ORIGINAL RESEARCH PAPER

Volume-8 | Issue-6 | June-2019 | PRINT ISSN No. 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

UNUSUAL CASE PATHOLOGIES–A SYSTEMATIC APPROACH TO DIFFERENTIAL DIAGNOSIS-A REPORT OF 4 CASES

Oral Pathology

Prasanth Panicker Department of Oral & Maxillofacial Surgery, Kannur Medical and Dental College, Anjarakandy, Kannur, Kerala and Kinder Hospital, Kochi, Kerala

Archana Shenoy* Department of Oral & Maxillofacial Surgery *Corresponding Author

Ashford Lidiya George Department of Oral & Maxillofacial Surgery Sree Anjaneya Institute of Dental Sciences, Calicut, Kerala

Dr.Arjun Gopinath Department of Oral & Maxillofacial Surgery Kannur Dental College, Anjarakandy, Kannur, Kerala

ABSTRACT

Correct diagnosis of a lesion or a condition is of utmost importance for proper treatment planning and its appropriate management. A multitude of pathologies may affect the head and neck region and may have multiple ways of presentation. Some of these may have significant pathognomonic signs and symptoms and may ease the diagnosis whereas some lesions might have overlapping signs and symptoms which may lead the clinician or the surgeon in the wrong direction. Following a systematic approach including the clinical diagnosis, radiographic aids, histopathological analysis and the use of advanced diagnostic modalities as and when required, is thus, the key to accurate diagnosis and its subsequent appropriate management.

KEYWORDS

differential diagnosis, systematic approach

INTRODUCTION:

Correct diagnosis of a lesion or a condition is of utmost importance for proper treatment planning and its appropriate management. Numerous conditions affect the head and neck region and may have multiple ways of presentation. It should be kept in mind that diagnosis solely based on clinical presentations or imaging may not always suffice even though they may aid in differential diagnosis and narrowing down the possibilities most of the times. A purely clinically oriented diagnosis might lead us in a wrong direction. Thus, a systematic approach including the clinical diagnosis, radiographic aids, histopathological analysis and the use of other advanced diagnostic modalities as and when required, is necessary for accurately diagnosing and appropriately managing them.

Presented here are 4 cases with unusual presentations reported to our institution, and the systematic approach we followed for their accurate identification, our aim being to stress on the importance of following a proper step by step protocol for accurate differential diagnosis.

Case 1:

A 77-year-old female patient reported to our Department of Oral and Maxillofacial Surgery, with a chief complaint of swelling and pain in the right lower jaw region of around 6 months duration. She gave a history of extraction at the same site 8 months back. A significant facial asymmetry was evident owing to the swelling. Intra oral examination revealed a hard-palpable swelling of the buccal shelf at the right mandibular posterior region. A provisional clinical diagnosis of osteomyelitis was arrived upon based on the history, keeping in mind the possibilities of malignancy based on age and rate of progression.

The blood reports were within normal limits except for a significantly increased Alkaline phosphatase. A dental orthopantomogram showed osteolytic destructive lesion at the mandibular posterior region (Fig 1A). A further evaluation with CT scan showed lytic destructive lesion with bone formation and soft tissue mass suggesting a chronic inflammatory or malignant lesion (Fig 1B).

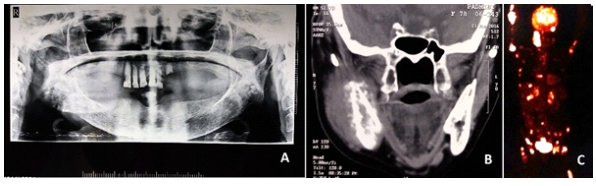


Fig 1: A-dental orthopantomogram showing osteolytic destructive lesion at the right mandibular posterior region; B -CT Coronal view-lytic destructive lesion with specks of bone formation and soft tissue mass, C – PET CT showing extensive and multiple metastatic lesions.

