Gorlin's syndrome - Report of a case and management of cystic lesions

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Abstract Gorlin's syndrome associated with multiple cystic lesions is a rare and uncommon condition. It is characterized by cutaneous basal cell carcinomas, multiple keratocysts in the jaw bones and skeletal anomalies. A case is presented with multiple odontogenic keratocyst and dentigerous cysts occurring in all quadrants of oral cavity with features suggestive of Gorlin's Syndrome. The clinical presentation and the management of these multiple cystic lesions and a two year follow up study is presented.

Keywords Keratocyst · Dentigerous cyst · Carcinoma · Nevoid

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Introduction

Gorlin – Goltz Syndrome is an autosomal dominant disorder and was first described in 1960 [11]. It is characterized by cutaneous basal cell carcinomas, multiple keratocysts in the jaw bones and skeletal anomalies [16]. It affects male and female equally and seen during the first, second and third decades of life [6,8]. Various studies have been proposed to study the genetic mechanism. It was suggested that causative mutations which occur are equivalent to that of Patched (PTCH) gene, which functions as a tumor suppressor gene [5,8,9,15,11]. The principle clinical features of Gorlin's syndrome comprise multiple odontogenic keratocyst, Basal cell nevi and skeletal anomalies [16].

The incidence of keratocyst is 75% in patients with Gorlin's syndrome. The cysts can be the first features of the syndrome and are found on incidental radiographic examination or the patient may present with symptoms, in case of secondary infection [4]. Radiographically the lesions were bilateral but can occur unilaterally also. They can appear either as unilocular or multilocular radiolucencies of the posterior body, angle or ramus of the mandible [17]. Evidence of displacement of developing teeth, root resorption and presence of unerupted teeth within the lesion are common. These are locally destructive, spreads within the bone and causes lateral expansion [16].

The histologic features of odontogenic keratocyst includes a thin parakeratinised or orthokeratinised stratified squamous epithelium (usually five to eight cell thickness), a well defined basal cell layer generally without retepeg formation, an uninfamed fibrous capsule and a lumen containing desquamated keratin. The connective tissue wall often contains small islands of epithelium and small satellite or daughter cysts [19].

The recurrence rate of odontogenic keratocysts is high i.e. 60% due to its thin lining and/or presence of satellite cysts within the capsule [2]. Various combination of treatments have been proposed i.e. surgical excision followed by cryotherapy or application of Carnoy's solution [21,19]. It has been suggested that the recurrence rate in patient's with Gorlin's syndrome may be higher than of cyst occurring in healthy individuals. This may be due to the inherited tendency within the basal layer of epithelium to develop new cysts in the patient with Gorlin's syndrome [20]. Hence early diagnosis and treatment with close clinical and radiological followup is as important as surgical treatment [10,14].

70% of patients with Gorlin's syndrome have some degree of craniofacial anomaly [11]. This can comprise of high and broad forehead, frontal and parietal bossing, a broad nasal root associated with ocular hypertelorism [10,14]. A hypoplastic maxilla and hyperplastic mandible may be found giving rise to crossbite teeth

relationship. Other anomalies include high arched palate, cleft lip and palate, malocclusion and impacted teeth [18].

Thoracic cage anomalies amount for 43% which includes bifid and fused ribs. Vertebral anomalies which can arise upto 31% includes spins bifida occulta, kyphoscoliosis, fusion defects and hemi vertebrae [7,14].

Case report

A 13-year-old Saudi boy reported to Oral and Maxillofacial unit of Regional Dental Centre, King Khalid Hospital, Najran, with a referral and a history of large swelling over the right side of the face since five days. Previously patient had undergone extraction of over retained maxillary right deciduous canine. History revealed similar swelling over the left side of the face approximately six months ago. On examination it appeared that the boy had slight mental retardation, mild hypertelorism, frontal bossing and kyphoscoliosis. Medical history revealed that the patient had undergone orchidopexy for both the undescended testis. There was evidence of diffuse swelling over the right side of maxilla, which extended superiorly till the infraorbital rim, obliterating the nasolabial fold. There was expansion of labial cortical plate, tenderness and 'egg shell crackling' was noted. Pus discharge was seen from extraction socket of

Fig. 1 OPG showing multiple impacted teeth with radiolucent lesions

Fig. 2 PNS view showing lesions involving maxillary antrums

Fig. 3 Postoperative 6 months evidence of new cystic lesion

Fig. 4 PA view of chest revealing fused ribs on right side

Fig. 5 Postoperative OPG revealing satisfactory bone healing

maxillary right deciduous canine, pathologic migration of adjacent lateral incisor was noted. Similar minimal expansion of labial and anterolateral wall of left maxillary sinus was present. Anterior teeth were in cross bite relationship, malocclusion, ectopic position of teeth and high arched palate.

Radiographic examination of PNS view (Fig. 2) revealed presence of pathologies in both the maxillary sinuses and impacted teeth, maxillary right second molar, maxillary right and left canines, and maxillary left first premolar. OPG (Fig. 1) revealed radiolucent lesions in the right and left retromolar regions. Water's view

showed clear demarcation between radiolucent lesions of both maxillary canines and maxillary antrum. Routine investigations were done including post-anterior view of chest (Fig. 4) which revealed presence of bifid ribs. Initial diagnosis of infected cysts was made and plan was exploration of all radiolucent lesions, and surgical enucleation under general anesthesia as it will be difficult to carryout biopsy of each individual lesions.

