Desmoid Fibromatosis of the Maxilla

Shiva P. Daram, MD1, Charles Timmons, MD, PhD2,3, Ron B. Mitchell, MD1,3, and Gopi Shah, MD1,3

Introduction

Desmoid fibromatosis (DF) is a benign fibroblastic neoplasm arising from connective tissue, characterized by significant local destruction and invasion without potential for metastasis.1,2 In children, it typically presents as a painless, rapidly growing mass in the limbs or trunk and is rarely seen in the head and neck. We present a facial mass that was initially diagnosed as cranial fasciitis (CF) on biopsy, but was diagnosed as DF after resection. This case study highlights the close pathologic relationship between DF and CF and the challenges in the treatment of DF.

Case Report

A 5-month-old male presented with a 2-month history of a rapidly enlarging facial mass. The parents reported nasal congestion without fevers, drainage, or trauma. Computerized tomography (CT) showed a 19-mm mass adjacent to the right infraorbital nerve, with no definite extension to the orbit or nasolacrimal duct (Figure 1A). Magnetic resonance imaging (MRI) showed an ill-defined lesion in the right infraorbital soft tissues, hypointense on T1-weighted images, and heterogeneously enhancing with contrast.

The baby underwent an intraoral incisional biopsy. The cells displayed aberrant nuclear staining for β-catenin, with concern for more aggressive behavior (Figure 2A and B). The CF was diagnosed. The mass enlarged over 3 months; thus, a sublabial gross total excision of the mass was performed. The final diagnosis was DF with positive surgical margins (Figure 2C). The mass recurred 1 month later. After discussion at tumor board, the patient started weekly methotrexate and vinblastine chemotherapy. At 24 weeks, MRI showed a significant response to chemotherapy (Figure 3). The plan is for chemotherapy every 2 weeks, until the mass completely resolves or stabilizes with imaging every 3 to 6 months.

Figure 1. A, CT noncontrast axial section shows a 1.9 cm mass adjacent to infraorbital nerve (star) with underlying bony erosion (arrow). B, Postcontrast T1 axial MRI scan shows hypointense mass with underlying bony scalloping (arrow). CT indicates computed tomography; MRI, magnetic resonance imaging.
Desmoid fibromatosis is a locally destructive, benign, fibroblastic neoplasm arising from connective tissue seen in the head and neck in 7% to 15% of cases. These lesions typically occur in a bimodal distribution in infants and young adults without a gender predilection. The etiology of DF is unknown, but trauma and hormonal influences have often been cited as possible causative factors. The DF has also been associated with hereditary genetic syndromes such as familial adenomatous polyposis and hereditary desmoid disease.

The differential diagnosis for a scalp or facial mass in a child includes hemangioma, dermoid cyst, fibrous dysplasia, Langerhans cell histiocytosis, CF, rhabdomyosarcoma, osteosarcoma, or Ewing sarcoma. The DF can be distinguished from other lesions by history, radiological features, or biopsy.

Microscopically, DF has spindle-shaped fibroblast-like cells without atypia, surrounded by abundant collagen, with rare mitoses and no necrosis. It typically stains with vimentin and smooth muscle actin, but aberrant nuclear staining of β-catenin is highly specific in differentiating DF from other similar tumors. However, CF also has similar histological staining including aberrant staining of β-catenin. Due to the higher rates of recurrence seen in cases of CF that stain positive for β-catenin, it may exist on a continuum with, or may transform into, DF.

The DF is classified as intermediate grade by the World Health Organization due to its locally aggressive natural history. Traditional treatment has been wide local excision with negative surgical margins to decrease recurrence. Meazza et al reported a 22% versus 76% recurrence rate with DF with negative compared to positive surgical margins. This has led to the use of adjuvant chemotherapy when positive margins are reported or recurrences occur, for tumor stabilization and regression. However, studies using chemotherapy in primary and recurrent head and neck DF have reported resolution of DF after incomplete resection with positive margins and have not advocated adjuvant treatment with positive margins. Instead, they recommend salvage surgery for clinical recurrences and chemotherapy only for unresectable primary or progressive recurrent disease.