An incisional biopsy from the intra-bony lesion on histopathological evaluation showed irregular bony trabecular pattern with intervening fibroblastic stroma infiltrated by nests of columnar epithelial cells having hyperchromatic nuclei and area of necrosis suggestive of metastatic adenocarcinoma.

A further evaluation and diagnostic workup with triple endoscopy were done. The primary lesion was identified to be the adenocarcinoma of recto-sigmoidal region of the colon on procto-sigmoidoscopy which showed the tumour mass with luminal narrowing. Other endoscopies were found to be normal.

Suspecting additional possibilities of metastasis, a PET CT evaluation was done which showed extensive and multiple metastatic lesions in the liver, right renal pelvis, adrenal glands, bony metastatic deposits in the vertebral bodies and pelvis indicating a terminal disease stage (Fig 1C). In this case, the symptoms due to the secondary metastatic deposits in the right mandibular ramus appeared with no characteristic gastrointestinal symptoms of constipation, diarrhoea, vomiting or abdominal distension from the primary lesion. The other metastatic lesions were also asymptomatic.

The patient was referred to a higher centre for further palliative management considering the extensive nature of the disease and age of the patient.

Case 2:

A 65-year-old male patient reported with a chief complaint of swelling in the left upper cheek region (Fig 2A) of 3 months duration. He also gave a history of an extra-oral swelling which was drained by a general surgeon who he had consulted a few weeks back. Examination revealed extra oral draining sinus and was suggestive of an infected lesion or an abscess. Intraoral examination revealed obliteration of the left buccal sulcus. Routine laboratory investigations were within normal limits. Appropriate antibiotics were prescribed with no regression of the swelling or sinus. A PNS view showed no significant findings. Considering the age of the patient, a contrast enhanced CT of maxillofacial region was done which gave an impression of an expansile lytic lesion with large associated soft tissue component in the left zygomatic bone. Multiple areas of cortical breach were noted. The soft tissue lesion was closely related to the left globe, lateral rectus and lacrimal gland with loss of fat plane between the structures suggestive of possibility of neoplasia. (Fig 2B)

Fig 2: A -swelling in the left zygomatic region, B - CT Coronal view- expansile lytic lesion with large associated soft tissue component

An intraoral incisional Biopsy was done. The zygomatic bone resorption was well appreciable on table during the biopsy procedure. Histopathological evaluation confirmed the diagnosis to be carcinoma in-situ with moderate to severe dysplasia.

Case 3:

3rd case is of a 70-year-old man who reported with pain and swelling on the right side of the lower jaw of around 8 months duration along with a history of halitosis and distaste. He gave a H/o Mucoepidermoid carcinoma of right parotid in 1975 for which he underwent radiation therapy as the treatment modality. He is a known case of diabetes, on medical management and also gave a history of CVA 4 years back.

On extraoral examination, there was no gross facial asymmetry, TMJ seemed normal and no lymph nodes were palpable. On intraoral examination, a well-defined solitary dome shaped, hard non-tender lesion of size 4x1x1cm was evident with significant buccolingual expansion of the mandibular cortices at the posterior mandibular region corresponding to 47, 48 tooth region. Further evaluation with an orthopantamograph revealed well defined multiloculated radiolucency extending from the posterior mandibular body region involving the ascending ramus, sparing the condylar head region (Fig 3A). A CT evaluation of the mandible revealed a lytic lesion involving the right body and ramus of mandible with multiple areas of cortical breach and sclerotic changes with associated soft tissue component involving the retromolar trigone area and the medial pterygoid muscle, atrophic changes in the right Parotid and submandibular salivary gland region with dense calcification of facial artery indicating a likely post-radiotherapy change (Fig 3B).

