Introduction: Hereditary dentin diseases are challenging in terms of both diagnostics and therapy, largely because the clinical presentation is highly diverse and all available treatment strategies lack supporting evidence. In the present paper, we report on a 25-year-old female patient with late-diagnosed dentinogenesis imperfecta (DI) in which the hard tissue defects were treated minimally invasively using direct adhesive restorations. Based on this case, the currently established classification of DI is reviewed critically and the selected treatment approach contrasted with alternative treatment strategies.

Methods: Hard tissue defects were restored using directly placed composite. The patient was followed up over 12 months.

Results: The direct and minimally invasive strategy that was selected allowed the restoration of teeth without additional hard tissue loss in a short time period and with limited costs. The functional and esthetic results were satisfactory.

Conclusion: A range of treatment strategies is available for managing DI. Nevertheless, all lack supporting evidence. For this specific case, the chosen strategy offered advantages over indirect restorations. The long-term prognosis remains unclear.

Keywords: Dentinogenesis imperfecta; minimally invasive therapy; composite restoration; Shields classification; interdisciplinary diagnostics
Introduction

Dentinogenesis imperfecta (DI) represents one of the autosomal-dominant inherited dentin disorders and is characterized by clinically manifested pathological dentin structures. DI is rather rare with an estimated prevalence of 1:6,000 to 1:8,000 [2, 8, 6]. In the classification which was developed by Shields in 1973 and is still in use today, DI is differentiated from dentin dysplasia and divided into 3 types [2, 14]:

1. Type I is based on a mutation of the genes (COL1A1 and COL1A2) encoding collagen I. Type I is associated with osteogenesis imperfecta [10] whereby the dental anomalies are considered more of an accompanying disorder. The dentin is hypomineralized, resulting in frequent fractures of the enamel which is inadequately supported.

2. Type II and type III only affect the dentin whereby type III depicts the more severe form and its occurrence appears to be regionally very restricted (Maryland and Washington, DC, USA) [2]. Types II and III are not based on mutations in the collagen genes (as is the case with type I) but are instead based on genetic mutations in chromosome 4q22. The dentin sialophosphoprotein (DSPP) gene, which encodes the noncollagenous dentin matrix protein of the same name, is located on this chromosome [1, 2]. DSPP is primarily involved in the formation and organization of the dentin structures [9, 14]. DSPP can also be detected in the tissues of other organs such as bones, kidneys, liver, or lungs although its expression in the dentin is about one hundred times higher [8].

Clinically, the types of DI are characterized by a number of common characteristics which nevertheless vary across the types. The teeth have an amber to blue or gray color and are often significantly worn or have multiple fractures since the enamel easily detaches from the dentin [8, 12, 14]. This can be associated with a loss of the vertical dimension. The crowns also tend to have a rather bulbous anatomy.

Radiographs show partly or completely obliterated pulp chambers along with roots that are shortened and/or very tapered. These findings can involve both the primary and permanent dentition (DI–I). Sometimes, the primary teeth can be more severely affected than the permanent teeth (DI–II) [2, 9, 14]. Patients often complain about pain due to apical infections with no discernable cause.

Figure 1a Initial situation of the patient at the first consultation. The grayish to amber-colored staining of the teeth was particularly remarkable, although they were affected to differing degrees. The anterior mandibular teeth were greatly affected and also had enamel fractures, while, for example, teeth 11 and 21 were clinically unaffected. The patient had a slight midline shift to the right.

Figure 1b Intraoral view of the initial situation of 45. On the distal and occlusal surfaces the fracture gap can be seen in the mesiodistal direction.

Figure 1c Intraoral view of the initial situation of 47. The enamel fracture along the temporary filling can be seen distally. The cervical fracture margin was overgrown by gingiva.

Figure 1d Mandibular anterior teeth with considerable abrasions and enamel fractures of the central incisors.

