

Atypical Site of Fibromatosis in a Child: A Case Report

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Abstract: Fibromatosis is a benign locally infiltrating soft tissue tumor seen in middle age females. Most of them arise in abdomen especially involving abdominal wall and the extra abdominal sites include inguinal region, chest wall, shoulder region etc. We report a case of fibromatosis of submental region in a 9 year old girl and discuss imaging findings.

Key Words: fibromatosis- soft tissue masses- children

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I. Introduction

Fibromatosis also called as desmoid tumor constitute about 3 % of soft tissue Tumors. They are benign but show local invasion and recurrence after surgery without producing any metastases. The tumor show spindle shaped myofibroblastic proliferation, myxoid matrix, elongated vessels and inter cellular collagen fibers¹. Fibromatosis is grouped into superficial and deep types based on the location of the lesion. The superficial group include palmar fibromatosis, plantar fibromatosis, penile fibromatosis as they arise from the aponeurosis at these locations. They are seen mostly in men between 3-6 decade. The deep or desmoid -type fibromatosis is further divided into extrabdominal, intraabdominal and abdominal wall types. They are characterized by infiltration into adjacent structures and local recurrences². About 2 % of all desmoids are associated with Gardner syndrome. Extra abdominal and abdominal wall fibromatosis are seen mostly in women in 2-3 decades. Extraabdominal fibromatosis (desmoid tumor) is seen in women in the age group of 20-40 years and less than 5 % occur below 10 years. Though described to occur anywhere in the body, the more common sites include upper arm, chest wall, head & neck, thigh etc. They are generally solitary but multiple can occur. Desmoid tumors are locally aggressive invading adjacent muscles, vessels and the tumor extends along the fascial planes and recur after excision (recurrence rate 19-77 %).² Desmoid tumor appear as well defined hypoechoic mass on ultrasound with vascularity in 66 % cases. "Fascial tail sign" and staghorn pattern was described as specific in desmoids.³ CT scan findings are non specific. On MR the lesions are either well defined or infiltrative type which show intermediate signal intensity on T1 weighted sequences. Based on the tumor cellularity the lesions can show T2 hyper or isointensity signals. One of the characteristic imaging feature of desmoid on MR is the presence of low signal intensity band like regions which does not enhance with contrast and the specificity of this sign is 91 %.⁴ Another important typical feature is the fascial tail sign (linear extensions along the fascial planes).

II. Case Details

A 9 year old girl presented with painless swelling in the floor of the mouth since 6 Months. The mass was slow growing, non tender. There was mild discomfort during swallowing. Past history, family history were not significant. General examination was normal. Local examination revealed a non tender soft tissue mass in the submental location in the midline. High resolution ultrasound revealed a mixed echogenic solid mass 3.3 x 3.4 cms in the submental region which showed partial vascularity along with subcentimeter perilesional lymph nodes. MRI of neck showed a relatively well defined lobulated mass which was hypointense to muscle on T1 W, heterogeneously hyperintense on T2 Weighted sequences with moderate enhancement with Gd contrast administration. The mass was Diffusion restricted and showed no blooming on gradient sequence (Fig 1 A,B,C,D). Complete en block resection of the tumor was done. Microscopy description: Sections show a proliferation of spindle cells in long fascicles. They have uniform spindle nuclei with no significant pleomorphism, mitoses or necrosis. IHC: the spindle cells are positive for SMA. Beta catenin and S100 are negative. Impression: Morphology and immune profile consistent with myofibromatosis. (Fig 2)

III. Discussion

Desmoid type fibromatosis are poorly marginated painless solid masses. The head and neck is an unusual site for fibromatosis. Angiero^F and others⁵ reported a case of fibromatosis arising from hard palate and subcutaneous area of frontal bone in adult group. Another case of submandibular region fibromatosis was reported in a 32 year old male⁶. Aggressive fibromatosis in paediatric age group is extremely rare. Sharma and others⁷ reviewed a series of fibromatosis

