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From tooth extraction to Gorham-Stout disease: A case report

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ABSTRACT

INTRODUCTION: Gorham-Stout disease (GSD), or vanishing bone disease, is a very rare condition of unknown aetiology. It is characterised by progressive osteolysis and angiomatosis.
CASE PRESENTATION: We report the discovery of this very rare disease following a trivial deciduous tooth extraction in a 14-year-old female. We focus initially on the difference between the preoperative orthopantomography and the whole-body computed tomography and magnetic resonance images obtained post-haemorrhage, and then on the improvement of strategies for the correct diagnosis and treatment of this disease.
DISCUSSION: Bone loss and the proliferation of vascular structures can occur in a single bone or spread to soft tissue and adjacent bone; areas commonly affected by GSD include the ribs, spine, pelvis, skull, clavicle, and the maxillofacial area. The clinical presentation of GSD includes pain, functional impairment, and swelling, although a few asymptomatic cases have been reported, similar to our case.
CONCLUSION: We report a very rare case of this multicentric disease in an asymptomatic child who presented for dental extraction, almost died, and was then diagnosed with and treated for GSD.

1. Introduction

Gorham-Stout disease (GSD), also known as vanishing bone disease, phantom bone disease, disappearing bone disease, or massive osteolysis, is a very rare idiopathic bone disorder characterised by spontaneous and progressive osteolysis, associated with angiomatous proliferation and soft tissue swelling without new bone formation [1,2]. Its diagnosis is based on clinical, radiological, and histological features [3]. Only 200 cases of GSD have been reported since the first description in 1838, followed by Jackson's report in 1872 on the case of a young man with a gradually vanishing humerus [4,5]. The first facial case was described by Romer in 1924 [6], and in 1955 Gorham and Stout [7] defined a specific disease entity and reviewed 24 cases from the literature. Here, we describe a case of this very rare disease that was discovered during an intraoperative, potentially life-threatening complication that developed during a deciduous tooth extraction in a 14-year-old girl with the Pierre Robin sequence and deletion of the long arm of chromosome 4. The preoperative workup featured only orthopantomography [8].

2. Case report

A 14-year-old girl with the Pierre Robin sequence and deletion of the long arm of chromosome 4 was referred to the Outpatient Unit of the Department of Maxillofacial Paediatric Surgery, Children's Hospital, ASST degli Spedali Civili, Brescia, Italy. Physical examination revealed only mandibular hypoplasia with multiple deciduous teeth and caries; the patient was noted to be 'fragile' (syndromic child). Orthopantomography revealed mixed dentition with nothing of note in the mandible or maxilla (Fig. 1). The patient had no history of trauma, previous medication, gingival bleeding, blood transfusion, or elective surgery. The dentist planned the avulsion of tooth #85 to facilitate the eruption of permanent teeth and avoid the formation of an abscess. The decision was taken to perform the extraction and deliver the necessary tooth care in the operating room, following our protocol for the treatment of 'fragile' children. Surgical intervention was performed under sedation and local anaesthesia. During the mobilisation of tooth #85 with a dental elevator, massive bleeding (approximately 700 mL over the course of a few seconds), triggering tachycardia and hypotension, occurred. The next few moments were life threatening; however, the team responded admirably. The patient was intubated immediately after the surgeon had momentarily controlled the bleeding by compressing the bleeding area with a finger and aspirated the blood in the oral cavity because the airway was unprotected. An acute blood transfusion was administered to avoid hypovolaemic shock. The haemorrhage was subsequently controlled by the initial use of bipolar cautery, followed by the application of bone wax and fibrin glue.

