Case Report
Congenital Craniofacial Deformities

Orthognathic surgery in pycnodysostosis: a case report

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Abstract. Pycnodysostosis is an extremely rare genetic osteosclerosis caused by cathepsin K deficiency. It is a human autosomal recessive genetic disorder characterized mainly by osteosclerosis of the skeleton due to decreased bone turnover. It is characterized by short stature, brachycephaly, short and stubby fingers, open cranial sutures and fontanelle, and diffuse osteosclerosis. Multiple fractures of the long bones and osteomyelitis of the jaw are frequent complications. The authors describe an 18-year-old girl with a clinical and radiological diagnosis of pycnodysostosis and the ortho-surgical treatment undertaken. Bimaxillary orthognathic surgery was carried out using rigid fixation and bone grafts. The authors recommend bimaxillary orthognathic surgery as a choice for treating the dentofacial deformities of pycnodysostosis, emphasizing the good and stable results obtained in terms of facial aesthetics and occlusion.

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Pycnodysostosis is a rare osteopetrotic clinical entity. It belongs to the group of craniotubular bone dysplasias, first described in 1962 by Maroteaux and Lamy as a form of dwarfism with craniofacial malformation similar to cleidocranial dysplasia[1,5–7,10]. The disease has also been called Toulouse-Lautrec syndrome, after the French artist Henri de Toulouse-Lautrec who suffered from the disease[1]. Pycnodysostosis has an autosomal recessive inheritance[1–10] and is characterized by systemic high bone density due to decreased bone turnover[1–7,9]. In the 1990s, the defective gene responsible for pycnodysostosis was located in chromosome Iq21, offering accurate diagnosis, carrier testing and a more thorough understanding of this disorder[1,3,5–7,10]. Pycnodysostosis represents a lysosomal storage disease of the bone caused by a mutation in the gene that codes the enzyme cathepsin K. This protease plays a major role in osteoclast-driven bone resorption[6,7,9,10] and is responsible for degrading collagen type 1, which constitutes 95% of the organic bone matrix[1,9]. The bones in individuals afflicted with pycnodysostosis are abnormally dense and brittle as a result of this insufficient reabsorption process[5,9]. Pycnodysostosis is usually diagnosed at an early age due to the typical phenotype with proportionate dwarfism and peculiar facies[1,3,5,6]. The diagnosis is sometimes made late, as a result of high susceptibility to long bone fractures and infections, because of the severe bone fragility resulting from increased bone density and impaired bone vascularity[1–10].

Extragnathic skeletal involvement includes short stature, clavicular and craniofacial dysplasia, total or nearly total phalangeal dysplasia and generally radiographic osteosclerosis. The acromial ends of the clavicles may be aplastic[1,2,5]. This syndrome usually presents with typical craniomaxillofacial deformities, such as a dolicocephalic or brachiocephalic skull with prominent forehead, hypoplastic maxilla and mandible with micrognathia, and hypoplastic midface with exophthalmus, hypopneumatization of the maxillary sinuses, beaked nose, open and anterior cross-bite, obtuse mandibular angle, grooved palate, longer soft palate and narrowing of the airway[1–3,5–7,10]. Dental crowding, dental abnormalities and impaction are observed, as well as alterations in eruption[1–3,6].

The diagnosis of pycnodysostosis is primarily based on clinical features and radiographs, but the confirmatory test is a cathepsin K gene mutation analysis. Other uncommon clinical disorders of reduced bone resorption include osteopetrosis, osteogenesis imperfecta, cleidocranial dysplasia and idiopathic acroosteolysis[1,5]. In osteopetrosis, the bone marrow may be absent so hematopoietic alterations are common. Signs of compression of the cranial nerves include facial paralysis, deafness or pain. Cleidocranial dysplasia may seem like pycnodysostosis because it presents with agenesis or clavicular aplasia, and alterations of the skeletal bone membranes, however, bone density is not increased. In idiopathic acroosteolysis, the appearance of the patients is typical, with hypotelorism, exophthalmos, and an upturned nose but increased bone density is not present[1,9].

