International Journal of Pathology; 2013; 11(1): 26-28

Case Report

Cherubim: Fine Needle Aspiration Cytology Findings

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Abstract: A 52 years old lady presented with history of weakness and breathlessness on exertion. She noticed changes in her facial appearance around twenty years of age which continued to advance progressively over the last three decades. On physical examination her face showed advanced deformity and asymmetry in both horizontal and vertical planes. There was significant deformity of maxilla with prominent and unequal enlargement of malar eminences, stretched overlying skin and prominent superficial veins. The Fine Needle Aspiration Cytology from prominent right maxillary area was consistent with the diagnosis of cherubism.

Keywords: Cherubism, Fine Needle Aspiration Cytology, FNAC, Fibrous Dysplasia

Case presentation

A 52 years old lady was presented with history of weakness and breathlessness on exertion for the last three to four months. She had decreased appetite and insomnia for the last two months. According to her husband she had halitosis. She had history of tooth extraction 26 years ago. She noticed changes in her facial appearance around the age of 20 years. Those changes continued to advance progressively over the last three decades. Patient and her attendants are little concerned about those changes. She covers her face and she is more concerned about her present symptoms. She was married at the age of 18 year and had 3 sons and four daughters. There was no family history of significant illness. She was uneducated, non smoker and not addicted to any substance.

On physical examination, the patient was pale and had swelling and deformity of face. Her radial pulse was 88/ min and regular; BP 130/80 mmHg; Temp 98.6 F. Her JVP was not raised and she was not jaundiced and not cyanosed. There was no pedal edema. She had no palpable lymph nodes. The heart sounds were normal. Pulmonary examination revealed bilateral normal vesicular breath sounds.

The abdomen had scaphoid shape and no tenderness on deep palpation. There was no visceromegaly and no shifting dullness. The bowel sounds were normal.

Examination of musculoskeletal and nervous system revealed decreased muscle mass with no other abnormality.

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The face showed advanced deformity and asymmetry in both horizontal and vertical dimensions. There was significant deformity of maxilla with prominent and unequal enlargement of malar eminences, stretched overlying skin and prominent superficial veins. There was involvement of both orbits changing the alignment of eye balls. The right eye ball was pushed upwards and outwards. The more advanced swelling on the right side had encroached over the nose and mandible. The chin appeared to be pushed backwards and angle of mouth stretched towards right side. The left maxillary swelling was less prominent as compared with the right and left orbit was not involved. (Figure 1)

The x- rays of face showed sclerotic and cystic involvement of maxilla. Her complete blood count (CBC) showed hemoglobin of 9.6 gm /dl, MCV 56.4 fl, with normal leukocyte and platelet counts. The routine biochemistry, Liver Function Tests, Urine analysis and screening for Hepatitis B and C were normal. The patient's chest radiograph was normal.

The Fine Needle Aspiration Cytology (FNAC) was performed using a 23 gauge needle on right malar swelling. Adequate material was obtained which was spread on several slides. These were stained with hematoxylin and eosin as well as Field Stain. The smears showed many trabeculae of woven bone containing eosinophilic osteoid and fibrobalsts. Some osteoid woven trabeuclae had focal calcification. The phase contrast microscopy displayed remarkable 3 dimensional features of the woven osteoid trabeculae. (Figures 2-5)

The FNAC findings were consistent with cherubism.

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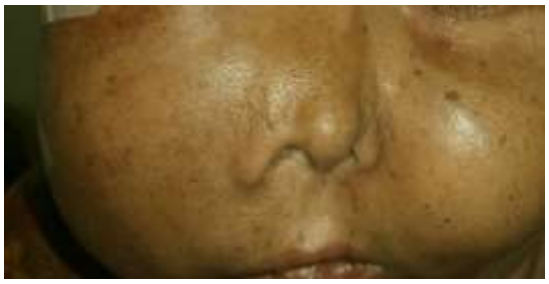

Figure 1. Gross Facial features

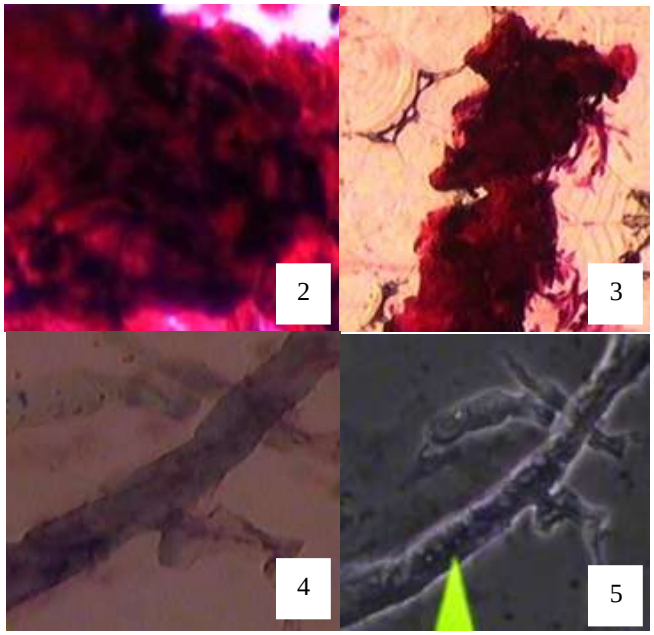

Figure 2: Fibroblasts & Osteoid (H&E X 100)

Figure 3: Woven bone: Osteoid with calcification (H&E X 100)

Figure 4: Woven bone trabecula (Field Stain X 100)

Figure 5: Woven Bone trabecula in phase contrast

Discussion

Cherubism is very rare, non-neoplastic, benign fibro osseous disease of facial bones[1]. It most commonly involves maxilla but mandible can also be involved. This fibro-osseous disease is characterized by painless expansion of these bones[2].