Angio-computed tomography (CT) was performed on the intubated patient, with remarkable results. The horizontal branch of the mandible appeared to have expanded; the chin region, normally

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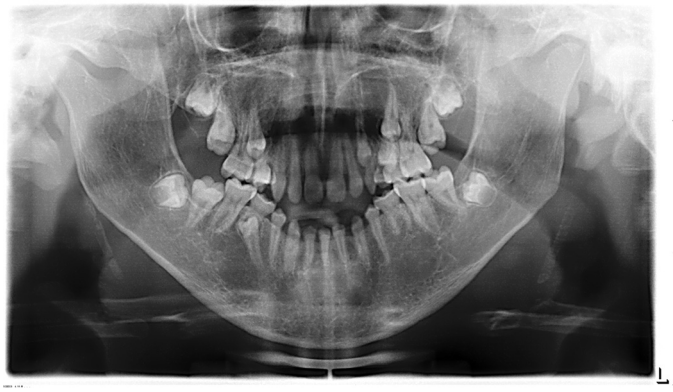

Fig. 1. The preoperative orthopantomogram.

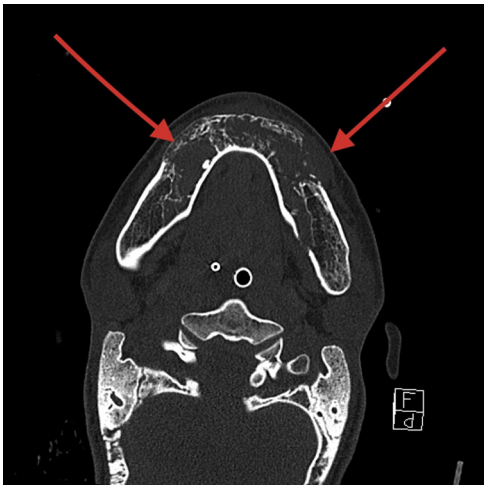

Fig. 2. The axial CT view of the mandibular lesion.

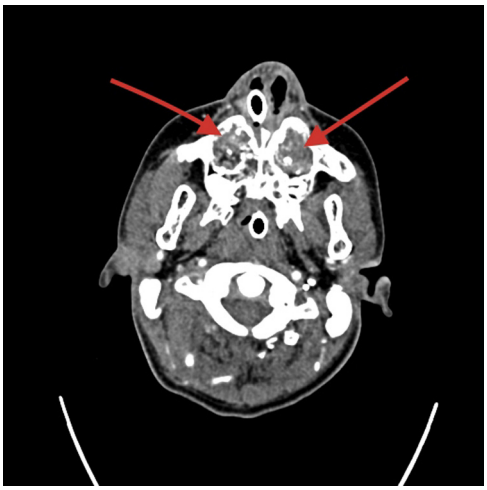

Fig. 3. The angio-CT axial view of the maxillary lesion.

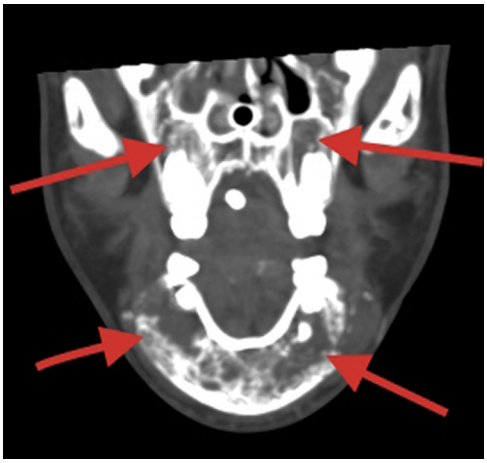

Fig. 4. The angio-CT coronal view of the mandibular and maxillary lesions.

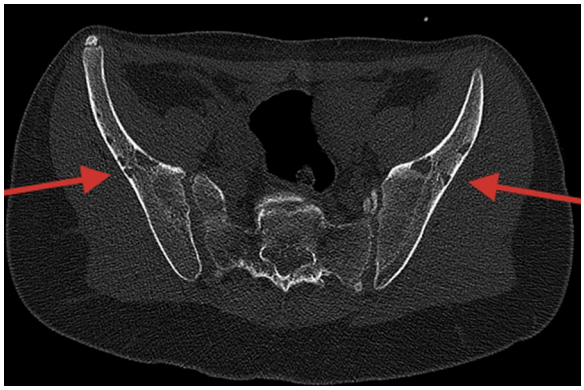

Fig. 5. The axial CT view of the small areas of osteolysis in the middle thirds of both iliac wings on the lateral and medial sides.