The choice of surgical technique was difficult because the high risk of infection, secondary fractures and nonunion render

the classical choices of distraction and iliac bone graft hazardous[10]. There have been few reports of rigid fixation of such dysplasic bone in the craniomaxillofacial area[4,7,8,10]. The purpose of this paper is to report bimaxillary orthognathic surgery and bone grafting for treating the dentofacial deformities of pycnodysostosis, emphasizing the good and stable results obtained in terms of facial aesthetics and occlusion.

Case report

The authors present the case of an 18-year-old girl who had been clinically and radiographically diagnosed with pycnodysostosis and referred to them for treatment. The cathepsin K gene mutation test was performed and confirmed the presence of the disease.

The patient's main complaints were her facial appearance, occlusal alterations and snoring. Physical examination revealed a severely distorted face as a result of anteroposterior and vertical maxillary and mandibular hypoplasia. A severe short face was accompanied by a relative rhinomegaly. A class II occlusion was present (Fig. 1). Other findings included short stature and clavicular and phalangeal dysplasia. A cone beam CT scan revealed severely distorted facial architecture with abnormally small facial bones.

Treatment began with fixed orthodontics to achieve enough alignment to allow for further orthognathic surgery.

Test surgery was carried out to evaluate the patient's bone response to facial osteotomies, xenografts and rigid fixation with osteosynthesis material, because of the high risk of infection and nonunion as a result of the poor bone quality. For that purpose, a vertical and sagittal augmentation genioplasty was carried out under general anaesthesia with 8 mm advancement and 7 mm downgrafting with interposition of a bovine hydroxyapatite block (Bioss). Postoperative recovery and healing was uneventful.

After 14 months of presurgical orthodontics the patient was considered ready for bimaxillary surgery. The orthodontist used skeletal anchorage to aid levelling. The preoperative work up included a cone beam CT and conventional model surgery with the generation of two surgical splints.

Surgery was performed under hypotensive general anaesthesia. The chin plate was removed at the time of orthognathic surgery and excellent bone healing was observed. A mandibular advancement of 13 mm with bilateral sagittal split osteotomies was performed. A Le Fort I osteotomy followed with maxillar advancement of 10 mm and downgrafting of 8 mm. Rigid fixation was achieved with four miniplates in the maxilla and four more in the mandible. Two plates on each mandibular osteotomy were used to hold the advancement and to provide enough stability. Interpositional blocks of Bioss were placed in the gaps both at the Le Fort I and the sagittal split. An open reduction rhinoplasty with dorsal, alar and septal reduction was performed. No postoperative intermaxillary fixation was applied apart from two light box elastics to guide mandibular movements.

Postoperative recovery was uneventful and the patient was discharged 48 h after surgery. A post-surgical cone beam CT scan revealed adequate repositioning of the bony segments. The patient followed a liquid diet for 10 days and then a soft diet for 2 months.The postoperative orthodontics lasted for 12 months and yielded a functional class I occlusion with stable periodontal parameters. A cone beam scan, 24 months post-surgery, revealed stability of skeletal movements and adequate consolidation at the osteotomies. Patient satisfaction with facial and occlusal results was very high. As a result of her high satisfaction level, orthodontics is still in progress due to low cooperation from the patient after surgery. She was able to resume her social life, which had been severely compromised (Fig. 2).

Discussion

Pycnodysostosis is a rare autosomal recessive disease. The incidence is estimated to be 1.7 per 1 million births[1,2,5,10]. Multiple fractures of long bones and osteomyelitis, stridorous breathing and snoring due to a narrow chest and airway are frequent complications[5,6,10]. Muto et al.[6] reported pycnodysostosis presenting as severe snoring, caused by pharyngeal narrowing in two siblings. Cephalograms[3,6] showed pharyngeal narrowing at the level of the soft palate and the base of the tongue caused by the long soft palate and mandibular hypoplasia. They suggested that respiratory insufficiency, such as snoring or

Fig. 1. Clinical examination. (A) Frontal view and (B) lateral view, revealed a severely distorted face because of anteroposterior and vertical maxillary and mandibular hypoplasia. A severe short face was accompanied by a relative rhinomegaly. A class II occlusion was present.

