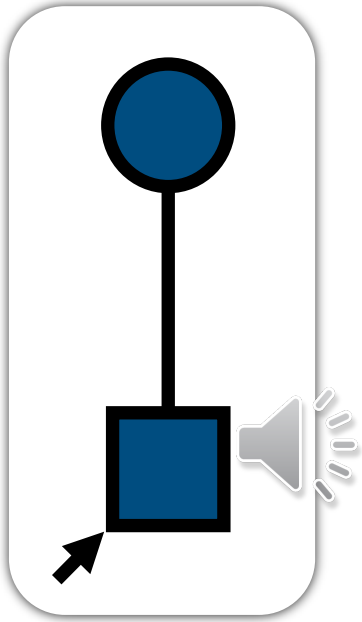
Diagnosis

- Clinical presentation
- Family history
  - Pedigree construction
- ? Genetic diagnosis ?





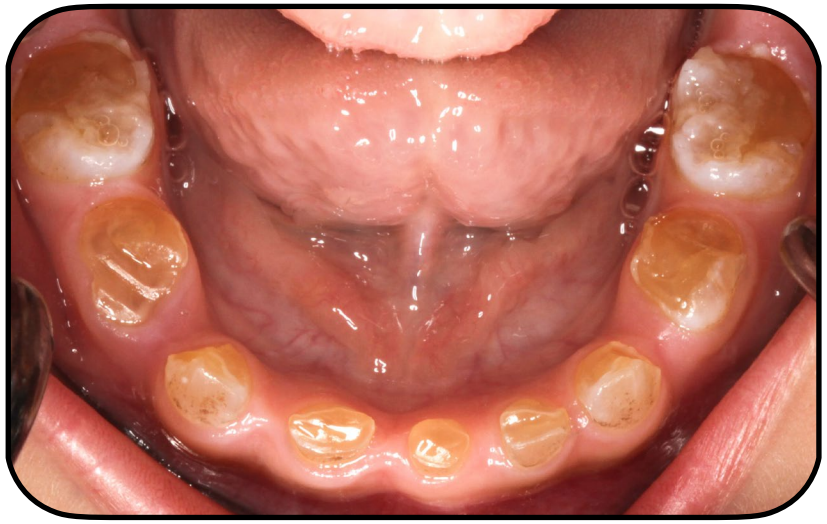


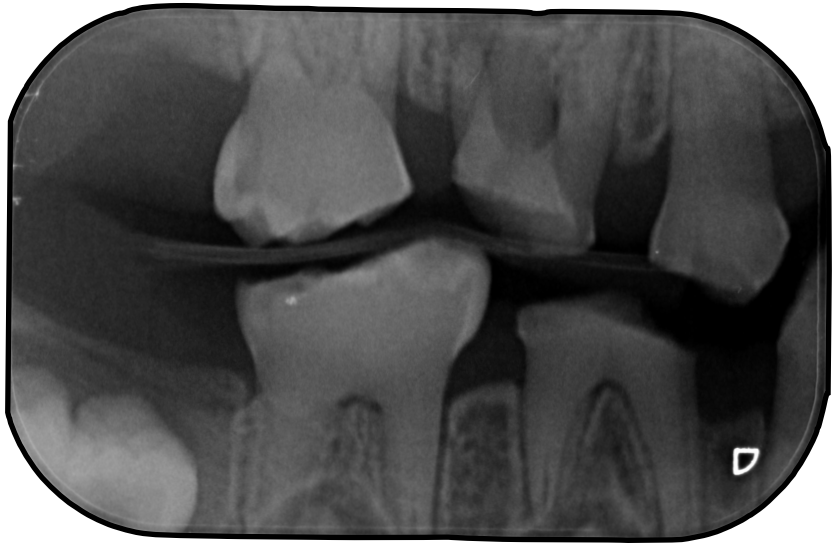


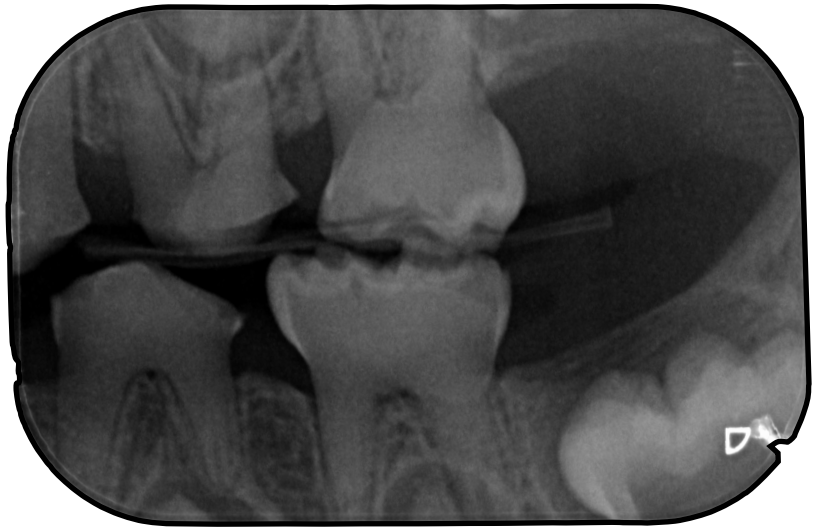
Family 1







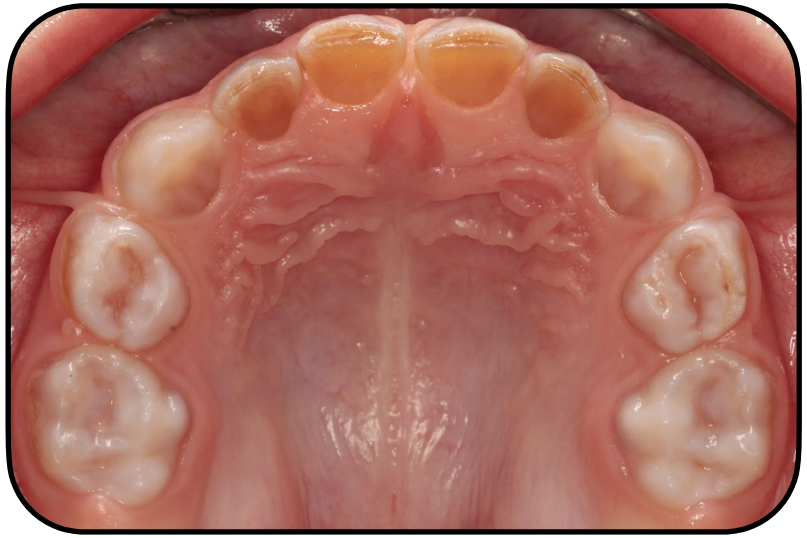


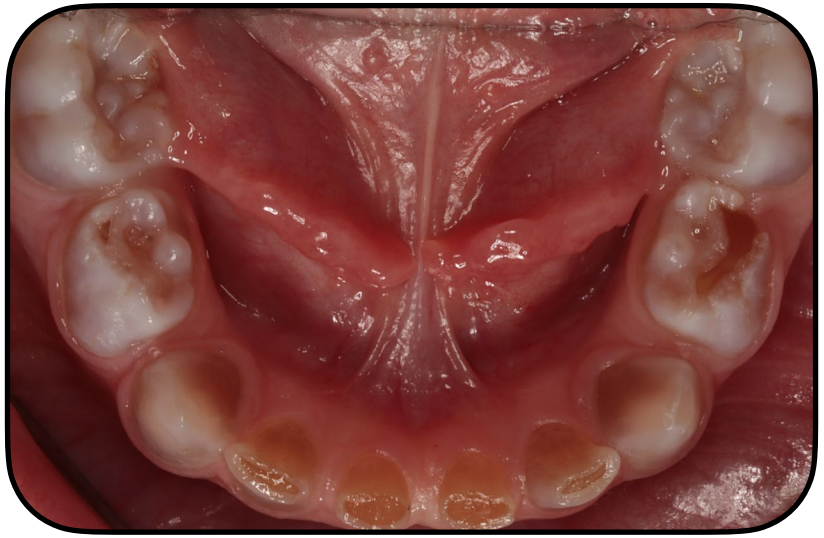


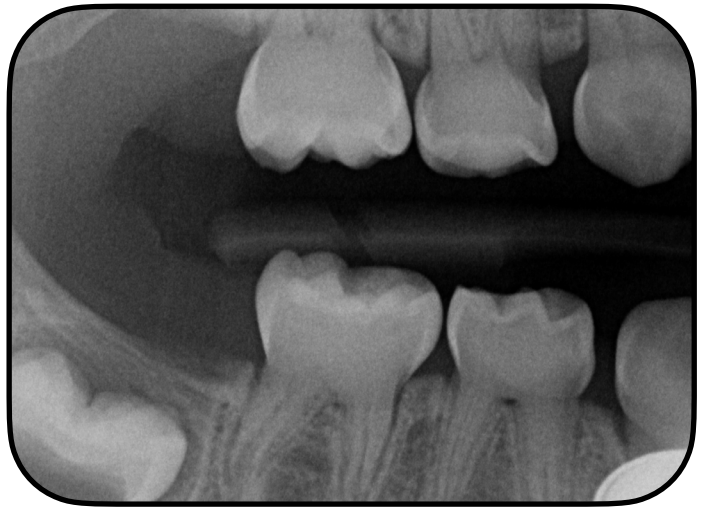
Images courtesy of Dr Maleeha Gilani

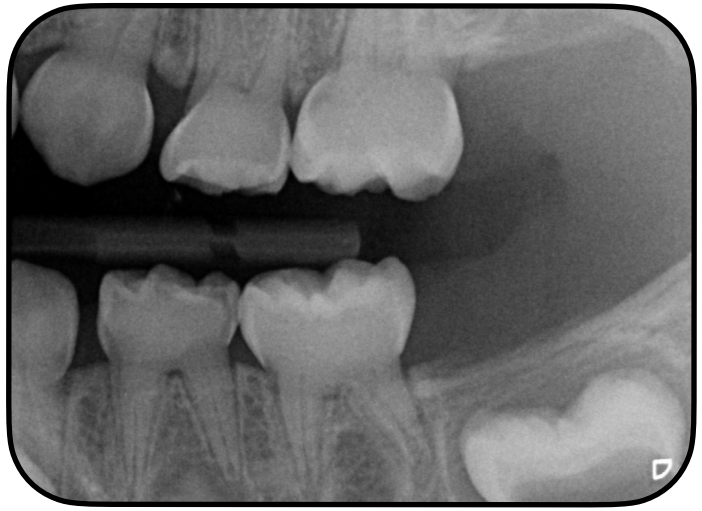
Family 2





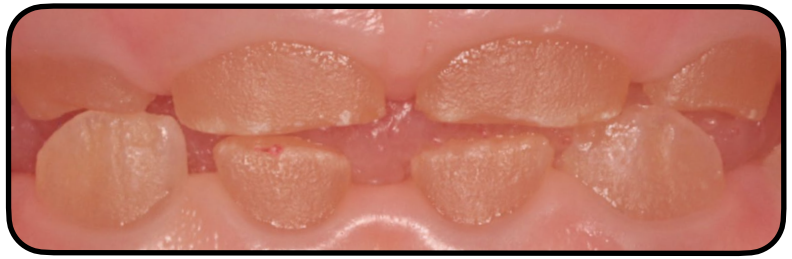


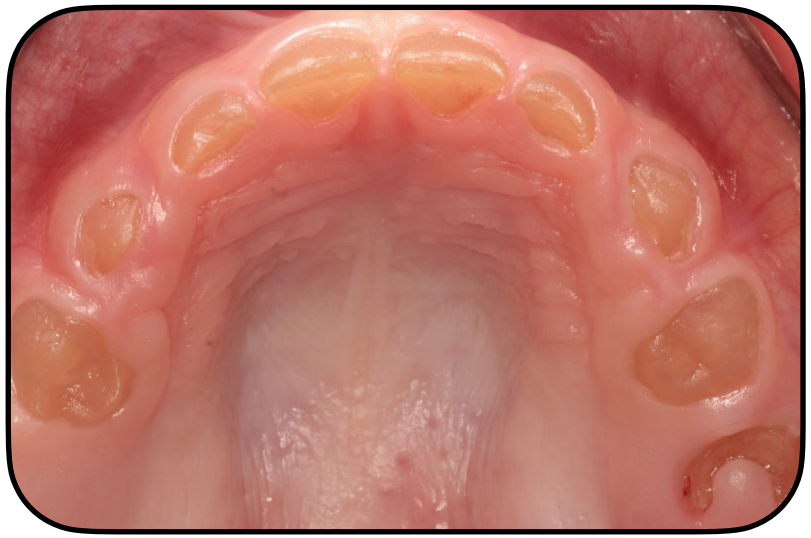


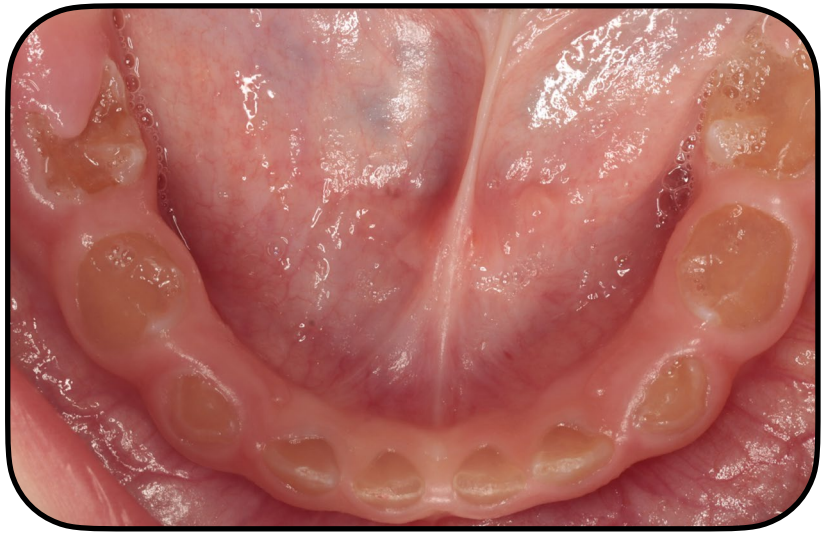


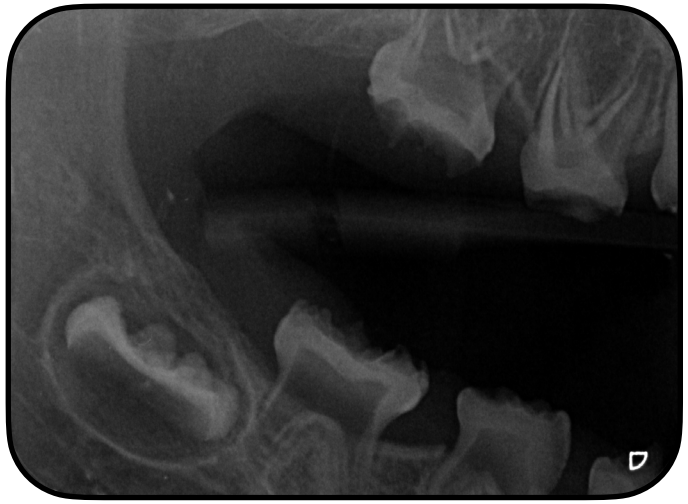