filled with cancellous bone, was composed entirely of vascularised tissue. Although the tissue was solid, it lacked the typical appearance of a high-flux arteriovenous malformation and was evidently a low-flux angiomatous formation. The outer cortical bone (between the premolars and left canines) was focally interrupted by solid, vascularised subcutaneous tissue. The expanded cortical bone exhibited marked thinning, and the solid vascularised tissue extended to the right mandibular angle. The maxilla also appeared to have expanded, containing similar, solid vascularised tissue, and even the sphenoid appeared to have partially expanded; the ethmoid was partially occupied by hypodense material (Figs. 2–4).

Two days later, the patient underwent contrast-enhanced maxillofacial magnetic resonance imaging (MRI), which revealed widespread structural subversion of the bones of the head and neck. Almost every bone was affected. The cancellous bony cores of the zygomatic and nasal bones, and those of the alveolar processes and horizontal branches of the jaw, were replaced by tissue with the signal characteristics, and the impregnation and bone development modes, of angioma. At several points, the resulting structural alterations had remodelled and disrupted the cortex. A Chiari type I malformation was also evident. To determine the radiological features of the facial skeleton, it was decided to perform CT of the pelvis and spine, whole-body MRI, and positron emission tomography (PET).

Pelvic and spinal CT revealed small areas of osteolysis in the middle thirds of both iliac wings on the lateral and medial sides, and showed that the cortical bone appeared to be interrupted for the normal entry of vessels. A similar 15-cm alteration was found along the rear edge of the intermediate and distal diaphyses of the right femur; on the right side, an isolated phenomenon of identical appearance affected the posterior region of the distal tibia (Fig. 5–7)

The PET scan enabled exclusion of the presence of hypermetabolic lesions in the area of explored bone volume.

Whole-body MRI, in addition to confirming the CT findings, also showed subcortical hyperintensity of the anterior and posterior dorsal spine and lumbar triangular areas on T2-weighted

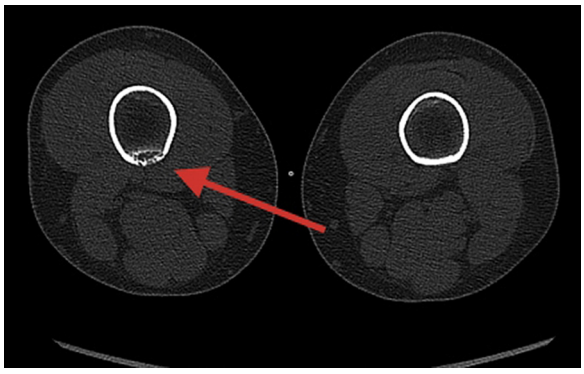

Fig. 6. The axial CT view of the small areas of osteolysis along the rear edge of the intermediate and distal diaphyses of the right femur.

sequences. These findings supported the first hypothesis of vascular deficiency.

The next step was to biopsy the lesion; in this case, the most important decision regarded selection of the best biopsy site to reduce the risk of bleeding. The ideal site for such a biopsy was determined to be the distal tibia. The patient underwent an incisional biopsy performed by an orthopaedic surgeon; no vascular complication was observed during this procedure.

Microscopically, the lesion showed haemangioma-like proliferation of thin-walled vessels with bone destruction. No malignant cell or cellular atypia was noted (Fig. 8). To reach a better diagnosis, immunohistochemical staining, including the use of D2-40 for specific lymphatic endothelial markers and CD31 and CD34 for vascular markers and vascular endothelial growth factor, was undertaken.

The histopathological and radiological findings, in association with the clinical aspect, finally revealed a diagnosis of GSD.

