Case Report

www.ijrhs.com
ISSN (o):2321–7251

Desmoid Fibromatosis: A neonatal presentation

Varun Hathiramani[1], ShrutiTewari[2], AnantBangar[3], VikramMardhekar[4], Rajeev G Redkar[5]

Department of Pediatric Surgery,Lilavati Hospital and Research Centre, Mumbai.1- 6th year resident, 2- 1stYear Resident, 3- Junior Consultant. 4- Consultant Pediatrician and Neonatologist. 5- Consultant Pediatric Surgeon

Corresponding Address: Dr. Rajeev G Redkar, Consultant Pediatric Surgeon Lilavati Hospital and Research Centre, Mumbai, India.Affiliated to: Wadia Children's Hospital, Honorary Consultant Pediatric Surgeon, and Shushrusha Hospital- Consultant Pediatric Surgeon.E mail id: rajeev.redkar@gmail.com

Abstract:

Desmoid tumors describe a rare monoclonal, fibroblasticproliferation characterized by a variable and oftenunpredictable clinical course. Although histologicallybenign, desmoids are locally invasive and associatedwith a high local recurrence rate, but lack metastaticpotential.Although desmoid tumors are more common in persons aged 10-40 years than in others, they do occur in young children and older adults, but have rarely been reported in the neonatal age group. We report here a 17 day child who presented with a cervical swelling whose histology was desmoid fibromatosis.

Key words: Desmoid fibromatosis, neonatal presentation

Introduction:

The term desmoid tumour describes a fibromatous proliferative disease that in its biological behaviour is classified between benign fibrous tissue proliferation and fibrosarcoma. According to the World Health Organization, desmoid tumors are defined as "clonal fibroblastic proliferations that arise in the deep soft tissues and are characterized by infiltrative growth and a tendency toward local recurrence but an inability to metastasize" [1].

Case Report

A 17 day male child born of a non-consanguineous marriage presented with a gradually increasing mass in the left supra-clavicular region of the neck which was noticed when the child was seven days old. As the swelling increased in size the child kept the head turned towards the right side. Other than some redness associated with the swelling there was no discharge, difficulty breathing or feeding, decrease in activity, lethargy or excessive crying.

On examination a 2x3 cm sized, round, mobile hard swelling was present. There was redness of the overlying skin but the swelling was non-tender with no transmitted pulsations or movement with deglutition or adherence to overlying skin or underlying structures.

The child was shown to a local physician who advised an ultrasound scan of the neck which showed two closely placed oval masses noted anterior to left jugular vein. They were firm to hard non-compressible lesions measuring about 2.7 x 1.1 cm which raised a possibility of it being either an enlarged lymph node or possibly a sternocleidomastoid tumour. As lymphadenopathy is a rare at this age the child was referred to us for further evaluation.

We further investigated the child with a MRI of the neck to make sure what pathology we were dealing with. The MRI showed two hyperintense lesions [11.3 mm x 26.9 mm x 17 mm and 29.3 mm x 17.7 mm x 16.9 mm] on the left side of the neck deep to the sternocleidomastoid muscle superior to the sternum and in continuity with the anterior portion of the thymus. There was posterior displacement of the trachea and anterior displacement of the right innominate artery. In view of these findings we decided to perform an excisional biopsy of the lesion to ascertain the diagnoses.

The intra and post-operative course was uneventful. The mass was completely excised without damage to any vital structures and the histopathology showed myofibroblastic proliferation suggestive of desmoid fibromatosis with clear margins. The child is on regular follow up and is doin well.

Discussion

Desmoid-type fibromatoses are rare tumors, accounting for less than 0.03% of all neoplasms, and less than 3% of soft tissue neoplasms in the general

population [2, 3]. Incidence of DTF is estimated at 2-4 per million per year. This benign but monoclonal proliferative soft tissue lesion arises from deep fascia or soft tissues and is derived from mesenchymal stem cells [4, 5]. Two relative incidence peaks are reported in the literature: 6 to 15 years and between puberty and the age of 40[6].

