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· 防治实践 ·

区域性牙发育不良伴发先天缺牙1例报道及文献回顾

张贝贝^{1,2}, 黄永清², 段小红¹

1. 口腔系统重建与再生全国重点实验室, 国家口腔疾病临床医学研究中心, 陕西省口腔医学重点实验室, 空军军医大学口腔医院口腔生物学教研室, 空军军医大学口腔医院口腔罕见病与遗传病门诊, 陕西 西安(710032);

2. 宁夏医科大学总医院口腔医院口腔颌面外科, 宁夏口腔疾病研究重点实验室, 宁夏 银川(750004)

【摘要】目的 探讨区域性牙发育不良(regional odontodysplasia, RO)伴发先天缺牙患者的临床特征及其影像学特点, 为临床诊疗提供参考。**方法** 报道1例RO伴发先天缺牙病例的影像学表现和诊治, 对其5年内患牙的影像学特征动态变化进行回顾性总结, 并结合文献进行对比分析。患者为9岁女性, 因“左下后牙拔除后不适7月余”就诊于本院口腔罕见病与遗传病门诊。根据其临床及影像学表现, 诊断为区域性牙发育不良(左下颌)、12和34恒牙先天缺失。制定治疗计划并长期随访。将本次影像学图片与既往影像学资料进行对比, 总结其牙齿发育变化; 结合文献对比分析与既往RO牙齿特征的异同点。**结果** 随访期间症状未加重, 患者选择保守性观察, 待患区牙齿萌出后再行治疗。三个时期(4.5岁、8.5岁、9岁)影像学资料对比结果显示该病例左下颌乳、恒牙均呈现典型“鬼影”状影像学特征, 伴12、34恒牙胚先天缺失及35牙发生延迟。文献回顾分析表明, RO表现为牙萌出延迟、牙体积小、牙冠色黄等临床特征, 并具有典型的“鬼影”状影像学表现。治疗需根据患者具体情况进行个性化设计。目前仅报告5例RO患者伴发先天缺牙, 而恒牙胚发育延迟尚未见报道。**结论** 动态影像学评估在RO早期诊断中具有关键作用, RO有先天缺牙和牙胚发生延迟的现象。长期随访及个性化治疗方案是RO治疗的关键。

【关键词】 区域性牙发育不良; “鬼影”牙; 牙发育; 先天缺牙; 牙萌出; 影像学; 锥形束CT; 乳牙列; 混合牙列

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Regional odontodysplasia accompanied by hypodontia: a case report and literature review ZHANG Beibei^{1,2}, HUANG Yongqing², DUAN Xiaohong¹. 1. State Key Laboratory of Oral & Maxillofacial Reconstruction and Regeneration, National Clinical Research Center for Oral Diseases, Shaanxi Key Laboratory of Stomatology, Department of Oral Biology, Clinic of Oral Rare and Genetic Diseases, School of Stomatology, the Fourth Military Medical University, Xi'an 710032, China; 2. Ningxia Key Laboratory of Oral Disease Research; Ningxia Key Laboratory of Craniomaxillofacial Deformities Research; Department of Oral and Maxillofacial Surgery, Hospital of Stomatology, the General Hospital of Ningxia Medical University, Yinchuan 750004, China

Corresponding author: DUAN Xiaohong, Email: xhduan@fmmu.edu.cn

【Abstract】Objective To explore the clinical and imaging characteristics of patients with regional odontodysplasia accompanied by hypodontia and to provide a reference for clinical diagnosis and treatment. **Methods** This report presents the imaging manifestations, diagnosis, and treatment of a case of regional odontodysplasia (RO) accompanied by hypodontia. It includes a retrospective summary of the dynamic changes in the imaging characteristics of the affected teeth



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【作者简介】 张贝贝, 医师, 硕士研究生在读, Email: 1438638478@qq.com