3. Discussion

According to Heffez et al. [9], eight inclusion criteria are used for the diagnosis of GSD: biopsy positivity for angiomatous tissue; absence of cellular atypia; minimal or no osteoblastic response and the absence of dystrophic calcification; evidence of local progressive osseous resorption; non-expansile, non-ulcerated lesion status; absence of visceral involvement; osteolytic radiographic pattern; and negativity for hereditary, metabolic, neoplastic, immunological, or infectious aetiology.

Hardegger et al. [10] classified five types of idiopathic osteolysis that together comprise a heterogeneous group of very rare diseases; this classification was based on reports by Torg et al. [11] and Macpherson et al. [12]. This classification system considers familiar and sporadic cases, as well as multicentric and unicentric osteolysis. GSD is considered to be a form of type IV osteolysis, according to the Hardegger [10] system.

The mechanism of osteolysis in GSD remains unknown and under debate. Gorham and Stout [7] proposed a trauma-induced change in pH and osteolysis caused by mechanical force. Heyden et al. [13] speculated that osteolysis results from the enhanced activity of local hydrolytic enzymes induced by local hypoxia and acidosis. Lala et al. [14] suggested that lymphatic vessels result in the pathogenesis of GSD. Fretz et al. [15] and Kulenkampff et al. [16] thought that angiomatosis was responsible. Thompson and Schurman [17] suggested that the disease is a primary aberration of vascular tissue in bone related to hyperemic granulation tissue, whereas Young et al. [18] believed that osteolysis is attributable to a basic underlying endothelial dysplasia of the lymphatics and/or blood vessels.

The disease occurs within a large spectrum of bones, such as the scapula [19,20], clavicle [21,22], ribs [22,23], femur [24,25], and maxillofacial bones [9,26], but the majority of case reports refer to the shoulder and pelvis [16,27–29]. GSD can occur in males and females, with a slight predominance in males [30], and has been reported in patients aged 1 [31] to 75 [32] years. Our case occurred in a 14-year-old girl.

The clinical presentation of GSD most frequently includes pain, functional impairment, and swelling of the affected region, although asymptomatic cases such as ours have been reported [30]. Clearly, the clinical manifestation depends on the affected area.

CT and MRI play crucial roles in the radiographic characterisation of GSD, and enable division of the disease process into four stages [33]. CT is used to delineate the extent of osteolysis, and 3D images are useful for the planning of surgery [31]; MRI, the use of which in cases of GSD has been described only rarely, is useful for visualisation of the vascular structures and fibrosis [20]. Nevertheless, the gold standard criterion remains biopsy positivity for the presence of angiomatous tissue [9].

The typical histological findings show non-specific vascular proliferation intermixed with fibrous connective tissue and chronic inflammatory infiltration of lymphocytes and plasma cells [34].

The differential diagnosis includes hereditary multicentric osteolysis, osteolysis with nephropathy, osteomyelitis, rheumatoid arthritis [35], osteolysis due to intraosseous malignancy, hyperparathyroidism, eosinophilic granuloma, and osteolysis due to diseases of the central nervous system [36].

The proper course of treatment for GSD remains unclear, and no standard therapy is available; current treatment modalities include

Fig. 7. The axial CT view of an isolated phenomenon of identical appearance affected the posterior region of the distal tibia.