The risk factors associated with the development of desmoid-type fibromatosis include high oestrogen states [like pregnancy], prior surgical trauma, and Gardner's syndrome, which is caused by inherited mutations in the adenomatous polyposis coli [APC] tumour suppressor gene. The most common tumour sites are the extremities [shoulder and thigh], trunk [chest wall and back], mesenteryand head and neck .Among cases of desmoid fibromatoses 7 to 15 % of tumors occur in the head and neck [7-9]. A literature search suggested that neonatal presentation of this tumour is rare and very few cases have been reported.

The clinical presentation is variable depending on location and extent of the lesion. Patients with extra-abdominal desmoid fibromatoses may complain of a slow-growing, deep-seated, firm, painless or minimally painful mass. Alternatively, depending on lesion location, patients may complain of neurologic symptoms or even decreased mobility if joints are involved. Patients with intra-abdominal desmoid fibromatoses, particularly mesenteric lesions, may complain of some mild abdominal pain or simply an asymptomatic abdominal mass. Less frequently, these patients may present acutely with bowel perforation, obstruction, or gastrointestinal bleeding as a result of local desmoid tumour invasion. The clinical behaviour of DTF is varied, with some lesions spontaneously regressing[10].

Imaging modalities like ultrasound, CT scan are non-specific where the lesion could be homogeneous or mildly heterogeneous on USG and hypodense or isodense compared to muscle on CT. MRI shows the lesion to be hypointense on T1 images and hyperintense on T2 images [11].

A paper on the European Organisation for Research and Treatment of Cancer [EORTC]/Soft Tissue and Bone Sarcoma Group's position on desmoid fibromatoses reported that a "watch and wait strategy" is the first choice in the treatment of desmoid fibromatoses in all populations and that resection with a clear margin should be considered to be a treatment option if postoperative morbidity is acceptable [12]. Although complete resection [negative microscopic margin] of the tumour is thought to be the reference standard for successful treatment in patients with DF uncontrolled by other treatment approaches, resection in the head and neck region is often difficult because of the presence of vital structures [13].This problem is worse in paediatric patients. In paediatric patients with desmoid fibromatoses uncontrolled by non-surgical treatments, surgeons sometimes are concerned about performing wide resections because of the high potential of postoperative morbidity. When a large tumour exists close to a vital structure, surgery with an adequate safety margin may be challenging. Although some successful cases with incomplete resection or without surgery have been reported [14-19], a randomized trial of treatment strategies has not yet been reported.

Other treatment alternatives include radiation therapy, chemotherapy and hormonal therapy. Drugs like methotrexate and vinblastine for low grade tumours and cyclophosphamide, doxorubicin, dacarbazine for aggressive lesions have been used. In slowly growing or mildly symptomatic tumours non-cytotoxic drugs, such as anti-estrogens,NSAIDs, imatinibmesylate, hydroxyurea, and interferon-α, orretinoic acid can be effective [20, 21]. Hormonal therapy[tamoxifen, toremifene] has also been used in paediatricpatients, but with controversial results. These therapies canbe combined with chemotherapy with benefit but the longtermclinical improvement is minimal [22] and the lateeffects in young children are unknown.