【通信作者】 段小红, 主任医师, 教授, 博士, Email: xhduan@fmmu.edu.cn

over a 5-year period, along with a comparative analysis of the literature. The patient was a 9-year-old female who presented to the Clinic of Oral Rare and Genetic Diseases of our hospital with the chief complaint of “discomfort for over seven months following the extraction of the teeth in the left mandibular region.” Based on her clinical manifestations and imaging findings, she was diagnosed with RO in the left mandible and with hypodontia of permanent teeth 12 and 34. A treatment plan was formulated, and long-term follow-up was conducted. The current radiographic images were compared with previous imaging data to summarize the developmental changes in her teeth, and a comparative analysis was also performed with the literature to identify similarities and differences with previously reported RO dental characteristics. **Results** During the follow-up period, the patient’s symptoms did not worsen, and a conservative observation approach was adopted; the treatment plan was decided after the eruption of the affected teeth. By comparing and analyzing imaging data from three ages (4.5, 8.5, and 9 years old), it was determined that the deciduous and permanent teeth in the left mandible of this patient exhibited typical “ghost” radiographic features, alongside hypodontia of teeth 12 and 34, as well as the delayed development of tooth 35. A literature review and analysis indicated that RO manifests clinical characteristics such as delayed tooth eruption, reduced tooth size, and yellow crowns, along with typical “ghost” radiographic appearances. Treatment requires a personalized approach based on the patient’s specific condition. To date, only five cases of RO patients with hypodontia have been reported, while the delayed development of permanent tooth buds has not yet been documented. **Conclusion** For patients with RO, dynamic imaging evaluation plays a critical role in early diagnosis. RO is associated with hypodontia and delayed tooth germ development. Long-term follow-up and personalized treatment plans are the key to RO treatment.

【Key words】 regional odontodysplasia; “ghost” teeth; odontogenesis; hypodontia; tooth eruption; imaging; cone beam CT; deciduous dentition; mixed dentition

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区域性牙发育不良 (regional odontodysplasia, RO), 又称局部牙发育不全^[1], 是一种罕见的同象限内数颗牙发育不良, 可同时累及患牙的外胚层和中胚层相关组织, 其特征性的影像学表现为髓腔宽大, 根尖孔开放, 牙釉质及牙本质层薄且密度降低、釉牙本质界不清, 又称“鬼影”牙^[2]。RO病例首次报告于1934年, 目前文献报道的病例不到300例^[3]。RO同时伴发先天缺牙属于罕见现象, 目前仅有5例报道^[4-8]。本文通过对1例伴发先天缺牙的RO患儿3个不同时期乳牙和恒牙的影像学特征回顾, 结合文献总结该病的发生和发展规律, 探讨该病的适宜诊治方法。

1 临床资料

1.1 基本信息

患者女性, 9岁, 因“左下后牙拔除后不适7月余”于本院口腔遗传病与罕见病门诊就诊。患者就诊7个月前因左下后牙疼痛行左下第二乳磨牙及第一恒磨牙拔除术, 术后偶感不适, 为明确诊治, 遂来本科就诊。患儿否认系统性疾病及传染

病史, 否认家族史。

1.2 就诊经历

经问诊, 患者就诊经历如下: ①2019年4月因“牙齿外形粗糙”于外院就诊并拍摄曲面断层片, 具体诊治不详; ②2023年4月因“左下后牙疼痛”于我院就诊, 建议其拔除左下颌区松动患牙; ③2023年5月患者于外院行左下第二乳磨牙及第一恒磨牙拔除术; ④本次就诊。

1.3 临床检查

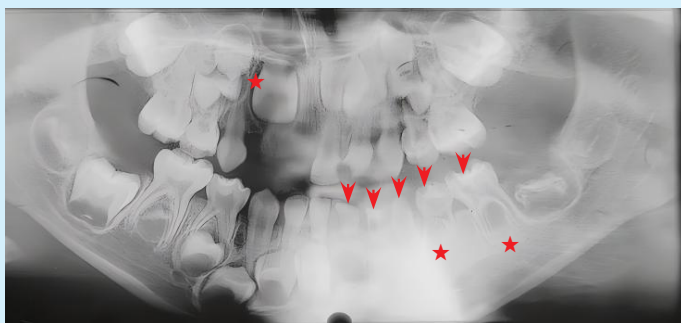
口外检查可见颌面部大致对称无畸形。口内可见混合牙列: 11、53-55、16、21、22、63-65、26、31、41、42、83-85、46; 牙列缺损: 上颌牙列12缺失、左下颌区仅见31且31牙齿体积小、萌出高度不足、表面粗糙、色黄, I度松动; 左下颌后牙区牙槽嵴较窄, 75、36牙拔牙创愈合良好, 其余无明显异常, 口腔卫生一般。

1.4 影像学检查

患者家长提供的既往影像学资料: ①2019年4月(患者4.5岁)的曲面体层X线片: 乳牙列期, 71-75牙呈低密度影像、牙体较小、牙釉质牙本质界不

清、呈“鬼影”状;12恒牙胚缺失;患区31-33恒牙胚发育迟缓且呈空壳状,34恒牙胚疑似缺失,35恒牙胚未见(图1)。②2023年4月(患者8.5岁)于我院拍摄的锥形束CT:替牙列期,12先天缺失;73、74过早脱落、75牙根大部吸收;31、32牙体低密度影