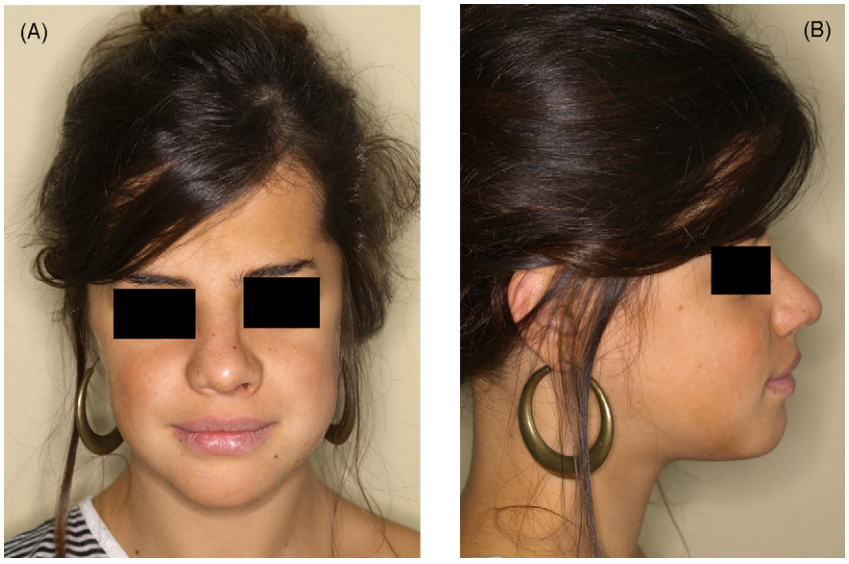

Fig. 2. Views 24 months after surgery. (A) Clinical frontal and (B) lateral views. The aesthetic facial results were very high and allowed the patient to resume her social life, which had been severely compromised.

obstructed sleep apnoea, are common and under-appreciated symptoms of pycnodysostosis[3,6]. The maxillary hypoplasia often results in a class III occlusal relationship with characteristic retrusion, and vertical reduction of the midface[3,7]. The body of the mandible also presented a generalized reduction in size, which could be appreciated more by Go-Me measurements, leading to the belief that the maxilla would be more involved in the development of the significantly unfavourable skeletal sagittal pattern of these patients[3].

Bone fragility and a history of frequent fractures may suggest a diagnosis of abnormalities in the material composition and structure of bone[5,9]. Tooth extraction, in patients who suffer from pycnodysostosis, requires special care, such as carrying out the surgery as atraumatically as possible and with proper asepsis, due to the risk of fracture, especially in the mandible. The impaired bone vascularity, increases the probability of developing post-extraction osteomyelitis[1]. Treatment of osteomyelitis of the jaws in pycnodysostosis is difficult and may lead to large resections[2,7].

No information is available in the literature regarding the efficacy and safety of orthodontics in children or young adults with pycnodysostosis[3]. Orthodontic and orthopaedic movements are dependent on osteoclastic activity and bone resorption and remodeling capacities. Few cases of elective skeletal facial surgery in pycnodysostosis have been reported[7,8,10] and there are few reports of rigid fixation in dysplasic facial bones[4,7,8,10]. The most important orthopaedic problem for surgical correction in this condition is the recurrent pathological fracture of bones and the high infection rates, due to the limited quality and vascularity of the sclerotic bone[1,2,5–8,10]. These facts represent a challenge for the maxillofacial surgeon. A case report of a successful conventional Le Fort I osteotomy was presented by Polley and Figueroa[8]. The size of surgical movement was smaller, they performed only monomaxillar surgery and there was no need for bone grafting[8]. Nørholt et al.[7] performed maxillary distraction for a 15-year-old girl with pycnodysostosis who presented with severe dentofacial deformity. They reported stable consolidation was only obtained 13 months after Le Fort I osteotomy followed by 6 weeks of external distraction.

