Case Report *รายงานผู้ป่วย*

Dental Management for Recessive Dystrophic Epidermolysis Bullosa Child-Patient

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Abstract Epidermolysis bullosa is a group of rare hereditary skin disorders characterized by the formation of bullae on the skin and mucous membranes. Blisters arise spontaneously and easily at points of trauma. Epidermolysis bullosa is classified into three main groups based on anatomical location and distribution of the lesions. Dystrophic epidermolysis bullosa, one of three subtypes, is characterized by blister formation occurring beneath the entire demo-epidermal junction (sublamina densa) and divided into two types, dominant and recessive, according to mode of autosomal inheritance. Recessive dystrophic epidermolysis bullosa has two subtypes, localized and generalized. In generalized form, bullae becomes blister and heals with severe atrophic scarring and contracture.The character and extent of oral involvement in generalized recessive dystrophic epidermolysis bullosa are more severe than other types. Subsequent scar formation causes alteration of soft tissue texture, obliteration of oral vestibule, ankyloglossia and microstomia. The teeth are prone to dental caries. This is a case report of a child-patient with recessive dystrophic epidermolysis bullosa who had dental problems. Dental management such as teeth extractions and restorations were performed under local anesthesia as well as preventive measures were rendered to improve his oral health. Complications, precautions, and some problems in this case were discussed.

Key words: **child-patient, dental management, recessive dystrophic epidermolysis bullosa**

Introduction

Epidermolysis bullosa (EB) is a group of rare hereditary skin disorders characterized by the formation of bullae on the skin and mucous membrane. Blisters arise spontaneously and easily at points of trauma.[1,2] EB encompasses many clinically distinctive disorders which share three major features: genetic transmission, mechanical fragility of the skin and blister formation.[3] EB is classified into three main groups based on anatomical location and distribution of the lesions.The main groups are epidermolysis bullosa simplex (EBS), junctional EB (JEB), and dystrophic

EB (DEB).[1-3] Dystrophic or scarring form (DEB) is characterized by blister formation occurring beneath the entire demo-epidermal junction (sublamina densa)[4] and divided into two types, dominant and recessive, according to mode of autosomal inheritance.[1,2,5,6] Both recessive and dominant forms of DEB result from mutations in the type VII collagen gene.[7] Collagen type VII is the major component of the anchoring fibrils which play an integral part in the structural integrity of the anchoring complex of the dermal-epidermis junction by attaching the lamina densa of the basement membrane to the underlying papillary dermis. The anchoring fibrils can be morphologically altered, reduced in number, or entirely absent in DEB.[8] Recessive DEB has two subtypes; localized and generalized DEB.[2,9]

The incidence of a various type of EB occurs in one to two people per 100,000[10]. In Thailand, EB was annually found in one to two patients of the average 49,000 new patients at Institute of Dermatology, Ministry of Public Health.[2] In the United States, the incidence of DEB has been estimated at 5.7 per million live births with the severe recessive DEB having an incidence of 2.0 per million live births.[8]

Generalized recessive DEB is characterized by wide spread bullae involving the skin and mucosa. Bullae becomes blister and heals with severe atrophic scarring and contracture. Cutaneous scarring may lead to a variety of complications, particularly to joint contractures, and to a fusion of the fingers and toes.[1] Digital fusions and mitten deformities of hands and feet are secondary to repeated episodes of blistering and scarring as well as flexural contractures of knees, elbows, wrists, and short stature secondary to malnutrition[6]. Moreover, blistering of the oral, pharyngeal and easophageal mucosa which are common in this type of DEB may caused dysphagia, submucous fibrosis in the oral cavity, contraction of the oral and pharngael opening and fixation of the tongue[1]. Extremely painful erosions cause patients great difficulties in chewing and swallowing food. Dental and gingival conditions become very poor due to these suffering hindrances and duly inappropriate cleansing. The character and extent of oral involvement in this type of EB are more severe than other types[11]. The entire oral mucosa is affected by continuous blistering. Subsequent scar formation causes alteration of tissue texture, obliteration of oral vestibule and ankyloglossia. Microstomia which is most profound in severe type[11] apparently results from chronic, severe intraoral blistering with subsequent scar formation.While the teeth are usually structurally normal when comparing with junctional EB which enamel hypoplasia is more prevalent, they are prone to severe dental caries[12,13]. The absence of normal physical cleansing due to restriction of tongue movement and lips, the shallow vestibule, contributes to food retention. Tooth brushing is difficult because blistering of the hands and fingers causes contraction reducing the ability to grip and hold a toothbrush[13]. Effective oral health care is impossible when painful blistering develops in the oral cavity. Continuous blistering may produce tissue change with malignant potential. Carcinoma are often associated with the generalized recessive dystrophic form[11].