Figure 1e Initial situation of the patient at the first consultation. The grayish to amber-colored staining of the teeth was particularly remarkable, although they were affected to differing degrees. The anterior mandibular teeth were greatly affected and also had enamel fractures, while, for example, teeth 11 and 21 were clinically unaffected. The patient had a slight midline shift to the right.
An established therapeutic concept is often recommended for the treatment of DI. This involves the insertion of preformed steel crowns of the first affected primary teeth. This aims to protect the teeth from occlusal stresses because of their susceptibility to fracture and to preserve the vertical dimension [2, 7]. Occasionally, the first permanent molars are treated with steel crowns until all permanent teeth have erupted to ensure that bite elevation develops properly. Ultimately, all affected teeth and (in most cases all teeth) are crowned in young adulthood to preserve or restore the vertical dimension and to protect against fractures [2]. In the present paper, we report on a female patient with late-diagnosed DI in whom the existing defects in the hard tissue were not treated in accordance with this concept, but instead using a minimally invasive technique.

**Medical history**

The 25-year-old patient presented initially due to acute pain in region of the lower right 5 (LR5, tooth 45). In her medical history, the patient stated “back problems”; the orthopedic diagnoses were hyperkyphosis, scoliosis, osteochondritis, and osteoporosis. In her dental history, the patient mentioned a “special situation” regarding her teeth which had not been precisely assessed to date by her attending doctor. In her childhood she had experienced traumatizing serial extractions of primary teeth. The lower 6-year molars were extracted shortly after eruption. Recently, when biting hard food, a tooth in her right mandible cracked. Since then the patient has had pain in region 45 but occasionally also “further back”. There are no similar cases in the family.

**Findings**

The patient did not have any pathological extraoral findings. The intraoral mucous membranes were also normal. During a visual-tactile examination the exceptional coloring of the dentition as well as two enamel fractures in the 4th quadrant were noticeable. Further (oral medical, periodontal) pathological findings were not detected.

Figure 2 Panoramic radiograph after the initial treatment of the patient. The lack of pulp chambers and canals, the bulbous crown morphology of the molars, and the pointed root anatomy of most teeth were particularly noticeable. Further (oral medical, periodontal) pathological findings were not detected.

Figure 3a/b Bitewing images after the initial treatment of the patient. Along with radiopacity due to coronal restorations, the bulbous crown shape and extensive obliteration of the pulp chambers were noticeable. No carious lesions were detected. The dentin structures were homogeneous.
(Fig. 1c). The cervical fracture margin could initially not be assessed. At this point, the mandibular anterior teeth were already affected by abrasion and the typical enamel fractures (Fig. 1d).

Sensitivity tests (using cold spray) in the molar region were uniformly negative, while in the premolar and anterior regions the tests were irregularly positive. Teeth 15, 11, and 21 reacted reproducibly positive. The PSI index was 1, 1, 1, 1*, 2, 1 and the BEWE index was 0, 0, 0, 0, 0, 0.

The patient’s caries risk was assessed as low because of her good oral hygiene and, apart from 16, all existing restorations having been attributed to fractures (and not caries). Furthermore, the risk factor profile analysis was favorable (limited amount and frequency of consumption of fermentable carbohydrates, good oral hygiene, use of fluoride toothpaste, etc.). The reason for the extractions of the now missing teeth in the mandible was no longer known. The vertical dimension was preserved.

For the radiographic examination a panoramic radiograph (Fig. 2) and bitewing images (Fig. 3) were used. The characteristic features of DI such as bulbous crowns, pointed roots in some cases, and obliterated pulp chambers and canals were apparent; however, not all teeth were equally affected. Teeth 15 to 25 showed normal anatomy, for example, with clearly visible root canals. No apical translucencies often described in DI patients were detected.

The bitewing images also revealed the characteristic bulbous crown shape (Fig. 3). While the molars and premolars in the mandible did not have any radiographically visible root canals, the pulp chambers of the premolars in the 1st quadrant were properly visible and those in the 2nd quadrant were only partly obliterated. No carious lesions were detected.