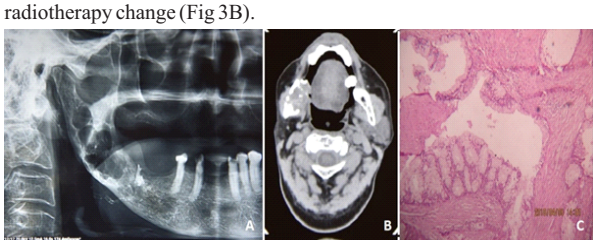
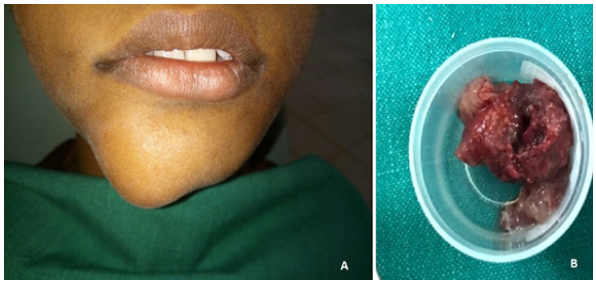


Fig 3: A - orthopantamograph showing well defined multiloculated radiolucency in the posterior mandibular region, B – CT image showing lytic lesion involving the right body and ramus of mandible with multiple areas of cortical breach, C – Histopathological slide showing characteristics of a low grade mucoepidermoid carcinoma - clear cell variant

The patient was managed with Hemimandibulectomy with preservation of the proximal stump. Reconstructive procedures involved use of Reconstruction plates, Sternomastoid transposition and Pectoralis major myocutaneous flap (PMMC).

Case 4:

A 11-year-old male patient reported with a chief complaint of swelling in the chin region (Fig 4A) of 4 months duration which gradually increased in size, non-tender, firm to hard in consistency, mobile and not attached to the underlying bone. Intra oral examination did not yield any positive findings. There were no significant radiographic findings. An incisional biopsy of the lesion and the histo-pathological evaluation was suggestive of fibrosarcoma. The patient was operated upon and lesion excised with sufficient margins (Fig 4B). A thin sticky frothy material was found oozing from the lesion during excision.



4: A - swelling in the chin region, B – excised specimen

Post excision histopathological evaluation was also suggestive of fibrosarcoma. Even though the peak incidence of fibrosarcoma in the head and neck region is reported in the 3rd and 5th decade of life, there has been a wide age range and many cases have been report with patients aged less than 20 years of age.[1,2] Thus, for the purpose of confirmation, a further evaluation with Immuno-histochemistry was done which was weakly positive for desmin and vimentin and was CytoKeratin and Myoglobin negative ruling out fibrosarcoma. A final diagnosis of proliferative fasciitis was reported.

DISCUSSION

A multitude of pathologies may affect the head and neck region. Some of these may have significant pathognomonic signs and symptoms and may ease the diagnosis whereas some lesions might have overlapping signs and symptoms which may lead the clinician or the surgeon in the wrong direction. Few cases might not give any clue making the diagnosis challenging. Following a systematic approach for diagnosis for every case is thus, the key to accurate diagnosis and appropriate management.

A thorough evaluation of the history, clinical presentations and radiological features are thus required for differential diagnosis of these lesions[3]. The age of the patient, the symptoms associated and the rate of growth of the lesions give clues as to whether the lesion is developmental or inflammatory, benign or malignant, aggressive or a less destructive lesion. Radiographic examination provides additional information necessary for diagnosis. Incisional biopsy followed by histopathological examination is required for final and definitive diagnosis of the lesion and in case of metastatic lesions of the jaws, histopathological features vary according to the tissue of origin which can direct the clinician towards diagnosing the primary.[4]

In addition to these routinely used diagnostic modalities, an array of both non-invasive and invasive diagnostic aids is available which need to be utilised appropriately and advertently for accurate diagnosis and proper formulation of treatment plan.

Posterior mandibular region is known to be the site of occurrence of most of the jaw lesions ranging from the more common odontogenic lesions to rarest lesions like metastatic disseminations with more or less, similar or overlapping signs and symptoms. The treatment protocols and management modalities vary significantly based on the type of lesion and its aggressiveness.