As this kind of diseases affect at the early age, dental problems are neglected. Most patients have suffered from severe dental problems and need extensive dental treatments. Delicate cutaneous and mucosal tissue challenge dentists in providing dental management and best oral care. The purpose of this report is to present the dental management for a recessive dystrophic epidermolysis bullosa child-patient.

Case Report

A 12-year-old Thai boy was referred to Dental Department, Rajavithi Hospital for an emergency treatment of toothache in May 2001.

Medical History

He was diagnosed as generallized recessive dystrophic epidermolysis bullosa and was admitted to Institute of Dermatology due to recurrent infected skin blisters.

Physical examination

The patient looked pale, thin and could not stand straight. He had atrophic skin with erythematous plague and dry scale on his trunk and extremities (fig. 1). He also had contracture of the fingers (fig. 2) and his both feet were mitten-like (fig. 3) due to repeated blistering and scarring. There were less plaque and scale on his face than his body. He had dry lips and no restricted oral opening.

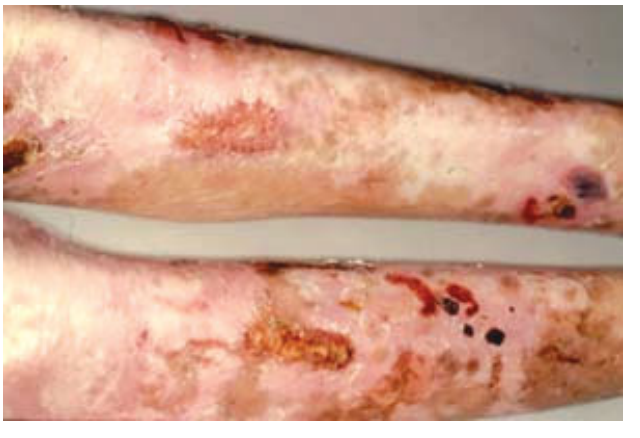

Fig. 1 Atrophic skin with erythematous plaque and dry scale on his both legs

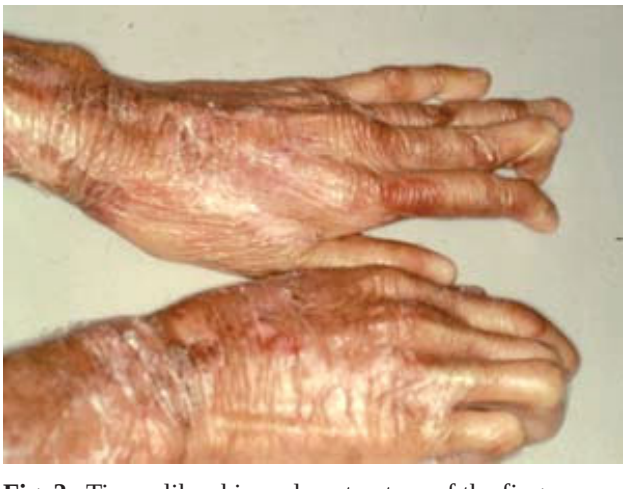

Fig. 2 Tissue-like skin and contracture of the fingers

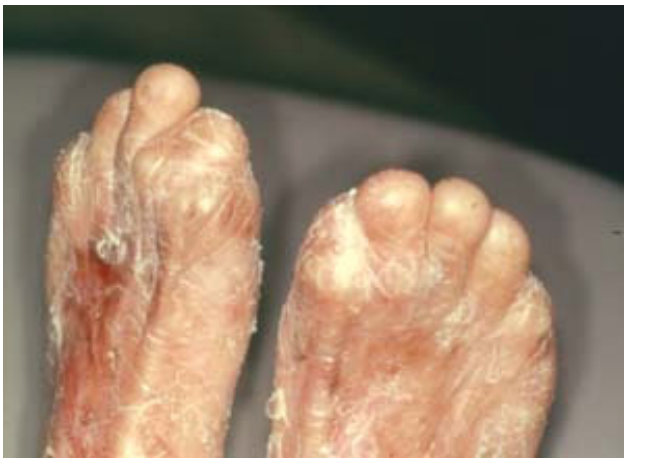

Fig. 3 Mitten-like feet due to repeated blistering and scarring