Images courtesy of Dr Maleeha Gilani

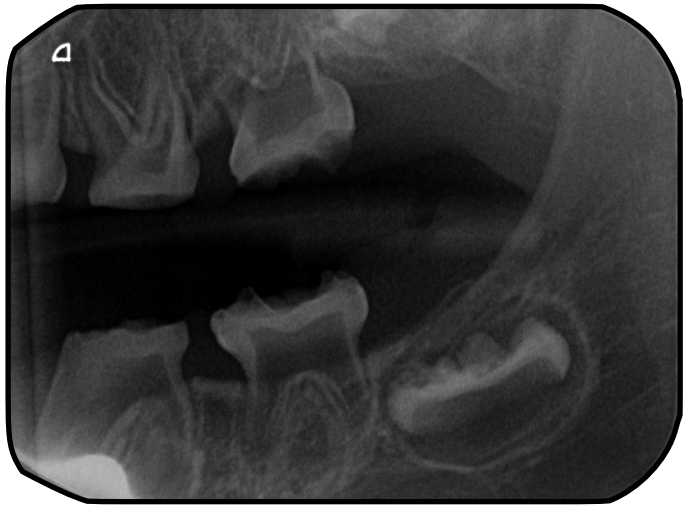
Family 3







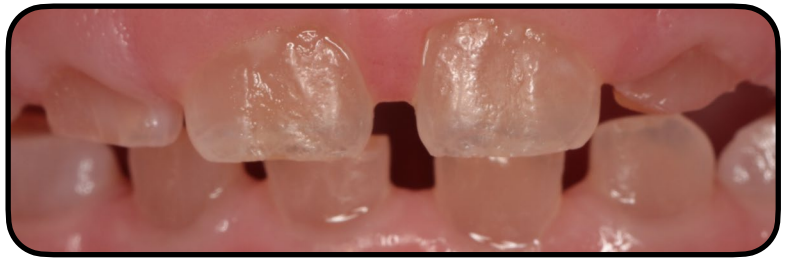


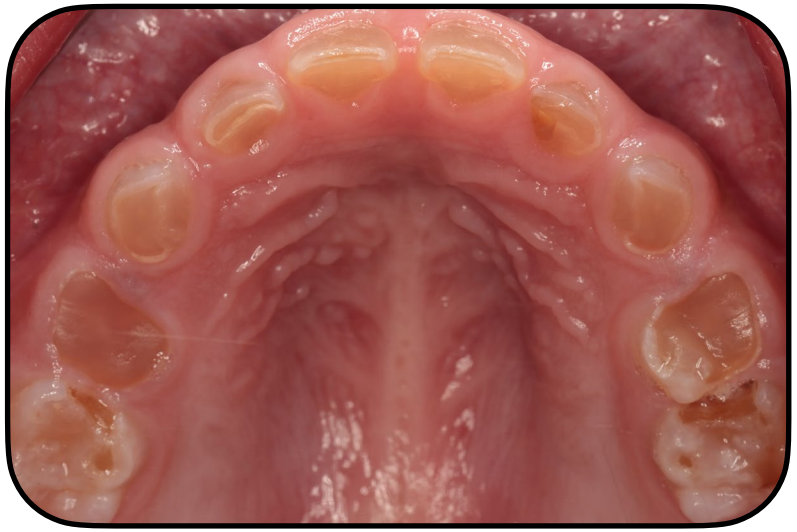


Images courtesy of Dr Maleeha Gilani

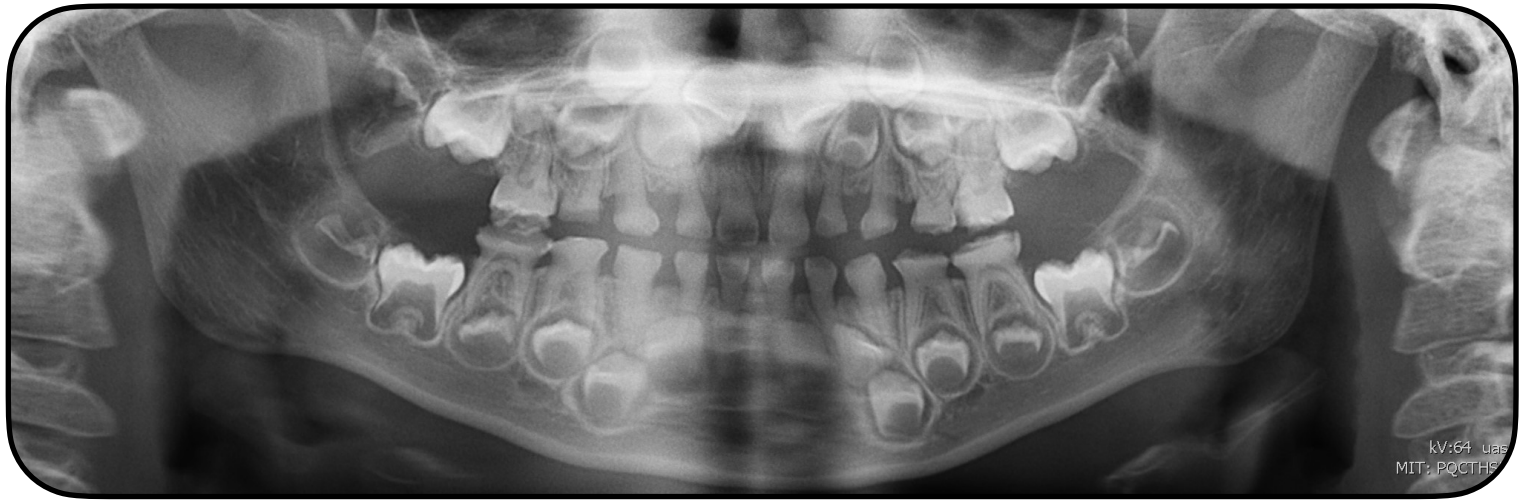
Family 4

Twin 1

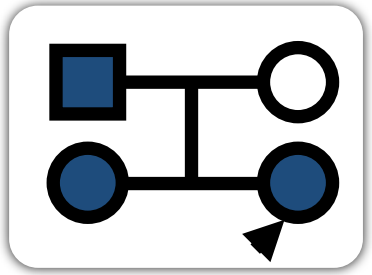








Images courtesy of Dr Maleeha Gilani

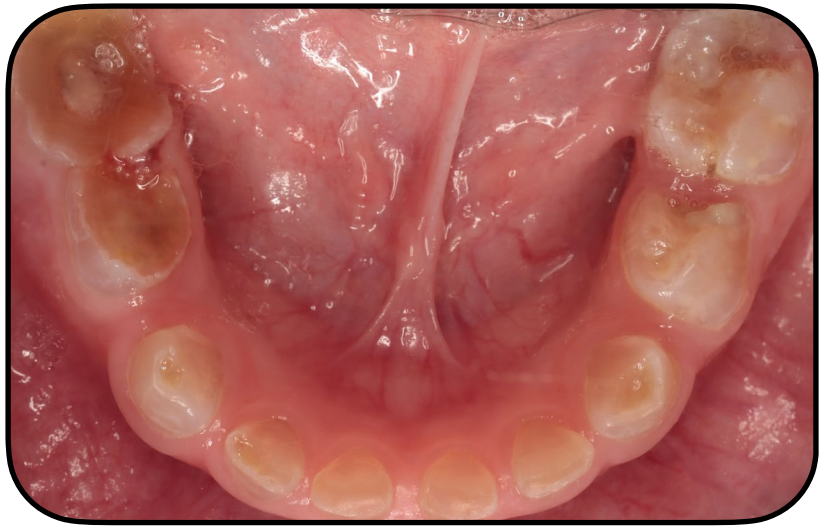


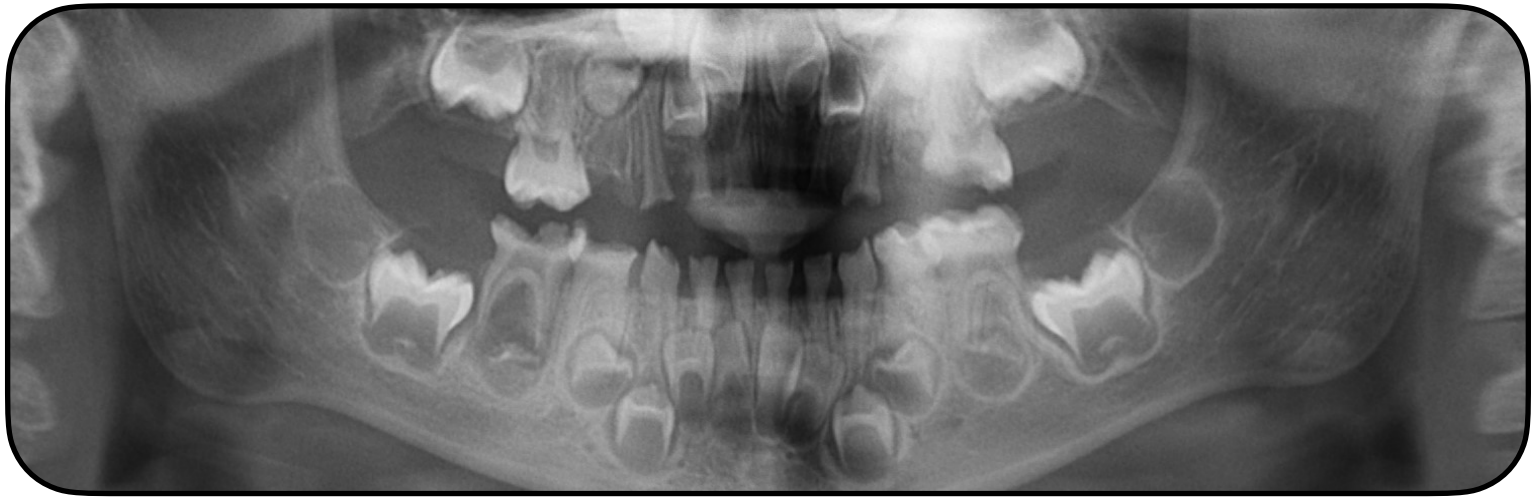
Twin 2

Family 4











Images courtesy of Dr Maleeha Gilani

Implicated Genes

DSPP	Involved in dentine mineralisation
COL1A1	Involved in type 1 collagen pro-α1 chain formation