With respect to the cases presented in this case series: In the first case report, the signs and symptoms were pointing towards osteomyelitis. The radiographic evaluation showed osteolytic lesion supporting our provisional diagnosis. On evaluating the patient, she didn't report any comorbidities except for Hypertension which was under medicinal control, the routine blood investigations were all within normal limits except for a significant rise in levels of alkaline phosphatase and few specks of radio-opacities on an osteolytic lesion in the mandibular posterior region on an Orthopantomograph. Considering the advanced age of the patient and the radiographic findings, a CT scan was done which was suggesting a chronic inflammatory or malignant lesion which prompted us for further evaluation with incisional biopsy and histopathological examination. The lesion was reported to be Metastatic deposits from an adenocarcinoma. On further work up, the primary was found to be an adenocarcinoma of the recto-sigmoidal junction. It is estimated that only 1 to 3 % of Gastrointestinal cancers have shown metastasis to the bone.[5] They usually involve bones which are rich in haematopoietically active bone marrow like the vertebral column, pelvis etc. Thus, metastasis to the jaw bones is even rarer due to the lack of rich sinusoidal vascular spaces which aid in tumour

spread.[6] Also, in this case, the primary was asymptomatic even though the patient was in the advanced stages. The advanced stage warranted a palliative management.

The second case report is of the patient who reported with an extraoral swelling in the infraorbital-zygomatic region with a draining sinus. The patient was referred to our centre after attempt of incision and drainage by a general surgeon following recurrence, to rule out any dental focus of infection but no significant dental finding was observed. There was no history of comorbidities and routine blood investigations were within normal limits. The signs and symptoms were suggestive of a non- healing infected draining sinus which did not respond to appropriate antibiotics prescribed empirically. On Further evaluation with contrast enhanced CT, the aggressive nature of the lesion was identified and incisional biopsy and histo-pathological evaluation was done. The report showed moderate to severe dysplasia and was suggestive of carcinoma in situ. In this case too, the patient presented with only a swelling and a non- healing infected draining sinus as the chief complaint, stressing on the importance of a proper systematic evaluation and differential diagnosis.

In the third case, even though there was a previous history of mucoepidermoid carcinoma involving the parotid, the current presentation of signs and symptoms were intraosseous and a systematic approach was used for its appropriate diagnosis. Biopsy of the resected specimen was confirmatory of a low grade mucoepidermoid carcinoma - clear cell variant (Fig 3C). Immunohistochemical examination of the specimen revealed focal positivity to S- 100, strongly positive for EMA, calcitonin negative and special stain mucarmine positive supporting our diagnosis. This was probably a second primary in the patient as there was no history of surgical management and a probable accidental seeding of the tumor into the mandible for the previously presented parotid tumor. Also, unlikely to be radiation induced as the patient underwent radiotherapy 45 yrs ago.

In the fourth case report, the young patient reported with an extra oral swelling of 4 months duration in the chin region with no other signs or symptoms. A history of gradual increase in size was reported. The lesion had a firm consistency and the general surgery department of our institution had made a provisional diagnosis of dermoid cyst and had referred him to our department to rule out any dental involvement. Intra-oral examination did not reveal any clinically significant findings. An incisional biopsy was suggestive of Fibrosarcoma. After a careful excisional of the lesion with appropriate marginal clearance, the final histopathological report also suggested the lesion to be fibrosarcoma. An Immuno-histochemical evaluation ruled out fibrosarcoma and a final diagnosis of proliferative fasciitis was reported. The application of Immuno-histochemical methods to histopathology has resulted in marked improvement in the microscopic diagnosis and has contributed to more accurate determination of patient prognosis as seen in this case as well.[7]

CONCLUSION

All these cases, presented to our department with unusual complaints, signs and symptoms. The accurate diagnosis and the subsequent management were possible only by following a systematic approach and use of advanced aids whenever necessary. These unusual presentations of cases stress on the importance of following a systematic approach to the differential diagnosis and thus aid in their appropriate management, Thus, improve the overall well-being and quality of life of patients.

Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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