Intraoral examination

Intraoral examination revealed that blisters and erosions were seen over buccal mucosa, tongue, hard palate,gingiva and inner lips epidermis (fig. 4). Some blisters were over 10 mm. in diameter. The tongue was smooth due to complete lost of papillae. Erosive lesions were found at dorsum and lateral borders of the tongue (fig. 5). Tongue movement was limited due to ankyloglossia. Labial and buccal vestibules were shallow. Heavy plaque and white spot lesions were evident at all visible surfaces of all teeth (fig. 6). The gingiva was generalized inflamed. Both maxillary canines were labio-version and caused irritation to the inner surface of the upper lip. Multiple dental caries were detected, especially at cervical and proximal surfaces of anterior and posterior teeth (fig. 7). The lower left first molar (#36) that he had complained of toothache was carious. The examination showed that the pulp was exposed by a large carious lesion at buccal cervical area with no intra-oral swelling or tooth

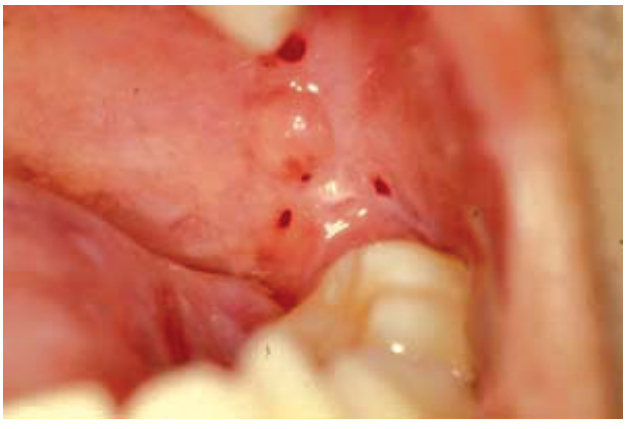

Fig. 4 Large bullae at the oral mucosa and erosive ulcers at lateral border of the tongue

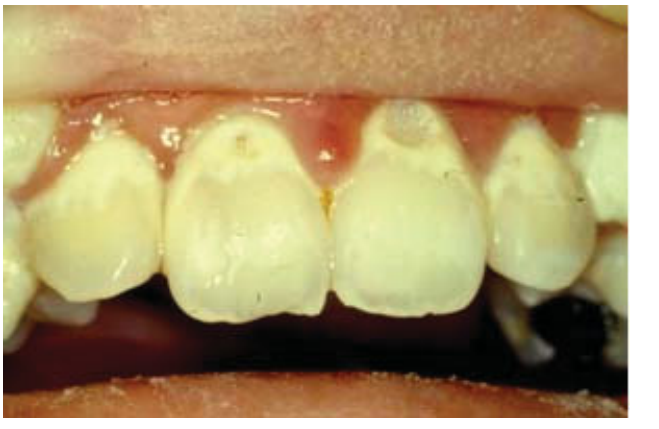

Fig. 7 Visible white spot lesions and dental caries at cervical area of upper anterior teeth plus gingival inflammation due to plaque accumulation

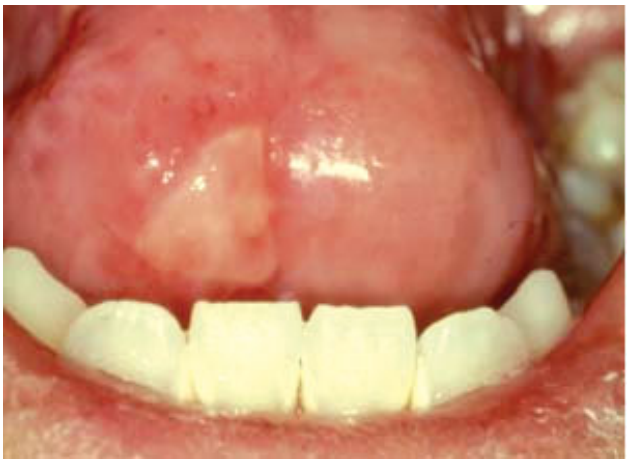

Fig. 5 Erosive ulcer at the tongue which was smooth due to complete loss of papillae