COL1A2	Involved in type 1 collagen pro-α2 chain formation

Molecular Aetiology

DGI-I:

- Oral manifestation of osteogenesis imperfecta (OI)
- Mutations in genes COL1A1 and COL1A2
- Mild form of OI – risk of misdiagnosis
    - DGI-I may be the only clinically apparent findings

DD-I:

- Pathogenesis largely unknown

Marini et al 2007

Molecular Aetiology

DGI-II, DGI-III and DD-II:

- Conditions are allelic and represent a single disease with variable expressions
- Mutations in the gene which encode DSPP
  - DSPP – key non-collagenous proteins involved in tooth development and mineralisation

Barron et al 2008
de la Dure-Molla 2015
MacDougall et al 1999

Management

- Dependent upon the clinical expression
  - Standard “prescription” not possible

- Multidisciplinary approach
- Early identification critical
- Preventative care and management of tooth sensitivity

- Long term rehabilitation in phases:
  - Primary dentition
  - Mixed dentition
  - Permanent dentition

Management

Primary Dentition:

- Early stage –
  - ~18-20 months
  - Treat and protect first primary molars and incisors

- Second stage –
  - ~ 20-30 months
  - Treat and protect primary canines and second primary molars

- Treatment provided under general anaesthesia

Sapir & Shapira et al 2001



Images courtesy of Dr John Winters



Images courtesy of Dr John Winters