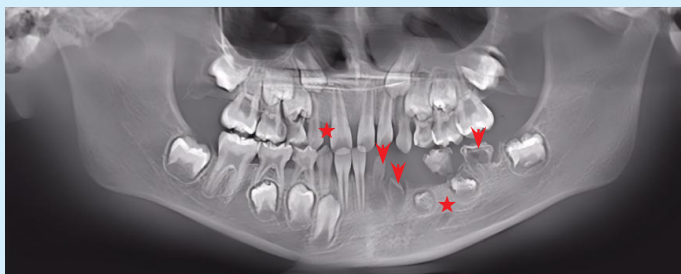
像、呈“鬼影”状,牙根发育不足,萌出高度不足;34恒牙胚缺失;33、35、37恒牙胚发育迟缓、体积小、呈低密度影像;36牙体积小、表面形态不规则、釉牙本质界不清、牙髓腔扩大、牙根吸收、呈“鬼影”状(图2)。



Panoramic X-ray of the primary dentition taken at 4.5 years of age revealed a “ghost” appearance of the deciduous teeth and permanent tooth buds in the left mandibular region (indicated by arrow); the permanent tooth buds for teeth 12, 34, and 35 were not observed(indicated by pentagram)

Figure 1 Radiographic image of the patient at 4.5 years of age showing regional odontodysplasia accompanied by hypodontia (April 2019)

图1 区域性牙发育不良伴发先天缺牙患者4.5岁时影像学图片(2019年4月)



Cone beam computed tomography (CBCT) scan taken at 8.5 years of age revealed that the teeth in the left mandibular region appeared as a “ghost” (31, 32, 36) (indicated by arrow). The permanent tooth buds (33, 35) exhibited rough contours and inadequate root development; neither permanent tooth 12 nor the permanent tooth bud for tooth 34 was observed (indicated by pentagram)

Figure 2 Radiographic image of the patient at 8.5 years of age showing regional odontodysplasia accompanied by hypodontia (April 2023)

图2 区域性牙发育不良伴发先天缺牙患者8.5岁时影像学图片(2023年4月)

本次就诊(2023年12月,患者9岁)拍摄的锥形束CT显示:75和36拔牙创口恢复较好,周围骨质无明显异常(图3)。

1.5 诊断

区域性牙发育不良(左下颌)、12、34恒牙胚先天缺失。



Cone beam CT scan taken at 9 years of age revealed that, in comparison to Figure 2, teeth 75 and 36 were absent, and there are no significant abnormalities in the surrounding bone tissue.

Figure 3 Radiographic image of the patient at 9 years of age showing regional odontodysplasia accompanied by hypodontia (December 2023)

图3 区域性牙发育不良伴发先天缺牙患者9岁时影像学图片(2023年12月)

1.6 诊断依据

①发育不良的牙局限在左下颌象限内;②影像学显示该象限内乳牙及恒牙胚体积小、轮廓粗糙、呈典型“鬼影”状、牙根发育不足。

1.7 治疗计划及过程

综合考虑患儿的现病史、牙槽骨和颞下颌关节的发育、咀嚼、美观和心理健康等诸多因素的情况下,制定以下治疗计划:①目前暂不处理未萌患牙,定期随访,观察患牙萌出情况,并对牙槽骨及颞下颌关节等的发育进行动态评估检测。②若后期恒牙萌出且达到一定高度、稳固性佳,则配戴预成冠以保护牙齿外形、提高咀嚼功能、改善咬合关系,保护牙齿进一步发育。③观察期间若患儿症状加重或牙齿萌出后稳固性不佳则建议其在全麻下拔除异常牙齿以降低继发感染的可能性,并于后期制作局部义齿恢复患儿咀嚼功能;长期随访,待颌骨发育完善后行永久性义齿修复治疗。

患儿家长同意继续观察,故暂不处理患牙并嘱其定期复诊。

2 结果

2.1 治疗结果

对患者进行定期随访1年余,近期随访过程中患者家长表示患者症状无加重,选择继续观察患区牙齿及牙槽骨发育情况,待患区牙齿萌出后按原计划进行治疗。

2.2 影像学资料回顾分析

为明确该患者5年内的牙齿发育变化,对其健侧同名牙影像学特征进行对比(见表1&表2),对区域性牙发育不良伴发先天缺牙的乳牙列和恒牙列患牙特征总结:①乳牙列:牙发育不良;牙冠体积小,牙根发育不足,髓腔和根管宽大,牙釉质和牙本质难以区分,呈“鬼影”状;易脱落、早失。②恒牙列:牙发育不良;牙冠体积小,牙根发育不足,髓腔和根管宽大,牙釉质和牙本质难以区分,呈“鬼影”状;牙发生延迟;牙萌出延迟或延迟到达咬合面。