Figure 1: Clinical picture showing swelling in left supra-calvicular region

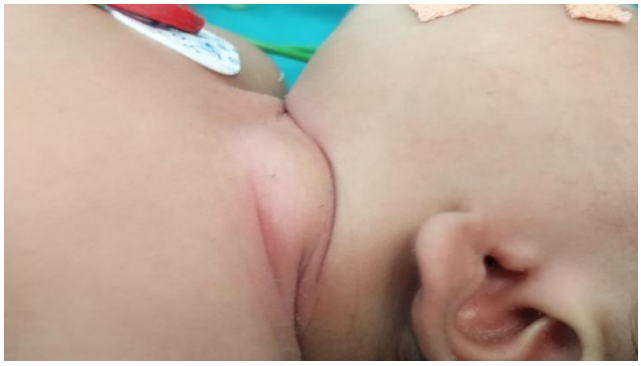


Figure 2: MRI hyper intense lesions deep to the left sternocleido mastoid muscle

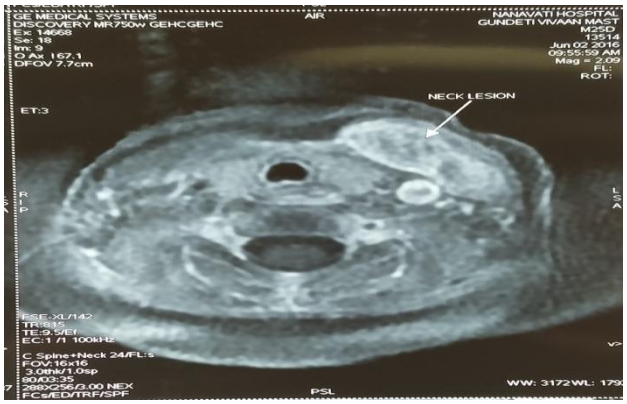


Figure 3: Intra-operative picture showing superficial and deep components of the swelling

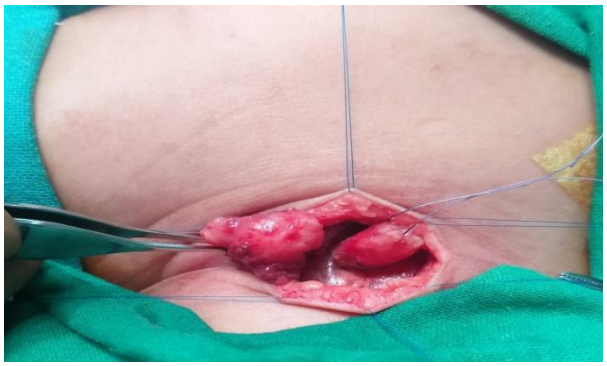
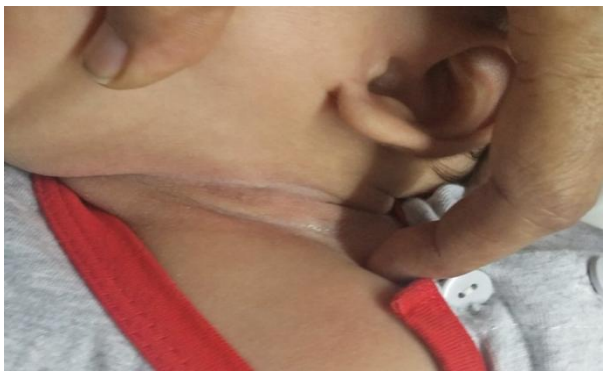


Figure4: Post-operative picture showing a healthy scar with no swelling



Conclusion

Desmoid fibromatosis is a locally invasive but non-metastatic disease. Surgical excision with clear margins is the best modality of treatment. In cases where surgical excision carries a significant risk of morbidity additional modalities like limited resection and close follow up, radiation therapy, chemotherapy can be incorporated in the patient's management.

Source of Funding: Nil
Source of Conflict: Nil

Acknowledgement

The authors are grateful to authors/editors/publishers of all those articles, journals and books from where the literature is discussed.

References

1. Biermann JS. Desmoid tumors. Curr Treat Options Oncol 2000;1:262–266.

2. Escobar C, Munker R, Thomas JO, Li BD, Burton GV. Update on desmoid tumors. Ann Oncol. 2012 Mar;23[3]:562-9.

3. Reitamo JJ, Hayry P, Nykyri E, Saxen E. The desmoid tumor. I. Incidence, sex-, age- and anatomical distribution in the Finnish population. Am J ClinPathol. 1982 Jun;77[6]:665-73.

4. C. A. Kingston, C. M. Owens, A. Jeanes, and M. Malone,"Imaging of desmoid fibromatosis in pediatricpatients,"American Journal of Roentgenology, vol. 178, no. 1, pp. 191–199, 2002.