Under general anesthesia right and left buccal mucoperiosteal flaps were raised in the retromolar regions. Enucleation of cystic lesions was done along with extraction of mandibular left third molar.

The cystic bony walls were freshened with rotary bur, packed with surgical haemostatic material and flaps closed with vicryl sutures. In the maxilla too buccal flaps were raised on right and left side involving the gingival margin. Two separate cystic lesions involving the maxillary canines and maxillary antrums were noted and enucleation was done including extraction of impacted maxillary right second molar, both the canines, and left first premolar teeth. Maxillary antrums were packed with antibiotic-impregnated gauze rolls separately and flaps closed with 3–0 vicryl sutures. A total of six cystic lesions were enucleated and specimen sent for histopathological examination. On the sixth postoperative day, under local anesthesia antibiotic packs were removed by releasing two sutures and flaps were resutured.

Healing was uneventful and patient was followed up periodically. Histopathology of the six specimens were reported as, five lesions were dentigerous cysts arising from both the retromolar regions, canine regions bilaterally and right maxillary sinus, and. One specimen was reported as Odontogenic keratocyst from left maxillary sinus region.

Six months postoperative radiograph showed good healing of cystic cavities with the evidence of new bone formation. Also

it was noted that there was small radiolucent lesion between mandibular left lateral incisor and partially impacted canine. (Fig. 3) It was decided to keep the patient under observation for one month. Orthopantomograph was done later which revealed mild increase in the dimension of new radiolucent lesion in the mandible and pathologic migration of canine was noted. It was informed to the patient and the relatives. Surgical enucleation of new pathology was done under local anesthesia, by raising the labial flap. The lesion was treated as odontogenic keratocyst, and the specimen was sent for histopathologic examination, which was reported as odontogenic keratocyst. The cystic cavity healed well during subsequent months. The last follow up was done 24 months postoperatively, and OPG (Fig. 5) revealed excellent healing process, with no evidence of any recurrence and any new lesion.

Discussion

Gorlin – Goltz Syndrome also called as Nevoid basal cell Carcinoma Syndrome is a rare entity principally a triad of multiple basal cell nevi, jaw keratocysts and skeletal anomalies [12]. The diagnostic criteria to designate this condition was given by Evans DG et al. in 1991. Other anomalies associated with this syndrome were published by Manfredi M in 2004. The age predilection for this condition is in the 1–3rd decade of life [14,6]. This supports the case reported here, which is in the first decade. The patient was around 13-years-old with over retained maxillary canine teeth. Patient did not have basal cell carcinoma, but there were other major criteria like bifid ribs and histologically proven odontogenic keratocyst of jaw. It was interesting to note that out of the seven cystic lesions, only two were odontogenic keratocysts and rest were all dentigerous cysts. These findings are supported by the fact that a similar case was presented by Andras Ezsias in 2001 [1]. Patient had other minor criteria's i.e skeletal anomalies like frontal bossing, bifid ribs, scoliosis. Neurological anomaly like mild mental retardation. Oropharyngeal anomalies like malocclusion, ectopic position of teeth, impacted teeth, high arched palate and ophthalmic anomaly like mild hypertelorism was noted in this case.

Odontogenic cystic lesions are asymptomatic and are found usually on routine radiographs. Other mode of presentation is when they are secondarily

infected causing swelling and facial asymmetry [4]. This supports the case as the patient reported with history of large swelling and pus discharge following extraction of maxillary right deciduous canine and upon routine radiographic examination other cystic lesions were noted.

The management of dentigerous cyst is not controversial as compared to keratocyst, due to the fact that keratocyst have high recurrence rate because of thin lining which is friable and presence of satellite cysts. Various conservative approaches have been proposed for young patients with large cysts to preserve soft and hard tissues including the teeth. These include marsupialisation of the cyst or decompression followed by secondary enucleation, but results are in long term are ineffective [3]. Other procedures include surgical excision of the keratocyst followed by cryotherapy or Carnoy's solution [19,21,22]. In this case surgical enucleation of all the cystic lesions was done followed by removal of 1–2 mm layer of bone around the periphery of cystic cavities with sharp curette or a bone bur with sterile irrigation to remove the epithelial remnants, which could proliferate into recurrence of the cyst [13].

The recurrence rate or incidence of new lesion being developed is higher in patients with Gorlin's syndrome compared to other patients. This is due to inherited tendency within the basal layer of their epithelium to develop new cyst [20]. This supports the case reported here, as this patient developed a new cystic lesion in relation to partially erupted mandibular left canine 6 months after the initial surgical procedure.

Conclusion

Gorlin's syndrome is a rare autosomal dominant disorder affecting the skeletal structure of the body. The other important features are cutaneous basal cell carcinomas, multiple keratocysts of jaw bones. Clinically it is important to recognize the condition and thorough radiographic evaluation is mandatory to assess involvement of other bones. The cystic lesions have to be diagnosed based on their radiographic appearances, aspiration contents and consistency of cystic lining and contents of lesions upon surgical exposure. It is relevant to subject all the specimens for histopathological examination as in this case which proved the presence multiple dentigerous cysts in

this syndrome, which is a rare. Long term followup is required to ensure that no recurrence of keratocyst occurs.

In conclusion this case is reported here is an addition to the cases of Gorlin's syndrome, which are very rare. It is interesting to note the occurrence of multiple dentigerous cysts and keratocyst in combination in all the quadrants of oral cavity.

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