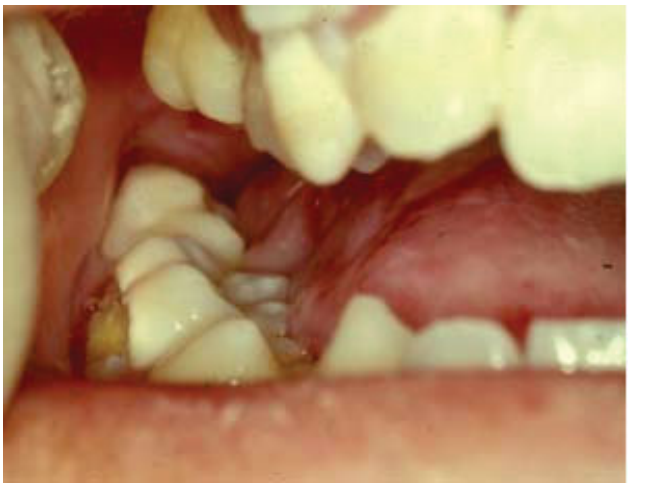

Fig. 8 Deep carious lesion at cervical area of lower right first mandibular molar

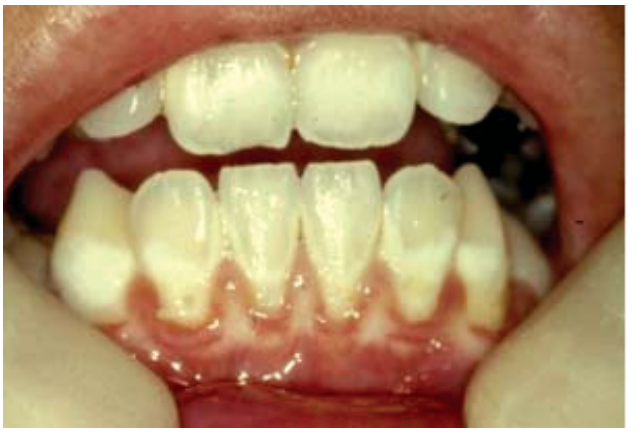

Fig. 6 White spot lesions on lower anterior teeth and shallow labial vestibule

mobility presented.

Radiograhic examination

Because of mucosal fragility and blisters easily developed from mechanical trauma, intraoral radiographs could not be taken.

Diagnosis

Generalized recessive dystrophic epidermolysis bullosa with dental problems;

1. Irreversible pulpitis on #36, #46
2. Dental caries on #11, #12, #14, #16, #21, #22, #24, #25, #26, #31, #32, #41, #42
3. Generalized gingivitis

Treatment plan

1. Medical consultation
2. Emergency treatment; extraction on # 36
3. Oral disease control;
   - 3.1 Prevention;
     - Patient education
     - Oral hygiene instruction; tooth brushing
     - Chlorhexidine and fluoride mouthrinses
     - Sealant on # 34, #35, #37
     - Topical fluoride application
   - 3.2 Periodontal treatment
     - Full mouth scaling
   - 3.3 Restorative treatment;
     - Amalgam fillings on #24 MOD, #25 MOD, #26 OM
     - Composite fillings on 11 DLi, MLi, La, #12 DLa, MLaLi, #21 MLi, DLi, La, #22 MLi, #31 DLi, #32MLa, #41 DLa, #42 MLa
     - Preventive resin restoration on #14 O, #16 O
   - 3.4 Surgical Treatment;
     - Extraction on # 46
4. Periodic recall

Precaution

Due to mucosal fragility, oral manipulation should be gently done with patience to avoid subsequent oral blisterings and ulcers.