A European Organization for Research and Treatment of Cancer position paper recommends a watch-and-wait approach to head and neck DF. For progressive cases, they recommend medical therapy, nonsteroidal anti-inflammatory drugs and antiestrogen medication, or chemotherapy. However, studies using chemotherapy in primary and recurrent head and neck DF...
show low rates of complete response, but good rates in stopping disease progression.\textsuperscript{14}

In our case, the lesion was initially diagnosed as CF. With the rapid recurrence of the mass, gross total excision was performed and pathology confirmed DF. Its rapid regrowth further complicated treatment options. The literature presents 2 options for children with DF, wide local excision, or chemotherapy.\textsuperscript{3,4,11} Given the morbidity that would be associated with wide local excision of the maxilla in an infant, the decision was made to pursue adjuvant treatment with chemotherapy, with good response since initiation of treatment.

Conclusion
Desmoid fibromatosis is a locally aggressive entity, rarely seen in the head and neck, with little consensus on treatment strategy. Cranial fasciitis positive for $\beta$-catenin is closely related to DF, and cases that do not resolve spontaneously or recur rapidly should be reanalyzed for DF. Management options for DF include observation, surgical resection, medical therapy, and chemotherapy.

Declaration of Conflicting Interests
The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding
The author(s) received no financial support for the research, authorship, and/or publication of this article.

References
1. Buitendijk S, van de Ven CP, Dumans TG, et al. Pediatric aggressive fibromatosis: a retrospective analysis of 13 patients and review of literature. \textit{Cancer.} 2005;104(5):1090–1099.
2. Sharma A, Ngan BY, Sandor G, Campisi P, Forte V. Pediatric aggressive fibromatosis of the head and neck: a 20-year retrospective review. \textit{J Pediatr Surg.} 2008;43(9):1596–1604.
3. Miyashita H, Asoda S, Soma T, et al. Desmoid-type fibromatosis of the head and neck in children: a case report and review of the literature. \textit{J Med Case Rep.} 2016;10:173–181.
4. Tostevin PMJ, Wyatt M, Hosni A. Six cases of fibromatosis of the head and neck in children. \textit{Int J Pediatr Otorhinolaryngol.} 2000;53(3):235–244.
5. Lips DJ, Barker N, Clevers H, Hennipman A. The role of APC and beta-catenin in the aetiology of aggressive fibromatosis (desmoid tumors). \textit{Eur J Surg Oncol.} 2009;35(1):3–10.
6. Curtin E, Caird J, Murray DJ. Cranial fasciitis located at the temporal region in a 2-year-old girl. \textit{Childs Nerv Syst.} 2014;30(12):2163–2167.
7. Wagner RD, Wang EK, Lloyd MS, Lam SK, Khechoyan DY. Cranial fasciitis: a systematic review and diagnostic approach to a pediatric scalp mass. \textit{J Craniofac Surg.} 2016;27(1):e65–e71.
8. Rakheja D, Cunningham JC, Mitui M, Patel AS, Tomlinson GE, Weinberg AG. A subset of cranial fasciitis is associated with dysregulation of the Wnt/beta-catenin pathway. \textit{Mod Pathol.} 2008;21(11):112.
9. Woods TR, Cohen DM, Islam MN, Rawal Y, Bhattacharyya I. Desmoplastic fibroma of the mandible: a series of three cases and review of the literature. \textit{Head Neck Pathol.} 2015;9(2):196–204.
10. Meazza C, Bisogno G, Gronchi A, et al. Aggressive fibromatosis in children and adolescents. \textit{Cancer.} 2010;116(1):233–240.
11. Wang CP, Chang YL, Ko JY, Cheng CH, Yeh CF, Lou PJ. Desmoid tumor of the head and neck. \textit{Head Neck.} 2006;28(11):1008–1013.
12. Stoeckle E, Coindre JM, Longy M, et al. A critical analysis of treatment strategies in desmoid tumours: a review of a series of 106 cases. \textit{Eur J Surg Oncol.} 2009;35(2):129–134.
13. Kasper B, Baumgarten C, Bonvalot S, et al; Desmoid Working Group. Management of sporadic desmoid-type fibromatosis: a European consensus approach based on patients’ and professionals’ expertise – a sarcoma patients EuroNet and European Organization for Research and Treatment of Cancer/Soft Tissue and Bone Sarcoma Group initiative. \textit{Eur J Cancer.} 2015;51(2):127–136.
14. Skapek SX, Ferguson WS, Granovetter L, et al; Pediatric Oncology Group. Vinblastine and methotrexate for desmoid fibromatosis in children: results of a Pediatric Oncology Group Phase II trial. \textit{J Clin Oncol.} 2007;25(5):501–506.