The condition is usually observed in young children and becomes more evident at the time of puberty[3]. It then gradually improves in the following years with advancing age. There are few case reports of this condition in elderly in 4th and 5th decades of life.

The disease was first described by Jones in 1933 as "familial multilocular cystic disease involving mandible and maxilla"[4]. It was given the name of "cherubism" due to round appearance of cheeks in this condition. Its mode of inheritance is autosomal dominant which appears to have 100% penetrance in males and 50–70% penetrance in females. There are however reports of non familial cases of cherubism in the literature[5].

There was no history of a similar disease in any of the family members of our patient, thus adding to the list of non-familial cases that have been reported in literature

The signs and symptoms of disease depend on the severity of the condition, and range from clinically and radiologically undetectable features to severe deformity of the jaws, upright palate, respiratory distress and impairment of vision[6]. The Mandible is involved more commonly than Maxilla. The involvement of orbits leads to deformity and mal-alignment of eye balls. The disease activity may remain progressive in orbits even after stabilization of disease in other areas[7]. This case was unique as lesions were large and aggressive.

The cherubism has been classified according to the severity of involvement by Seward and Hankey system[8].

- Grade I: Involvement of bilateral mandibular, malar region and ascending rami, mandible body or mentis.
- Grade II: Involvement of bilateral maxillary tuberosities as well as the lesion of grade I, diffuse whole mandible.
- Grade III: Massive involvement of the entire maxilla and mandible except the condyles.
- Grade IV: Involvement of both jaws with condyles.
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The clinical knowledge about the behavior of cherubism is based on a few case series and individual case reports. In most of these instances patients have not been followed for a sufficient duration of time to confirm the end results of disease progression. Our patient showed progressive deterioration with the passage of time with significant disfigurement and malalignment of eyeballs.

There are limited treatment options for this condition[9]. It may range from radical reconstruction surgery to conservative policy of wait and see. In the presence of wide spread involvement of major facial bones radical surgical procedures are not possible. The curettage has also shown poor results with tendency of relapse. The remodeling osteotomy is also not an option due to disrupted repair of pathological bones. The radiation therapy has high risk of developing osteosarcoma in irradiated area. There is no satisfactory medical treatment for this condition. Calcitonin may theoretically be useful as it has shown inhibition of bone resorption in cherubic

tissues[10]. It has not been approved for this indication and there is no clinical evidence of its efficacy.

Very few cases of FNAC of cherubism had been reported [11,12]. These cases have mainly shown spindly fibroblasts and osteoclasts. However beautiful osseous trabeculae with and with calcification are not mentioned. Osteoclasts were not observed in our case. Our case is unique in this regard as it showed numerous stunning osseous trabeculae and fragments of woven osseous tissue thus proving that a confident definite diagnosis of cherubism can be rendered on FNAC in correlation with clinical and radiological findings.

References

1. Hyckel P, Berndt A, Schleier P, Clement JH, Beensen V, Peters H, Kosmehl H. Cherubism-new hypothesis on pathogenesis and therapeutic consequences. J Cranio-Maxillofac Surg. 2005;33(1):61–68. doi: 10.1016/j.jcms.2004.07.006. [Cross Ref]
2. Katz JO, Dunlap CL, Ennis RL. Cherubism: report of a case showing regression without treatment. J Oral Maxillofac Surg. 1992;50(3):301–303. doi: 10.1016/0278-2391(92)90333-U. [PubMed] [Cross Ref]
3. De Tomasi DC, Hann JR, Shewart HM., Jr Cherubism: report of a non familial case. J Am Dent Assoc. 1985;111(3):455–457. [PubMed]
4. Jones WA. Further observations regarding familal multilocular cystic disease of the jaws. British Journal of Radiology. 1938;11:227–241. doi: 10.1259/0007-1285-11-124-227. [Cross Ref]
5. Ueki Y. et al. Mutations in the gene encoding c-Abl-binding protein SH3BP2 cause cherubism. Nat Genet. 2001;28(2):125–6. doi: 10.1038/88832. [PubMed][Cross Ref]
6. Kozakiewicz M, Perczynska-Partyka W, Kobos J. Cherubism--clinical picture and treatment. Oral Dis. 2001;7(2):123–30. [PubMed]
7. Colombo F. et al. Orbital involvement in cherubism. Ophthalmology. 2001;108(10):1884–8. doi: 10.1016/S0161-6420(01)00757-6. [PubMed][Cross Ref]
8. Seward GR, Hankey GT. Cherubism. Oral Surg Oral Med Oral Pathol. 1957;10(10):952–974. doi: 10.1016/S0030-4220(57)80047-4. [PubMed] [Cross Ref]
9. Kuepper RC, Harrigan WF. Treatment of mandibular cherubism. Journal of Oral Surgery. 1978;36(8):638–41. [PubMed]
10. Lannon DA, Earley MJ. Cherubism and its charlatans. Br J Plast Surg. 2001;54(8):708–11. doi:
11. Gupta N, Duggal R, Rajwanshi A, and Rattan V. Fine needle aspiration cytologic features of Cherubism: a case report. Acta Cytol. 2010 Jul-Aug;54(4):595-7
12. Jing X and Pu RT Fine-needle aspiration cytological features of Cherubism Diagn. Cytopathol. 2008;36:188–189