Treatment

Comprehensive treatments and preventive care were scheduled to improve his oral health. After consultation with his attending dermatologist, the lower right and left mandibular first molar (#46, #36) were removed. Three upper left posterior teeth (#24, #25, #26) were filled with amalgam whereas upper (#11, #12, #21, #22) and lower anterior teeth (#31, #32, #41, #42) were restored with composite resin. Preventive resin restorations were performed at small cavities on two upper right posterior teeth (#14, #16). Scaling with topical fluoride application and sealants were performed. Oral hygiene instruction such as tooth brushing with small soft-bristled brush was reinforced to the patients. Chlorhexidine and fluoride mouthwashes were prescribed for daily rinse. The treatment continued after he was discharged and took eight visits to complete in September 2001. His revisits were scheduled monthly three times and every three months afterwards.

Treatment outcome

Dental condition was improved but gingival inflammation and plaque accumulation still existed. Tooth brushing and mouth rinsing were reinforced. Oral prophylaxis and topical fluoride application were performed every recall visits. However, this patient lost a check-up visit six months later.

Discussion

Dental rehabilitation and anesthetic management of EB patients are dependent on the specific manifestation and type of disease[14]. This generalized recessive dystrophic EB subtype has the most soft tissue involvement, oral blistering is not only most prevalent but also most severe[15]. Most patients who suffered from this disorder are not brought to the dentist for dental care at the early age and blisters and ulcer lesions discouraged oral cleansing, rapid severe dental caries onset was observed in precooperative-age children[10]. The state of soft tissue involvement such as presence of bullae, fragile oral mucosa and microstomia complicates the ability to render both preventive and restorative care. Tooth extraction under gen-

eral anesthesia is the only practical option for severely carious teeth[1].

Due to mucosal fragility, dental treatment for patients with this subtype of EB can precipitate oral blistering[16] and require special approach. This is often overcome with patience, gentle oral manipulation and careful technique[17]. When manipulating oral tissues which are most prone to mucosal blisterings, only compressive forces should be applied because these are less likely than lateral traction or other shear forces to induce tissue separation[18]. When more blisters and erosion developed in oral cavity, even mouth opening could cause severe pain, any dental treatment had to be postponed until the patient recovered. Oral mucosal pain associated with dental treatment can be reduced by minimizing trauma and the use of palliative rinses such as a 1:1 mixture of a mucosal coating agent -magnesium hydroxide- plus a local anesthetic such as diphenhydramine. Alternately, topical application of 2% viscous xylocaine or benzocaine gel to affected areas may provide relief[19].

Extraction and restorative treatments in this case were performed under local anesthesia. This patient could not tolerate sensitive and painful treatments and anesthesia was also beneficial to ease pain of surrounding soft tissue. Gentle retraction of lips, cheek and tongue had to be done to avoid aggravating pain and trauma which might precipitate blisters later.

Comprehensive dental care for pre-cooperative children and for patients with severe soft tissue involvement are best managed with general anesthesia[17,20]. Since this patient was preadolescent and cooperative, local anesthesia could be administered although it eventually caused bullae at the site of injection. Wright et al.[18] recommended that when administering intraoral anesthesia, the anesthetic solution should be injected deeply into the tissue slowly enough to prevent tissue distortion, which may cause mechanical tissue separation and blistering. Nerve blocks are far less likely to form blisters since they do not place the mucosal surface under pressure by depositing a bolus of fluid near the tissue surface. To the author's experience on this case, even the sharp point of small guage needle (no. 30) could not easily push through the tough mucosa of which the texture gradually changed from scarring. Infiltration technique at some areas that comprise loose tissues, for example, upper and lower anterior teeth sometimes caused haemorrhagic large bullae at the injection site immediately. However this bullae would rupture spontaneously and left painful erosive lesion. This patient reported that he had ever used a needle to rupture blister himself. However he was informed to use a sterile needle or else he would be infected.

Preventive strategies[19] such as oral hygiene instructions, diet counseling, tooth brushing with small soft-bristled toothbrush, pit and fissure sealant, antimicrobial rinses, and home fluoride application are key for long-term success, but will require patient education and compliance. For this patient he can brush his own teeth in spite of his deformed hands but the effectiveness of tooth brushing depends on the prevailing condition of the lesions in his mouth and on his hands. Heavily flavored or alcohol-based fluoride rinses and topical applications are often not well accepted in patients with substantial oral involvement[18]. Neutral sodium fluoride topical application and nonalcohol-based rinses may prove to be effective[12]. Chlorhexidine mouthrinse was once prescribed to control cariogenic microorganisms but he was not able to tolerate the flavor. Even oral hygine instruction was encouraged as well as preventive measures, plaque accumulation still presented, so frequent dental checkup was scheduled. Since EB is an incurable and sometimes lethal disorder, any therapy is palliative and nonspecific. Dental treatment aims only at improving oral health and extent of care depends on individual patient[21], bearing in mind that improving quality of life

for these patients is an ultimate goal of health care providers.