Management

Permanent Dentition:

- Tooth whitening has been used with varying success
- Long term evidence lacking on performance of adhesively bonded restorations
- Dental implants
- Endodontic treatment
    - Often poor success due to partial or complete root canal obliteration

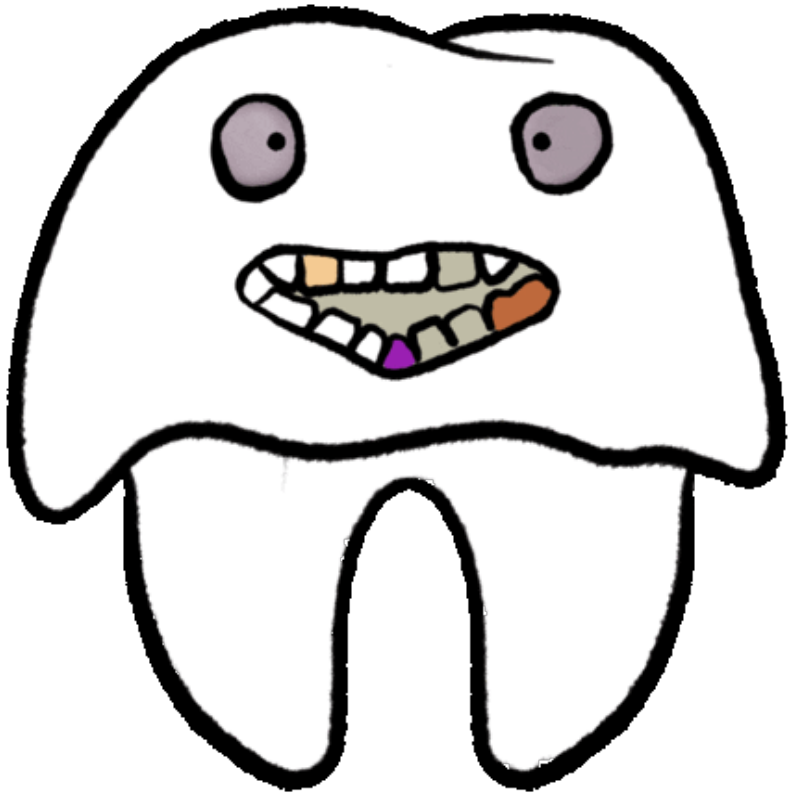
Hereditary Dentine Disorders Summary

- DGI type I = osteogenesis imperfecta
- DGI type II, II and DD type II = single disease with variable expressions
- DD type I = radicular dentin dysplasia

- Early identification important
- Multidisciplinary approach required for treatment
- Eligible through Medicare Cleft Lip and Palate Scheme (PCH)