2.3 既往相关文献复习结果

为进一步比较本病例与文献报道的RO特征异同点,笔者查询了178篇文献(1949年—2025年),收集到108篇有效文献,总结了6篇4岁~9岁患者的详细影像学特征^[9-14],结果显示:所有患者均有牙小、色黄、表面粗糙等临床表现和典型的“鬼影”状影像学表现;部分有乳牙早失(5/6)和迟萌(3/6)的表现;少数(2/6)描述有牙根发育不足的表现;但3个乳牙列患儿均无恒牙胚发生延迟的详细描述(表3)。

3 讨论

3.1 RO的临床及影像学特征

RO通常局限于1个象限内,少数病例累及多个象限^[15-16]。临床上通常根据患者典型的牙体积

表1 区域性牙发育不良伴发先天缺牙患者4.5岁时健侧同名牙特征对比(2019年4月)

Table 1 Comparison of the characteristics of the same tooth on the healthy and affected sides in the patient with regional odontodysplasia and hypodontia at 4.5 years of age (April 2019)

Tooth position	Healthy side	Affected side
I - V	The crown was moderate in size and complete in appearance, the pulp cavity was moderately sized, and a small amount of resorption was observed at the tips of the roots of I and II	The crown was small and had a rough surface, the pulp cavity was wide, and the tooth was “ghost” shaped
1, 2	The crown and part of the tooth root had formed	The crown had formed but was small, and the root was not obvious
3	The tooth germ was located at the root of III. The crown of the tooth germ had formed, and the enamel and dentin could be distinguished	A small amount of cusp tissue was visible
4	The tooth germ was located at the root of IV. The crown of the tooth germ had formed, and the enamel and dentin could be distinguished	No tooth germ
5	The tooth germ was located on the root side of V, a small amount of tooth cusp was formed, and the enamel and dentin could not be distinguished	No tooth germ
6	The tooth was close to eruption, the crown and a small amount of root were formed, and the enamel and dentin were highly differentiated	Crown formation, atypical appearance, “ghost” shape
7	The cusps were formed	Very little cusp tissue was formed

表2 区域性牙发育不良伴发先天缺牙患者8.5岁时健患侧同名牙特征对比(2023年4月)

Table 2 Comparison of the characteristics of the same tooth on the healthy and affected sides in the patient with regional odontodysplasia and hypodontia at 8.5 years of age (April 2023)

Tooth position	Healthy side	Affected side
I	Had been replaced by tooth 41	Had been replaced by tooth 31
II	Had been replaced by tooth 42	Had been replaced by tooth 32, but tooth 32 had not erupted
III	The root began to resorb	Had fallen off
IV	Not loose	Had fallen off
V	Not loose	Tooth defects and looseness (+++)
1	Emerged and reached the occlusal plane	Insufficient eruption height, “ghost” shape, tooth root resorption, and periapical shadows
2	Emerged and reached the occlusal plane	Delayed eruption, “ghost” shape, and incomplete tooth roots
3	The crown and 1/3–1/2 of the root were formed, and the enamel, dentin, and pulp were clear	A small amount of crown tissue was formed, which was irregular with low density
4	The crown and 1/3–1/2 of the root were formed, and the enamel, dentin, and pulp were clear	No tooth germ
5	The crown and 1/3–1/2 of the root were formed, and the enamel, dentin, and pulp were clear	The crown had formed, but it was small and exhibited a rough appearance. The density of the enamel and dentin was low, making them difficult to distinguish, and the structure of the pulp was not clearly defined
6	The crown and root had completed development, and the tooth had erupted and established an occlusal relationship with the opposing tooth	The tooth had erupted but was lower than the occlusal plane; the crown was small and had a “ghost” shape, and the tooth root was resorbed
7	The crown and a small amount of root had formed, and the enamel and dentin were highly differentiated	The crown and part of the root were formed, the surface of the crown was rough, and the density of the enamel and dentin was low and difficult to distinguish

表3 既往文献区域性牙发育不良牙齿影像学特点总结

Table 3 Summary of dental imaging characteristics of regional odontodysplasia in previous literature