5. C. Wu, S. Nik-Amini, P. Nadesan, W. L. Stanford, and B. A.Alman, "Aggressive fibromatosis [desmoid tumor] is derivedfrom mesenchymal progenitor cells," Cancer Research, vol. 70,no. 19, pp. 7690–7698, 2010.

6. C. Meazza, G. Bisogno, A. Gronchi et al., "Aggressive fibromatosisin children andadolescents: the Italian experience,"Cancer, vol. 116, no. 1, pp. 233–240, 2010.

7. Barnes L, Eveson JW, Reichart P, Sidransky D. World Health Organization classification of tumors. Pathology and genetics of head and neck tumors. Lyon: IARC; 2005.

8. Baumert BG, Spahr MO, Von Hochstetter A, et al. The impact of radiotherapy in the treatment of desmoid tumours. An international survey of 110 patients. A study of the Rare Cancer Network. RadiatOncol. 2007;2:12.

9. Tse GM, Chan KF, Ahuja AT, King AD, Pang PC, To EW. Fibromatosis of the head and neck region. Otolaryngol Head Neck Surg. 2001;125:516–9.

10.Julia A Ross, Xuchen Zhang. Soft Tissues: Desmoid-type fibromatosis. Atlas of Genetics and Cytogenetics in Oncology and Haematology 2013

11. Georg F. Eich, Jean-Claude Hoeffel, Heinz Tschappeler, Ingmar Gassner, Ulrich V. Willi. Fibrous tumours in children: imaging features of a heterogeneous group of disorders. PediatrRadiol 1998;28:500-9

12. Kasper B, Baumgarten C, Bonvalot S, et al. Management of sporadic desmoid-type fibromatosis: a European consensus approach based on patients' and professionals' expertise – a sarcoma patients EuroNet and European Organisation for Research and Treatment of Cancer/Soft Tissue and Bone Sarcoma Group initiative. Eur J Cancer. 2015;51:127–36.

13. De Bree E, Zoras O, Hunt JL, et al. Desmoid tumors of the head and neck: a therapeutic challenge. Head Neck. 2014;36:1517–26.

14. Humar A, Chou S, Carpenter B. Fibromatosis in infancy and childhood: the spectrum. J Pediatr Surg. 1993;28:1446–50.

15. Woltsche N, Gilg MM, Fraissler L, et al. Is wide resection obsolete fordesmoid tumors in children and adolescents? Evaluation of histological margins, immunohistochemical markers, and review of literature. PediatrHematolOncol. 2015;32:60–9.

16. Gronchi A, Casali PG, Mariani L, et al. Quality of surgery and outcome inextra-abdominal aggressive fibromatosis: a series of patients surgically treated at a single institution. J ClinOncol. 2003;21:1390–7.

17. Honeyman JN, Theilen TM, Knowles MA, et al. Desmoid fibromatosis inchildren and adolescents: a conservative approach to management. J Pediatr Surg. 2013;48:62–6.

18. Lev D, Kotilingam D, Wei C, et al. Optimizing treatment of desmoid tumors.JClinOncol. 2007;25:1785–91.

19. Wang CP, Chang YL, Ko JY, Cheng CH, Yeh CF, Lou PJ. Desmoid tumorofthe head and neck. Head Neck. 2006;28:1008–13.

20. J. Mace, J. Sybil Biermann, V. Sondak et al., "Response of extraabdominal desmoid tumors to therapy with imatinibmesylate," Cancer, vol. 95, no. 11, pp. 2373–2379, 2002.

21. C.Meazza,M. Casanova, G. Trecate, and A. Ferrari, "Objectiveresponse to hydroxyurea in a patient with heavily pre-treated aggressive fibromatosis," Pediatric Blood and Cancer, vol. 55, no. 3, pp. 587–588, 2010.

22. H. Lackner, C. Urban, R. Kerbl, W. Schwinger, and A. Beham, "Noncytotoxic drug therapy in children with unresectable desmoid tumors," Cancer, vol. 80, no. 2, pp. 334–340, 1997.