- Further research required into genes responsible, and management options





"Ghost Teeth"

Regional Odontodysplasia

Regional Odontodysplasia

- Localised non-inheritable developmental anomaly of dental tissues
- Affects both mesoderm and ectoderm tissue
  - enamel
  - dentine
  - pulp
- F>M
- No ethnic predilection



Clinical Features

- Hypoplastic, small teeth
  - surface pits and grooves
  - yellow / brownish discolouration
- Eruption delay
- Maxilla > mandible
- Usually unilateral
- Abscesses in the absence of caries



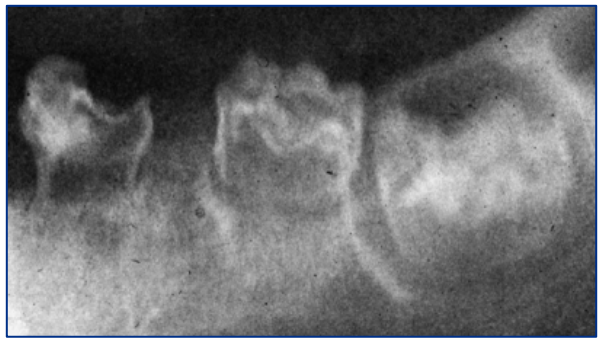
Rashidian et al 2010



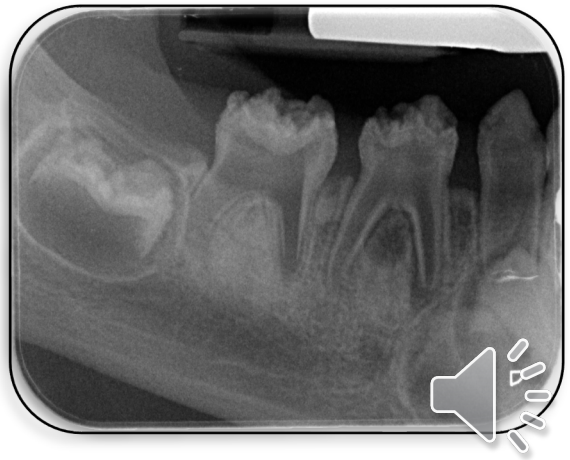
Lowry et al 1992

Radiographic Findings

- Lack of contrast between thin enamel and dentine layers
  - “Ghost-like” appearance
- Large pulp space
  - Pulp stones or denticles



Iizawa et al 2010



Histological Findings

- Enamel is hypoplastic
- Hypocalcified areas and invaginations from the enamel surface into the dentin layer
  - may result in bacterial contamination → pulpitis or necrosis
- Mixed areas of cellular, amorphous and interglobular dentin
- Bone not affected

Aetiology

- Unknown – likely localised insult rather than inheritable condition
  - Local circulatory disorders
  - Viral infections
  - Local trauma
  - Pharmacotherapy during pregnancy
  - Facial asymmetry
  - Combination of these factors

Treatment Options

- Medicare Category 7 - Cleft Lip and Palate Scheme
- Treatment dependant on severity of defect
- Maintenance of teeth if possible
  - Temporisation (GIC, composite, SSC)
  - Hall crown technique
  - Prosthodontic restoration
- Extraction
  - Removable acrylic orthodontic appliance ?
  - Implant
  - Autotransplantation
  - Removable denture

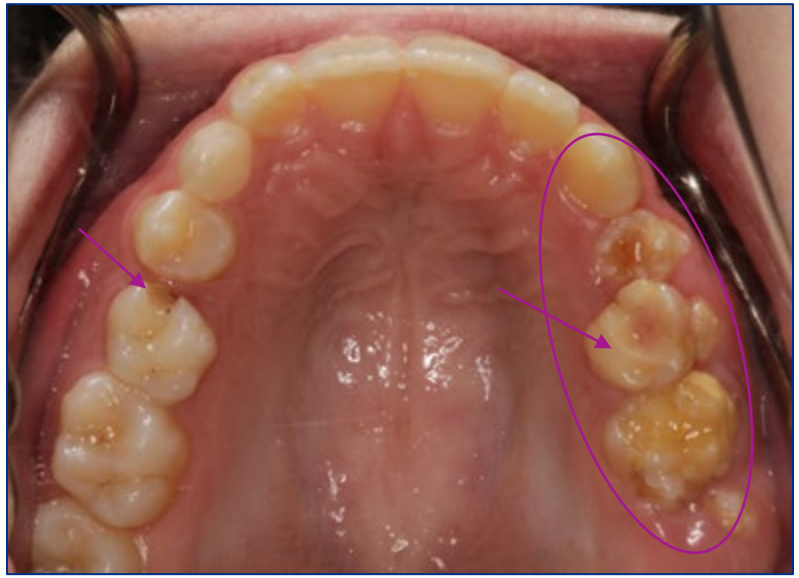


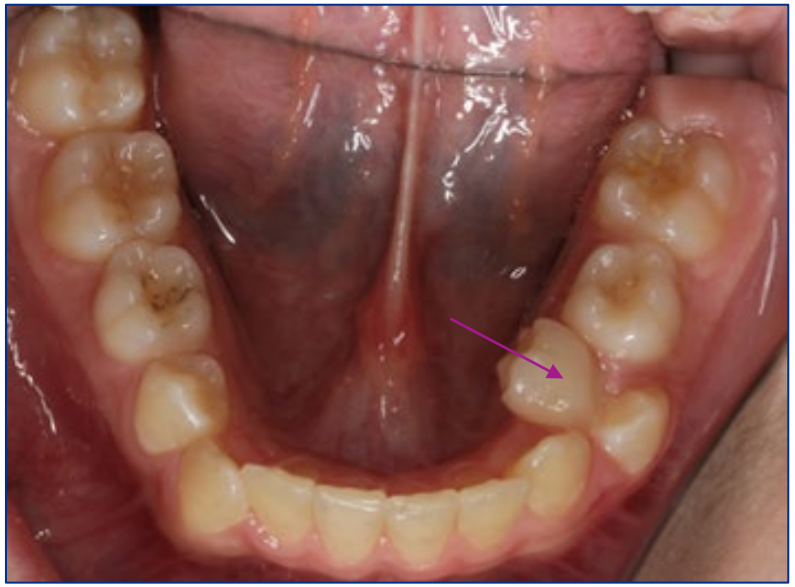
Patient 1

- 12-year-old girl
- Referred by private paediatric dentist for management of regional odontodysplasia
- No medical history of note
- Sensitivity to cold and toothbrushing – difficulties brushing Q2