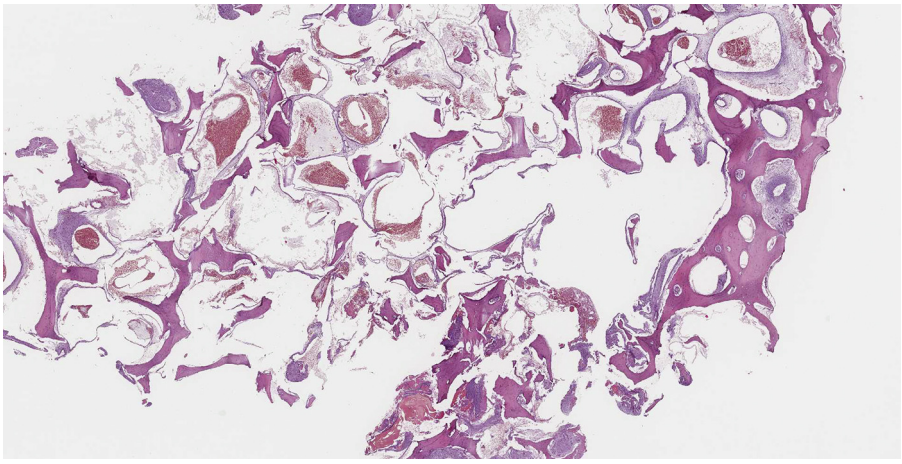

Fig. 8. Endothelial-vascular spaces without atypia interspersed with fibrous connective tissue stroma.

surgery, radiation therapy, and the use of bisphosphonates and interferon alpha-2b [37].

In our case, the patient initially underwent pharmacological therapy with vitamin D, calcium, and bisphosphonates. Due to the extent of the lesion, and because the disease is often self-limiting [37], conservative treatment appeared to be an appropriate choice in our case.

The follow-up period is currently 3 months; we are waiting a little bit more time to perform other radiographs.

This case report was written according to the Surgical CAse REport guidelines [38].

4. Conclusion

Decisions regarding the management of GSD are influenced by many factors, as the disease, although considered to be benign and spontaneously resolving, is characterised by an unpredictable prognosis and possible serious complications [39].

We present this rare case of GSD with several localisations to emphasise that any procedure appearing to be simple can go catastrophically wrong and can result in the discovery of something that is unexpected and extremely rare.

Therefore, in this article we wish to emphasise the extreme rarity of the case, focusing on the large difference between the preoperative panoramic radiograph and whole-body CT and MRI images obtained on the day after the first surgical procedure.

Conflicts of interest

No conflict.

Funding sources

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Ethical approval

No.

Consent

The consent has been given.

Author contributions

All authors have contributed in study concept, data collection, data analysis and interpretation and in writing the paper.

Registration of research studies

No.

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References

[1] Y. Liu, D.R. Zhong, P.R. Zhou, F. Lv, D.D. Ma, W.B. Xia, Y. Jiang, O. Wang, X.P. Xing, M. Li, Gorham-Stout disease: radiological, histological, and clinical features of 12 cases and review of literature, Clin. Rheumatol. 35 (March (3)) (2016) 813–823.
[2] T. Hirayama, A. Sabokbar, I. Itonaga, S. Watt-Smith, N.A. Athanasou, Cellular and humoral mechanisms of osteoclast formation and bone resorption in Gorham-Stout disease, J. Pathol. 195 (December (5)) (2001) 624–630.