**Diagnosis and therapy planning**

The patient was first informed about the tentative DI diagnosis. During the consultation possible complications and consequences for dental treatment were clarified and a preliminary treatment concept was prepared together with the patient. The issue of tooth preservation was a clear priority for the patient. The prognosis for tooth 45, for which a crownroot fracture with no pulp involvement was initially diagnosed, could only be assessed after removal of the fractured fragment. Likewise, the extent of the loss of dental hard tissue on tooth 47 was also only assessed during therapy. If the teeth

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**Figure 5a** Initial situation and fracture of tooth 47. The fracture margins were already overgrown by gingiva. The tooth had been treated alieno loco with glass ionomer cement.

**Figure 5b** Removal of the temporary restoration revealed a deep subgingival fracture margin. Like for tooth 45, the dentin was rust-colored and softened.

**Figure 5c** Status after restoration with composite. The anatomical structure of the tooth could be restored.

**Figure 6** The mandibular anterior teeth initially showed abrasions and minor enamel fractures (a). These were treated restoratively with composite (b).
were retainable, they were to be re-
stored adhesively using composite.

The therapeutic goal defined with
the patient was to preserve all exist-
ing teeth using non-invasive or min-
imally invasive procedures as far as
possible. The very limited financial
resources of the patient and her de-
sire for prompt treatment were im-
portant criteria here. Further loss of
hard tissue should also be avoided.

On tooth 47 the gingiva was like-
wise removed to expose the fracture
margin on the distolingual side.
Again, a deep subgingival defect was
revealed, this time in an area difficult
to access. As was the case with
tooth 45, this was not just an enamel
fracture but rather an enamel dentin
fracture (Fig. 5a). After removing the
glass ionomer cement filling (Fig. 5b),
the new restoration could be placed
under relative moisture control
(Fig. 5c). The procedure was ana-
logous to the restoration of tooth 45.

A proper anatomical build-up in
the anterior mandible was not pos-
sible, particularly for the central in-
cisors, due to existing static and dy-
namic occlusal contacts. Therefore,
the missing enamel areas were com-
penated within the now existing
restorative extent (Fig. 6). In the ap-
proximal areas a transparent matrix
was used where necessary. The incisal
edges and surfaces were built up with
a composite (ceram.x universal) after
using the etch-and-rinse two-bottle
adhesive system (Optibond FL).

The patient was then transferred
to supportive therapy. Fluoridation
with a 5 % NaF suspension (fluoridin
N5, VOCO, Cuxhaven) and a visual-
tactile examination of the restora-
gins. Subsequently, the softened den-
tin on the visible surface was care-
fully removed (Fig. 4b). An effective
adhesive bond could only be ex-
pected to a limited degree in the
dentin area due to the structural de-
fects. Hence, the aim was to achieve
strong adhesion to the remaining
enamel margins. On the mesial side,
a transparent matrix was positioned
with a wooden wedge for shaping
the proximal areas. For the adhesive
bond, 3-step etch-and-rinse system
(Optibond FL, Kerr, Bioggio, Switzer-
land) was used and the restoration
(SDR and ceram.x universal, Dentsply
Sirona, Constance) was subsequently
placed using a multi-layer technique.
All restorations were cured for 20 sec
for each increment using a polymer-
ization lamp with a light intensity
of 1,500 mW/cm². Figure 4c shows
the restoration of 45 after comple-
tion.

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The exposed dentin again showed a
rust-colored to brown appearance
and reduced hardness.

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Figure 7a Frontal view 12 months after treating the patient