Case	Age/years	“Ghost” shape	Premature loss of deciduous teeth	Delayed emergence	Insufficient tooth root development	Delayed odontogenesis of permanent tooth germ	References
1	5	(+)	(+)	(–)	N	N	[9]
2	5	(+)	(+)	(–)	N	N	[10]
3	5	(+)	(+)	(+)	N	N	[11]
4	8	(+)	(+)	(+)	(+)		[12]
5	8.5	(+)	(+)	(+)	(+)		[13]
6	9	(+)	(–)	(–)	N		[14]

The presence or absence of this phenotype is represented by (+) and (–) respectively; N: no detailed description

小,牙冠色黄、表面粗糙且易磨耗等临床表现和特征性的“鬼影”状影像学表现来确诊RO。本病例中患牙局限在左下颌区,其中31具有牙体积小、色黄、表面粗糙和萌出高度不足等临床表现,各时期影像学资料显示该区的乳牙和恒牙均表现出釉牙本质界不清、牙髓腔扩大等典型的“鬼影”状特征,诊断明确。

本研究通过对患儿3个不同时间点的影像学资料进行详细分析,发现本例患儿除具有上述RO典型的临床表现和影像学表现外,还具有以下特

点:①牙发生延迟。经文献回顾该特点为首次报道。以下颌第一和第二恒前磨牙为例,患儿4.5岁时其健侧已经有这两颗牙的影像,但患侧未见。牙的发生发育、矿化和萌出具有一定的时空特征。在本病例中,从最早萌出的下颌乳中切牙,到较早萌出的下颌恒中切牙、第一恒磨牙,以及最后萌出的第二恒磨牙等,一旦处于受累象限,则无一幸免。提示这种病变的发生跨度时间长,在胚胎发生早期开始,一直延续到出生后很长一段时间,具有很强的时空调控特征。②RO伴发先天缺牙。目

前仅少数RO患者伴发先天缺牙,具体病因不明。Koskinen等^[5]报道1例家族性少牙症的RO患者存在配对盒基因9(paired box 9, PAX9)突变,认为RO可能与该基因突变有关。多种基因的突变和环境因素的影响会导致先天缺牙^[17-18]。RO伴发先天缺牙的病因和机制有待进一步研究。

3.2 RO的病理学特征

RO在组织病理学上表现为牙釉质棱柱不规则^[19],成牙本质细胞变性,牙本质小管排列紊乱,牙髓钙化^[20],患牙周围的牙囊细胞呈螺旋状排列形成钙化螺旋结节(calcifying whorled nodules, CWN)^[21]。以上细微结构的改变最终导致本病例患儿左下颌区牙齿呈现出表面粗糙不规则、釉牙本质界不清等影像学特点。

3.3 RO的鉴别诊断

通过明确RO的上述特点可以与遗传性牙釉质发育不全(amelogenesis imperfecta, AI)、牙本质发育不全(dentinogenesis imperfect, DI)、节段性颌发育不良(segmental odontomaxillary dysplasia, SOD)等牙齿发常疾病相鉴别。AI几乎累及全口牙齿,患牙表现为棕色、表面有带状或窝状凹陷等特点^[22-23];DI患牙表现为棕色或灰蓝色,牙颈部紧缩,根细等特点^[24-25];SOD具有牙萌出延迟、牙吸收、牙本质发育不良、颌骨发育不良、软组织肿胀等多个特点^[26-27]。

3.4 RO的治疗

目前对RO首选治疗方法尚无共识,大多数病例以拔除患牙告终^[28]。部分研究人员提出RO治疗目标为短期内通过定期涂氟、龋洞充填等预防已萌患牙感染,同时长期监测对颌牙的萌出高度及未萌患牙的发育情况^[29-30];近年来多数学者认为需根据患者的年龄、病史、受累牙的畸形程度、有无感染及患者和家长的诉求,进行个性化评估治疗^[31-32]。对7岁患儿使用Essix保持器以保持美观和咀嚼功能,维持正常的垂直尺寸,有利于牙槽骨发育及后期修复^[33]。近期有研究人员报道对一例13岁RO患者的早期恒牙进行牙髓血运重建可促进牙根继续发育且有利于患牙根尖周病变的消退^[34]。对替牙期患儿拔除患牙后使用自体牙移植可实现牙槽骨与相邻牙齿同步生长并形成正常的牙齿间隙,同时适应功能刺激^[8]。

综上,对RO患者明确诊断后,应结合患者状况,综合考虑咀嚼、发育、发音、美观、心理等需求,多学科诊疗制定个性化治疗方案。此外应加强对

RO病因学的研究并探索新型治疗方法,为该类患儿提供更早期、短程、全面的治疗。

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