[3] P. Boyer, P. Bourgeois, O. Boyer, Y. Catonné, G. Saillant, Massive Gorham-Stout syndrome of the pelvis, Clin. Rheumatol. 24 (September (5)) (2005) 551–555.
[4] J.B.S. Jackson, A boneless arm, Boston Med. Surg. J. 18 (1838) 368–369.
[5] J.B.S. Jackson, Absorption of the humerus after fractures, Boston Med. Surg. J. 10 (1872) 245–247.
[6] O. Romer, Die pathologie der zahne, in: F. von Henke, O. Lubarsch (Eds.), Handbuch Der Speziellen Pathologischen Anatomie Und Histology, Springer, Berlin, 1924, pp. 135–499.
[7] L.W. Gorham, A.P. Stout, Massive osteolysis (acute spontaneous absorption of bone, phantom bone, disappearing bone). Its relation to hemangiomatosis, J. Bone Joint Surg. Am. 37 (1955) 985–1004.
[8] G. Bocchialini, L. Ferrari, D. Burlini, A massive haemorrhage developing during deciduous tooth extraction in a young child: a case report, Int. J. Surg. Case Rep. 31 (2017) 237–240, http://dx.doi.org/10.1016/j.ijscr.2017.01.061.
[9] L. Heffez, H.C. Doku, B.L. Carter, J.E. Feeney, Perspectives on massive osteolysis. Report of a case and review of literature, Oral Surg. Oral Med. Oral Pathol. 55 (1983) 331–343, http://dx.doi.org/10.1016/0030-4220(83)90185-8.
[10] F. Hardegger, L.A. Simpson, G. Segmueller, The syndrome of idiopathic osteolysis: classification, review and case report, J. Bone Joint Surg. 67 (1985) 89–93.
[11] J.S. Torg, A.M. DiGeorge, J.A. Kirkpatrick Jr., M.M. Trujillo, Hereditary multicentric osteolysis with recessive transmission: a new syndrome, J. Pediatr. 75 (August (2)) (1969) 243–252.
[12] R.I. Macpherson, R.D. Walker, M.H. Kowall, Essential osteolysis with nephropathy, J. Can. Assoc. Radiol. 24 (June (2)) (1973) 98–103.
[13] G. Heyden, L.-G. Kindblom, J.M. Nielsen, Disappearing bone disease: a clinical and histological study, J. Bone Joint Surg. Am. 59 (1977) 57–61.
[14] S. Lala, J.B. Mulliken, A.I. Alomari, S.J. Fishman, H.P. Kozakewich, G. Chaudry, Gorham-Stout disease and generalized lymphatic anomaly – clinical, radiologic, and histologic differentiation, Skeletal Radiol. 42 (July (7)) (2013) 917–924, http://dx.doi.org/10.1007/s00256-012-1565-4, Epub 2013 Jan 31.
[15] C.J. Fretz, W.F. Jungi, J. Neuweiler, M. Haertel, The malignant degeneration of Gorham-Stout disease, Rofo 155 (December (6)) (1991) 579–581.