Figure 7b Lateral view 12 months after treating the patient

Figure 7c Maxillary image 12 months after treating the patient

Figure 7d Mandibular image 12 months after treating the patient

Figure 7e Frontal view 12 months after treating the patient

Figure 7f Lateral view 12 months after treating the patient

Figure 7g Maxillary image 12 months after treating the patient

Figure 7h Mandibular image 12 months after treating the patient

Figure 7i Frontal view 12 months after treating the patient

Figure 7j Lateral view 12 months after treating the patient

Figure 7k Maxillary image 12 months after treating the patient

Figure 7l Mandibular image 12 months after treating the patient

Figure 7m Frontal view 12 months after treating the patient

Figure 7n Lateral view 12 months after treating the patient

Figure 7o Maxillary image 12 months after treating the patient

Figure 7p Mandibular image 12 months after treating the patient

Figure 7q Frontal view 12 months after treating the patient

Figure 7r Lateral view 12 months after treating the patient

Figure 7s Maxillary image 12 months after treating the patient

Figure 7t Mandibular image 12 months after treating the patient

Figure 7u Frontal view 12 months after treating the patient

Figure 7v Lateral view 12 months after treating the patient

Figure 7w Maxillary image 12 months after treating the patient

Figure 7x Mandibular image 12 months after treating the patient

Figure 7y Frontal view 12 months after treating the patient

Figure 7z Lateral view 12 months after treating the patient

Figure 7aa Maxillary image 12 months after treating the patient

Figure 7ab Mandibular image 12 months after treating the patient

Figure 7ac Frontal view 12 months after treating the patient

Figure 7ad Lateral view 12 months after treating the patient

Figure 7ae Maxillary image 12 months after treating the patient

Figure 7af Mandibular image 12 months after treating the patient

Figure 7ag Frontal view 12 months after treating the patient

Figure 7ah Lateral view 12 months after treating the patient

Figure 7ai Maxillary image 12 months after treating the patient

Figure 7aj Mandibular image 12 months after treating the patient

Figure 7ak Frontal view 12 months after treating the patient

Figure 7al Lateral view 12 months after treating the patient

Figure 7am Maxillary image 12 months after treating the patient

Figure 7an Mandibular image 12 months after treating the patient

Figure 7ao Frontal view 12 months after treating the patient

Figure 7ap Lateral view 12 months after treating the patient

Figure 7aq Maxillary image 12 months after treating the patient

Figure 7ar Mandibular image 12 months after treating the patient

Figure 7as Frontal view 12 months after treating the patient

Figure 7at Lateral view 12 months after treating the patient

Figure 7au Maxillary image 12 months after treating the patient

Figure 7av Mandibular image 12 months after treating the patient

Figure 7aw Frontal view 12 months after treating the patient

Figure 7ax Lateral view 12 months after treating the patient

Figure 7ay Maxillary image 12 months after treating the patient

Figure 7az Mandibular image 12 months after treating the patient

KHARBOT, SCHWENDICKE:
Minimally invasive therapy of a late diagnosed Dentinogenesis imperfecta
Discussions and all teeth were carried out every 3 months.

The obliterated or even missing pulp chambers and canals with the associated negative sensitivity tests are a known symptom of DI [2, 4, 9]. The affected teeth were regularly examined radiographically. Figures 7a–d show the situation 12 months after the restorative treatment of the patient.

Discussion

Both the diagnostic difficulties and the therapeutic approach that was selected will be discussed on the basis of the case presented.

The diagnosis of DI appears initially clear in this case. Despite a negative family history, the gray-brown tooth color and the presence of fractures of the hard tissue are typical features of DI. However, not all teeth were uniformly affected by the disorder. With an anomaly of the dentin caused by genetic mutation such as DI, it should be assumed that the entire dentition would be affected. Indeed, with the most common type DI–II all teeth are structurally changed without exception [2].

The radiographic diagnostics confirmed the symptoms of DI such as the bulbous crowns, shortened roots in some teeth, and obliterated pulp chambers and canals. Teeth 15 to 25 were not affected, however, and had clear pulp chambers and root canals.

In principle, hereditary dentin disorders such as DI or dentin dysplasia display considerable phenotypic variations, which can make diagnosis based on the clinically oriented Shields classification from 1973 difficult [2, 12]. As a consequence, clear differentiation of the mild DI type II from other dentin defects (dentin dysplasia I, II) is not always possible. Since mutations only in a single gene (DSPP) have been identified for all 4 isolated hereditary dentin disorders (DI II, DI, DD I, DD II) to date, it may as well be the same disorder with varying clinical presentation. Hence, differentiating these 4 disorders on the basis of the phenotype is only limitedly possible and useful, in part because it remains unclear what therapeutic consequences would result from such a classification.