Intraoral Findings





Intraoral Findings



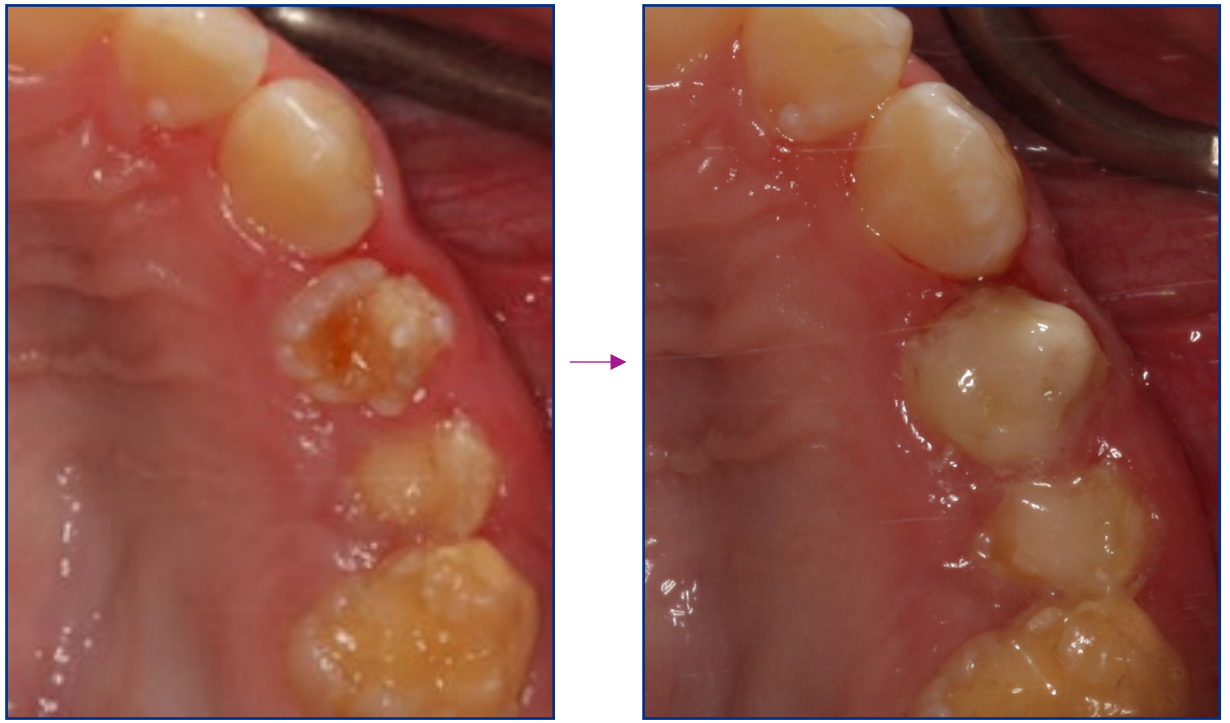


Radiographic Findings



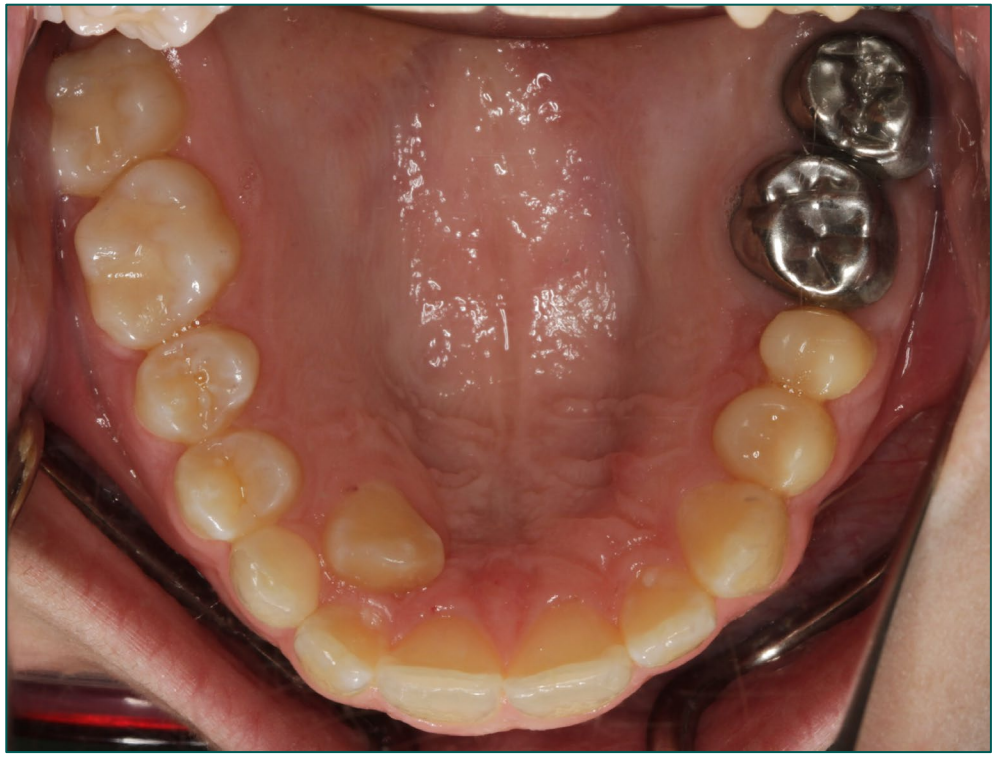
Treatment – Emergency Phase (RA/LA)