Teissier et al.[10], reported the case of a 3-year-old boy with pycnodysostosis who presented with severe snoring as a result of pharyngeal narrowing due to a hypoplastic mandible. They performed a bilateral rib graft to correct the micrognathia and to allow enlargement of the pharynx by acting on an anterior projection of the chin and a decrease of the glossoptosis. A significant reduction in snoring and an excellent mouth aperture were obtained. They stated that this risk-limited (compared with free fibular or iliac flap) surgical technique is ideal for patients presenting a high risk of infection, nonunion and secondary fracture (particularly if a fibular free flap was obtained) after such a surgical procedure[10]. In the reported case, plates and screws achieved enough stability and allowed uneventful bone healing. The osseous healing was verified by a cone beam CT scan. Neither the authors nor the patient encountered any problems during treatment. The long-term stability of the achieved occlusion and facial appearance confirm that adequate healing in such sclerotic bone is to be expected using standard fixation. From the literature[3,6] and their results, the authors suggest that respiratory insufficiency, such as snoring or obstructed sleep apnoea, are common and under-appreciated symptoms of pycnodysostosis, and bimaxillary advancement may be beneficial.

In conclusion, patients affected with pycnodysostosis with severe malocclusions and upper airway and aesthetic compromise may be treated with bimaxillary orthognathic surgery successfully. This patient remained stable for at least 2 years after surgery. The surgical technique is hazardous because of the high risk of infection and nonunion healing, but bimaxillary orthognathic surgery may be a better choice for achieving successful and stable aesthetic and functional results.

Competing interests

None declared.

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Ethical approval

Not required.

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Case Report
Dental Implants

Zygoma implant-supported prosthetic rehabilitation of a patient with a maxillary defect

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T. Shirota, O. Shimodaira, Y. Matsui, M. Hatori, S. Shintani: Zygoma implant-supported prosthetic rehabilitation of a patient with a maxillary defect. Int. J. Oral Maxillofac. Surg. 2011; 40: 106–123.

Abstract. This clinical report describes the successful management of a patient who underwent extensive resection of a maxillary cancer, by introduction of a maxillary obturator prosthesis using zygoma implants. The patient was a 57-year-old man with cancer of the upper anterior gingiva. The maxillary bone in the affected region had been extensively excised by radical surgery. Owing to loss of teeth retaining the denture, the existing prosthesis was unstable, and the patient experienced severe speech and mastication disorders. Four zygoma implants (two on each side), and two conventional dental implants (one each at both maxillary tuberosities) were used as denture retainers. The obturator prosthesis was stabilized by the implants, and the patient's oral function improved. High-level compatibility between the implant and surrounding tissue was obtained by mucosal regeneration around the implant. The results suggest that the combination of zygoma and conventional dental implants improves postoperative oral function by facilitating retention of the obturator prostheses.

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Severe maxillary defects resulting from surgical resection of oral neoplasms are associated with major difficulties in restoring oral function, such as mastication, speaking and swallowing, and impaired facial esthetics. In such cases, conventional dental implants have been used to improve the stability and retention of maxillofacial prosthetic obturators and to restore oral function[1,3]. Implant placement and the subsequent prosthetic treatment often become difficult following resection of maxillary cancer because of a lack of bone tissue in the areas where conventional dental implants can be placed, and extensive loss of soft tissues including the gingiva, mucosa and muscle.

The zygoma implant was developed for edentulous patients with insufficient bone mass for dental implants[5]. This implant is fixed in the zygoma so it is considered