Conclusion

Recessive dystrophic epidermolysis bullosa was a rare hereditary skin disorder characterized by wide spread bullae on skin and mucosa. This subtype of epidermolysis bullosa had the most severe soft tissue involvement, intraoral soft tissue morphology and mobility were drastically changed. A case of a child with recessive dystrophic epidermolysis bullosa was present. Dental management and preventive measures were performed to improve the oral health.

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บทคัดย่อ **การรักษาทางทันตกรรมในผู้ป่วยเด็กที่เป็นรีเซสซิฟดิสโทรฟิคอิพิเดอร์โมไลซิสบูลโลซา: รายงานผู้ป่วย**

เกศวดี ทรัพย์แสนดี

กลุ่มงานทันตกรรม โรงพยาบาลราชวิถี

วารสารวิชาการสาธารณสุข **2551; 17:SVI1559–66.**

อิพิเดอร์โมไลซิสบูลโลซาเป็นกลุ่มอาการของโรคผิวหนังที่ถ่ายทอดทางกรรมพันธุ์ที่พบได้ยากชนิดหนึ่ง มีลักษณะเป็นตุ่มน้ำเกิดขึ้นตามผิวหนังและเยื่อบุ ตุ่มน้ำพองจะเกิดขึ้นได้ง่ายตามร่างกายที่มีการกระทบกระแทกหรือเสียดสีเพียงเล็กน้อย ซึ่งแบ่งเป็นสามกลุ่มใหญ่ตามตำแหน่งทางกายวิภาคของตุ่มน้ำและการกระจายของรอยโรค ดิสโทรฟิคอิพิเดอร์โมไลซิสบูลโลซาเป็นกลุ่มโรคย่อยกลุ่มหนึ่งที่มีตุ่มน้ำพองเกิดขึ้นในตำแหน่งใต้รอยต่อของหนังแท้และหนังกำพร้า แบ่งเป็นชนิดถ่ายทอดทางกรรมพันธุ์ทางยีนลักษณะเด่นและลักษณะด้อย ชนิดลักษณะด้อยแบ่งแยกย่อยเป็นชนิดที่เกิดตุ่มน้ำในบางตำแหน่งและชนิดที่เกิดตุ่มน้ำทั่วไปตามผิวหนังและเยื่อบุ ชนิดที่เกิดทั่วไปนี้ตุ่มน้ำจะแตกเกิดแผลเป็นและมีการหดรั้งของผิวหนัง โรคชนิดนี้มีลักษณะของรอยโรคและอาการในช่องปากรุนแรงกว่าชนิดอื่น แผลเป็นที่เกิดขึ้นตามมาทำให้เกิดการเปลี่ยนแปลงความยืดหยุ่นของเนื้อเยื่อ การตื้นเขินของร่องข้างกระพุ้งแก้ม การยึดติดของลิ้นและการแคบลงของช่องปาก ผู้ป่วยโรคนี้มีความเสี่ยงที่จะเกิดฟันผุได้มาก บทความนี้เป็นรายงานผู้ป่วยเด็กหนึ่งรายที่เป็นโรครีเซสซิฟดิสโทรฟิคอิพิเดอร์โมไลซิสบูลโลซาและมีปัญหาสุขภาพในช่องปาก ผู้ป่วยได้รับการรักษาทางทันตกรรมเช่น การถอนฟัน การอุดฟัน โดยใช้ยาชาเฉพาะที่ และการให้ทันตกรรมป้องกันเพื่อให้ผู้ป่วยมีสุขภาพในช่องปากที่ดีขึ้น บทความนี้ได้กล่าวถึงผลข้างเคียง ข้อควรระวังและปัญหาที่ได้พบในการให้การรักษาผู้ป่วยที่เป็นโรคชนิดนี้

คำสำคัญ: **ผู้ป่วยเด็ก, การรักษาทางทันตกรรม, รีเซสซิฟดิสโทรฟิคอิพิเดอร์โมไลซิสบูลโลซา**