For the patient presented in this case report, the medical history (osteochondritis, osteoporosis) suggests the presence of DI in association with osteogenesis imperfecta (DI type I). Since autosomal recessive inheritance has been documented for mild cases of osteogenesis imperfecta (e.g., type IV), a negative family history does not necessarily allow to exclude this subtype of DI–I, which is the most serious symptom of the disorder [3, 12, 15]. Treatment of the DI–osteogenesis imperfecta combination should take place in close consultation with an internist.

Differential diagnoses must rule out hypocalcified forms of amelogenesis imperfecta, intrinsic discolorations (e.g., due to tetracycline exposure), or other dental development disorders such as rachitic defects [2]. Amelogenesis imperfecta could be ruled out, however, because the enamel overlaying the dentin defects was developed normally (see Fig. 4a). Similarly, tetracycline defects only affect the enamel and are characterized by horizontal bands, the localization and extent of which depend on the time of exposure and the corresponding status of the amelogenesis. Such typical involvement of the enamel was not encountered in this case. Symptoms of ongoing systemic disorders (rickets) were also not confirmed.

The therapeutic approach used here deviates considerably from the “conventional” therapeutic concept, in part because in most other cases an early diagnosis is made (these cases are often more severely affected and require a different therapeutic strategy than that selected for the patient presented here). Additionally, even the tissue-preserving preparation for modern ceramic crown restorations inevitably causes loss of healthy tooth structure, particularly enamel (which represents the only healthy tooth structure here). Furthermore, complications such as unwanted chipping of the existing enamel during cementation of indirect restorations has been reported, which would then further complicate restoration of the teeth [6].

A minimally invasive therapeutic approach using direct adhesive restoration was instead selected for this patient because the DI, which was also only recently diagnosed, was a mild form. The selected approach is characterized by its reversibility (preserves fall-back options, crowns remain possible), its tissue preservation, and the low costs and short time required (the latter were important aspects for the patient).

However, this concept also involves risks and an unclear prognosis: DI dentin has a considerably lower hardness than healthy dentin [11]; the microstructure of the dentin is also pathologically changed (the tubules and collagen network do not develop normally). DI dentin is also less densely mineralized than healthy dentin. The adhesive bond is therefore largely reduced by a restricted hybrid layer; the adhesion values for conventional adhesives are considerably lower for DI dentin than for healthy dentin [6]. Since the enamel structure remains unchanged, however, reliable adhesive bonding to the enamel margins is possible for defects surrounded by enamel (as was the case with this patient, at least in the coronal area). Overall, both the adhesive bond and the mechanical support of the adhesive restorations, however, were likely to be limited. The restorative treatment of the deep subgingival fracture margin was also difficult. Both moisture control and adaptation of the composite material on these margins proved to be challenging. Should there be a need for further treatment in these areas, e.g., due to a fracture or loss of the restoration or secondary caries, an indirect restoration or further pre-restorative surgical measures may be unavoidable.

Conclusion

Diagnosis and treatment of inherited dentin defects represent a challenge. Firstly, because these disorders are rare and consequently individual dentists have little daily experience with them. Second, the etiology and pathogenesis are little understood and accordingly, causal or biologically based therapies are not possible. Lastly, there are only limited studies supporting any diagnostic and therapeutic concepts. For the patient pres-
ent here, the DI was treated minimally invasively. The long-term prognosis of the placed restorations and the unrestored “DI teeth” is unclear and must be considered to be moderate at best since both the adhesion and the support of the restorations and the enamel coating by the underlying dentin structure were compromised. As part of a continuous supportive therapy, however, the selected therapy concept may represent a useful alternative to more elaborate and invasive treatments.

Conflicts of interest:
The authors declare that there is no conflict of interest within the meaning of the guidelines of the International Committee of Medical Journal Editors.

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