in 10 patients in a 20 year old retrospective study and they concluded that nuclear beta-catenin expression is not a specific for fibromatosis diagnosis . Isolated cases of fibromatosis of head and neck in children involving tongue ⁸ , paramandibular region ⁹ has been reported in the literature. Histologically fibromatosis have variable patterns. The most common type is the presence of elongated spindle cells of uniform size with long sweeping fascicles in a collagenous stroma , prominent blood vessels with perivascular edema, variable mitotic rate. The other patterns include hypocellular with prominent hyalinization of stroma, myxoid , staghorn vessel, keloidal and nodular fasciitis-like etc. Presence of spindle cells in long fascicular arrangement is typical. The immunohistochemistry findings are typical .They include 100 % positivity for Cathepsin D and Vimentin , variable positivity for SMA,MSA, 70-98 % positivity for beta catenin, cyclin D1, calretinin. Negative for CD 34,S100, cytokeratins, ALK1, DOG1 . CTNNB1 mutations are reported in about 90 % cases .Desmoid type fibromatosis has to be differentiated from related conditions like fibroblastic sarcoma, reactive nodular fasciitis , scars, keloids based on Betacatenin which is strongly positive in 80% cases in the former condition ¹⁰ . Presence of poorly marginated solid soft tissue mass which show variable echogenicity on ultrasound and the presence of linear extensions along the adjacent fascial planes and iso to hyperintense to skeletal muscle on T1W and T2 Weighted sequences on MR with marked enhancement on contrast administration is a fairly reliable signs to diagnose in imaging.

Our case was a 9 year old girl presenting as painless swelling in the floor of the mouth which showed heterogenous echogenicity on ultrasound and enhancing T2 hyperintensity on MR is very typical of fibromatosis which was histologically confirmed. Surgical excision followed by radiotherapy is the main line of treatment to prevent local recurrences. For advanced disease, systemic Tamoxifen, NAIIDS have shown good response

IV. Conclusion

Extra abdominal desmoid type fibromatosis has to be considered in a female who present with poorly defined , infiltrating solid soft tissue mass.

References

- [1]. S.W.Weiss ,J.R.Goldblum, and F.M.Enzinger, “ Fibromatoses,” in Enzinger and Weiss’ soft tissue tumors, S.W.Weiss and J.R.Goldblum Eds. pp 227-228 Mosby Elsevier. Philadelphia Pa,USA 2008
- [2]. Eric A.Walker , JonelleM.Petscavage, Pamela L.Brian, Chika IloanusiLogie, Kenneth M.Montini and Mark D.Murphey imaging features of superficial and deep fibromatoses in the adult population Hindawi publishing corporation 2012 sarcoma vol article id 215810 .
- [3]. C.C.Huang, S.F.Ko, M.C.Yeh et al, “ aggressive fibromatosis of the chest wall: sonographic appearance of the fascial tail and staghorn patterns ”Journal of ultrasound in medicine vol28, no 3 pp393-396 2009.
- [4]. J.C.Lee, J.M.Thomas ,S.Phillips,C.Fisher and E.Moskovic “ Aggressive fibromatosis: MRI features with pathologic correlation ”American Journal of Roentgenology vol 186 no 1 pp247-254, 2006
- [5]. Angiero F, Benedicenti S, Stefani M Fibromatosis of the head and neck : morphological , immunohistochemical and clinical features Anticancer Res 2008 may-jun : 28 (3B) : 1725-32
- [6]. Parthiban N, O’Dwyer T : Fibromatosis of submandibular region J Surg Tech Case Rep 2014 jan 6(1) 21-5 2006
- [7]. Sharma A, NganB.Y,SandorGK,CampisiP,Forte V Pediatric aggressive fibromatosis of the head and neck: a 20 year retrospective review J PediatrSurg 2008 43(sep ; 43(9) 1596
- [8]. Miyashita H,Asoda s, Soma T, Munakata K, Yazawa M, NakagawaT, Kawana H Desmoid type fibromatosis of the ehad and neck in children: a case report and review of the literatue J Med Case Rep 2016 jun 10
- [9]. Morillo C, Ruiz DieguezPR,NavarroM,Galeas Anaya FJ, Ruiz Delgado F, Valienta Alvarez. aggeressivefibromatosisi of the head and neck in pediatric age. a case report and review of the literature Cir Pediatr 2014 oct 25 (4) 213-207
- [10]. Bernd Kasper, Philipp Strobel , Peter Hohenbergerdesmoid tumors: clinical features and treatment options for advanced disease the oncologist 2011 ; 16 : 682-693.