- Extraction of 65 and 74
- GIC sealant on 24 and 25
- Flowable composite sealant 23B
- Wait for further eruption of teeth



Treatment Completed – Phase 2 (GA)

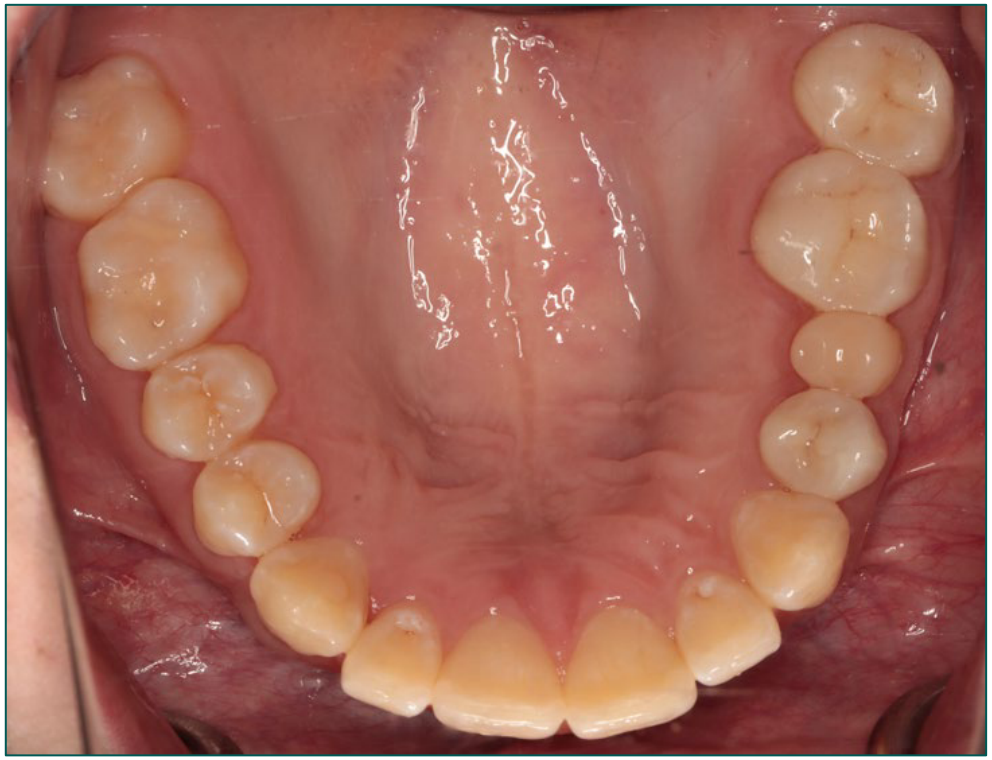




- Open surgical exposure of 13 (53 later extracted just prior to FFA)
- Composite crown 24 and 25 (3M bicuspid frasaco former)
- SSC 26 and 27
- Recalls during active ortho treatment

Treatment Completed – Phase 3 (GA)





- Monolithic zirconia crowns teeth 24, 26, 27
- Discharged from PCH to private dentist
- Aware 28 may require extraction or treatment

Patient 2

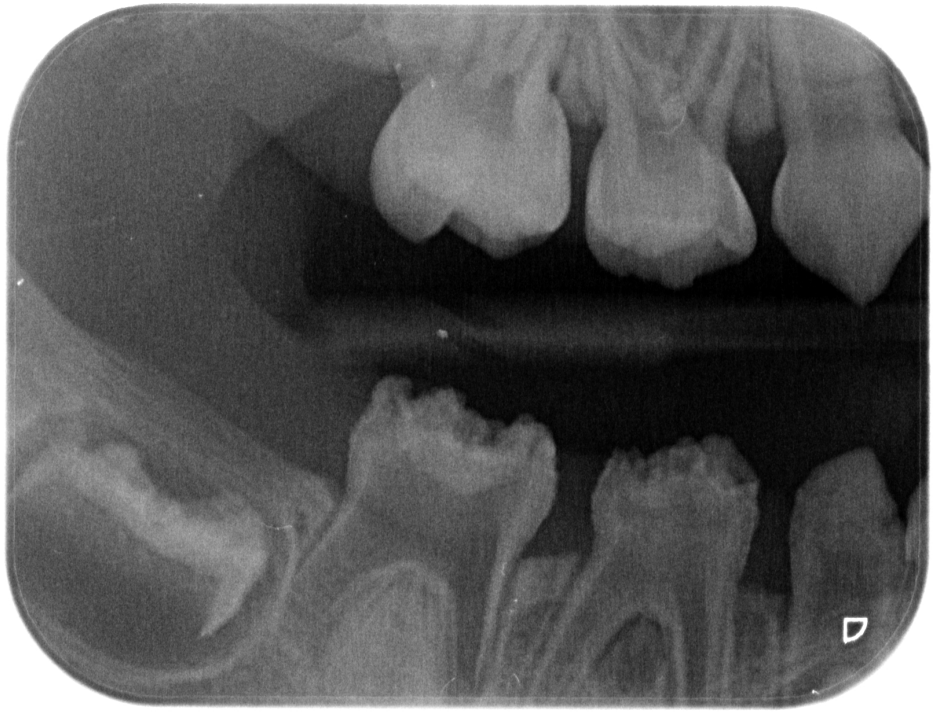
- 3-year-old boy
- Referred by community dentist for management of RHS facial swelling
- Medical history - multiple facial swellings in past month
  - prescribed 2 courses of antibiotics



Radiographic Findings



Radiographic Findings





Management

- Emergency treatment under general anaesthesia
  - 84 extraction
  - 85 SSC (Hall technique)
- Ongoing treatment
  - 83 six monthly Duraphat applications



Regional Odontodysplasia Summary

- Localised non-inheritable developmental anomaly of dental tissues affecting adjacent teeth
- Affects both mesoderm and ectoderm tissue
  - enamel
  - dentine
  - pulp
- Multidisciplinary care required