[16] H.A. Kulenkampff, G.M. Richter, W.E. Hasse, C.P. Adler, Massive pelvic osteolysis in the Gorham-Stout syndrome, Int. Orthop. 14 (4) (1990) 361–366.
[17] J.S. Thompson, D.J. Schurman, Massive osteolysis: case report and review of literature, Clin. Orthop. 103 (1974) 206–211.
[18] J.W. Young, M. Galbraith, J. Cunningham, B.S. Roof, I. Vujic, R.P. Gobien, et al., Progressive vertebral collapse in diffuse angiomatosis, Metab. Bone Dis. Relat. Res. 5 (1983) 53–60.
[19] M.I. Glass-Royal, M.A. Stull, Musculoskeletal case of the day: gorham syndrome of the right clavicle and scapula, AJR Am. J. Roentgenol. 154 (June (6)) (1990) 1335–1336.
[20] T.A. Damron, D.S. Brodke, J.P. Heiner, J.S. Swan, S. DeSouky, Case report 803: Gorham's disease (Gorham-Stout syndrome) of scapula, Skeletal Radiol. 22 (August (6)) (1993) 464–467.
[21] D. Feigl, A. Marmor, Gorham's disease of the clavicle with bilateral pleural effusions. Eight years later, Chest 92 (July (1)) (1987) 189.
[22] H.D. Sacristan, L.F. Portal, F.G. Castresana, D.R. Pena, Massive osteolysis of the scapula and ribs. A case report, J. Bone Joint Surg. Am. 59 (April (3)) (1977) 405–406.
[23] J. Romero, R. Kunz, U. Münch, U. Neff, Successful treatment of a chylothorax in lymphangiomatosis of the ribs (Gorham-Stout syndrome), Schweiz. Med. Wochenschr. 119 (May (20)) (1989) 671–677.
[24] J. Stöve, A. Reichelt, Massive osteolysis of the pelvis, femur and sacral bone with a Gorham-Stout syndrome, Arch. Orthop. Trauma Surg. 114 (4) (1995) 207–210.
[25] A.A. Mendez, D. Keret, W. Robertson, G.D. MacEwen, Massive osteolysis of the femur (Gorham's disease): a case report and review of the literature, J. Pediatr. Orthop. 9 (September–October (5)) (1989) 604–608.
[26] T. Ohya, S. Shibata, Y. Takeda, Massive osteolysis of the maxillofacial bones: report of two cases, Oral Surg. Oral Med. Oral Pathol. 70 (1990) 698–703, http://dx.doi.org/10.1016/0030-4220(90)90003-B.
[27] L.F. Remia, J. Richolt, K.M. Buckley, M.J. Donovan, M.C. Gebhardt, Pain and weakness of the shoulder in a 16-year-old boy, Clin. Orthop. Relat. Res. 268 (February (347)) (1998), 268-71, 287-90.
[28] L.C. Hofbauer, R.A. Klassen, S. Khosla, Gorham-Stout disease (phantom bone) of the shoulder girdle, Rheumatology (Oxford) 38 (September (9)) (1999) 904–905.
[29] N. Hejgaard, P.R. Olsen, Massive Gorham osteolysis of the right hemipelvis complicated by chylothorax: report of a case in a 9-year-old boy successfully treated by pleurodesis, J. Pediatr. Orthop. 7 (January–February (1)) (1987) 96–99.
[30] P. Hu, X.G. Yuan, X.Y. Hu, F.R. Shen, J.A. Wang, Gorham-Stout syndrome in mainland China: a case series of 67 patients and review of the literature, J. Zhejiang Univ. Sci. B 14 (August (8)) (2013) 729–735, http://dx.doi.org/10.1631/jzus.B1200308.
[31] P. Vinée, M.O. Tanyü, K.H. Hauenstein, G. Sigmund, B. Stöver, C.P. Adler, CT and MRI of gorham syndrome, J. Comput. Assist. Tomogr. 18 (November-December (6)) (1994) 985–989.
[32] C. Szabo, W. Habre, Gorham syndrome: anaesthetic management, Anaesthesia 55 (February (2)) (2000) 157–159.
[33] R. Kotecha, L. Mascarenhas, H.A. Jackson, R. Venkatramani, Radiological features of Gorham's disease, Clin. Radiol. 67 (August (8)) (2012) 782–788, http://dx.doi.org/10.1016/j.crad.2012.01.009, Epub 2012 Mar 15.
[34] L.P. Zhong, J.W. Zheng, W.L. Zhang, S.Y. Zhang, H.G. Zhu, W.M. Ye, Y.A. Wang, Z.Y. Zhang, Multicentric Gorham's disease in the oral and maxillofacial region: report of a case and review of the literature, J. Oral Maxillofac. Surg. 66 (May (5)) (2008) 1073–1076, http://dx.doi.org/10.1016/j.joms.2007.06.677.
[35] M. Ozbayrak, M.H. Yilmaz, F. Kantarci, H. Ozer, K. Harmanci, M. Babacan, S. Dervisoglu, A case of an idiopathic massive osteolysis with skip lesions, Korean J. Radiol. 14 (November–December (6)) (2013) 946–950, http://dx.doi.org/10.3348/kjr.2013.14.6.946, Epub 2013 Nov 5.
[36] M.H. Motamedi, S.M. Homauni, H. Behnia, Massive osteolysis of the mandible: a case report, J. Oral Maxillofac. Surg. 61 (August (8)) (2003) 957–963.
[37] S.M. Gondivkar, A.R. Gadbail, Gorham-Stout syndrome: a rare clinical entity and review of literature, Oral Surg. Oral Med. Oral Pathol. Oral Radiol. Endod. 109 (February (2)) (2010) e41–e48, http://dx.doi.org/10.1016/j.tripleo.2009.08.043, Epub 2009 Dec 6.
[38] R.A. Agha, A.J. Fowler, A. Saetta, I. Barai, S. Rajmohan, D.P. Orgill, the SCARE Group, The SCARE statement: consensus-based surgical case report guidelines, Int. J. Surg. 34 (2016) 180–186.
[39] V.S. Nikolaou, D. Chytas, D. Korres, N. Efstathopoulos, Vanishing bone disease (Gorham-Stout syndrome): a review of a rare entity, World J Orthop. 5 (November (5)) (2014) 694–698, http://dx.doi.org/10.5312/wjo.v5.i5.694, eCollection 2014.