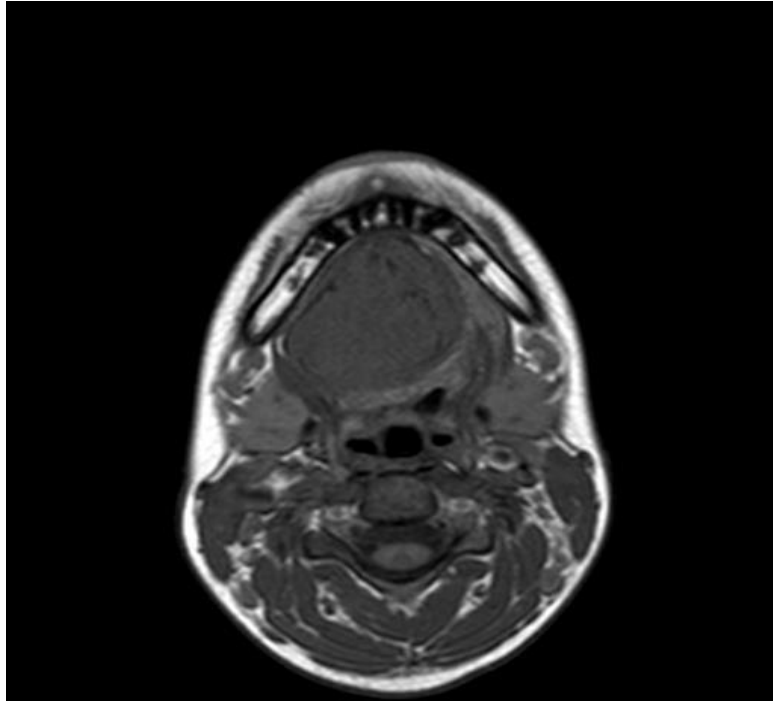


Fig 1 A

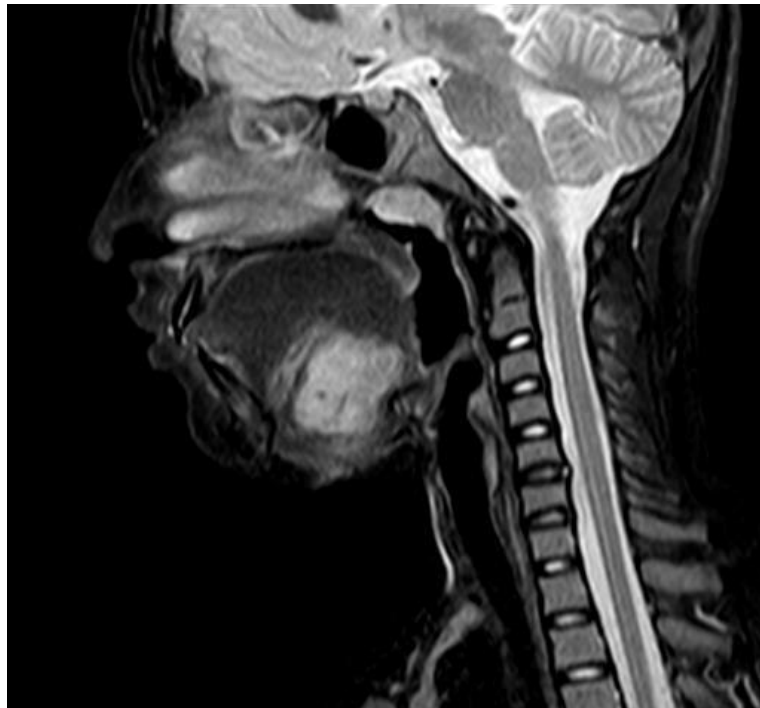


Fig 1 B

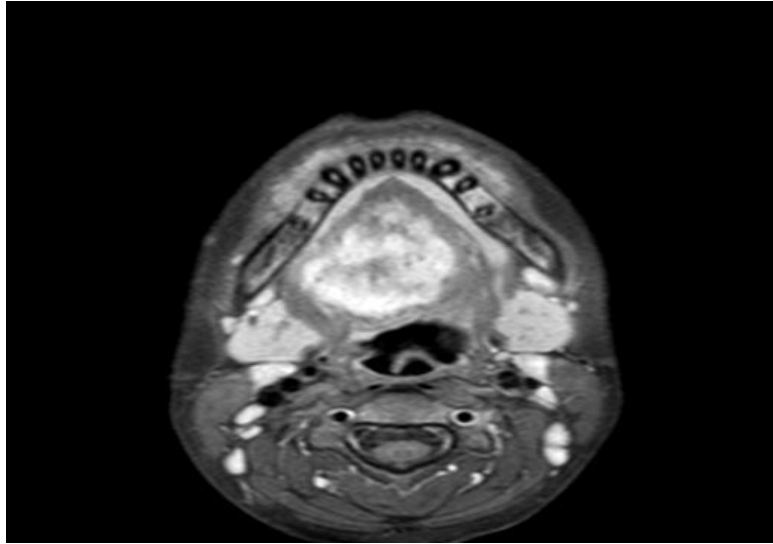


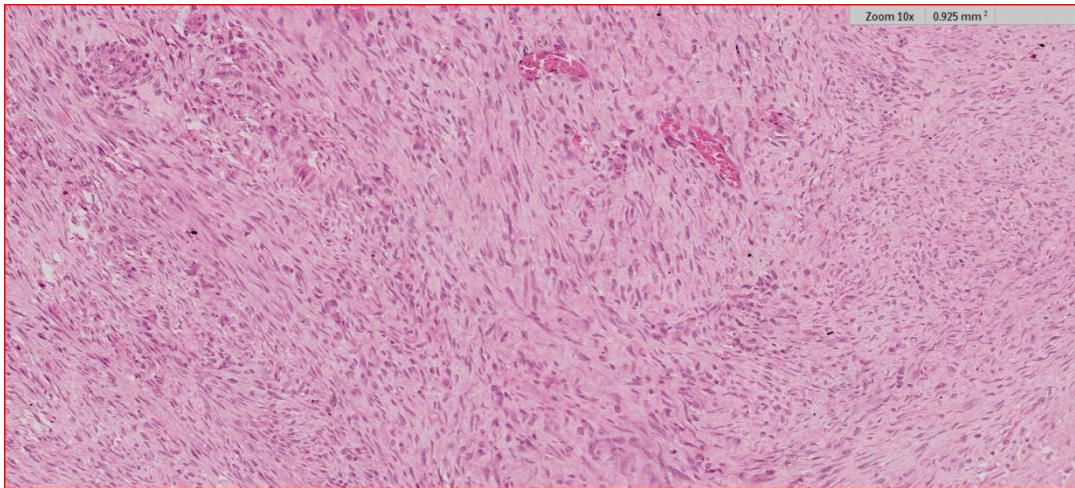
Fig 1 C



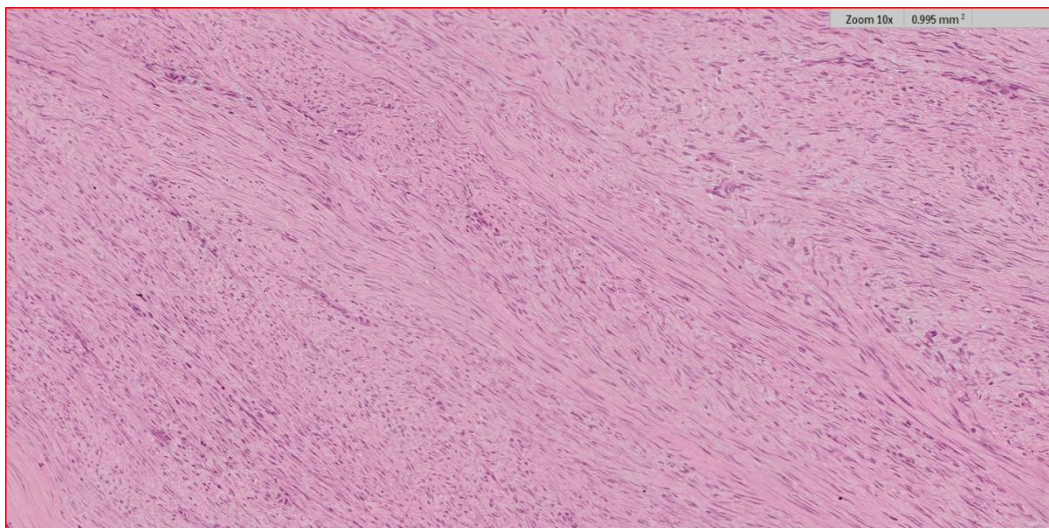
Fig 1 D



Fig 2 A.



B)



C)

LEGENDS: Fig 1 (A-D) : MRI images of neck showing soft tissue mass which is hypointense on T1 W and heterogenous on T2 W sequence with enhancement on contrast.

Fig 2 (A,B C) : Proliferation of uniform spindle cells in a collagenous stroma (H and E).

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