Craniofacial Mass in a Pediatric Patient: A Diagnostic Dilemma

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BACKGROUND INFORMATION
Fibrous dysplasia is a disease of bone wherein the cartilaginous matrix is replaced with fibrous connective tissue. This process can occur in one bone (monostotic), two or more bones (polyostotic), or can be part of a triad known as McCune-Albright Syndrome (MAS) that includes polyostotic FD along with café-au-lait skin macules and endocrinopathies, typically precocious puberty.12 Craniofacial polyostotic FD and MAS occurs in the temporal bones >70% of the time, which is in contrast to those patients with monostotic FD where temporal bone involvement is less common.45 Temporal bone involvement is uncommon in MFD.6 One of the difficulties in diagnosing FD relates to the wide range of symptoms associated with the disease. In one cohort of 66 FD patients, only 63% were detected based upon investigation of presenting symptoms.11

CASE PRESENTATION
Our patient presented to El Paso Children’s Hospital following a six month history of dizziness, nausea, and vomiting. She also described that in the last three months she had syncope several times during the week, lasting between 1-5 minutes and none with seizure like activity. There have been no changes with her weight, appearance, social activities, or attitude. Her description of the dizziness did not follow a vertigo pattern. She described a five month history of bilateral maxillary discomfort, which she and her family initially attributed to her active orthodontic care. She is being treated with orthodontic appliances as well as a palatal expander. She has had headaches for three months occurring 3-4 times a week that are located in the temporal region bilaterally as well as in the temporomandibular joint areas that were non-throbbing and last under one minute.

She started menstruation at 12 years old and describes normal flow and regular pattern. She is in the 8th grade, has exceptional academic standing, involvement with three athletic and four extracurricular organizations. Along with her parents she notes that she is always under a lot of pressure to perform well in all aspects of life. All cranial nerves were intact, normal funduscopic and otoscopic examinations. She currently has no abnormal skin pigmentation, nor any history of skin pigmentation issues. She had extreme difficulty arising from bed and had to be supported or else she would have fallen from the onset of dizziness.

Cardiology found no abnormalities and she had a normal 24-hour Holter monitor EKG. Neurology noted no gross abnormalities and she had a normal EEG. A psychiatry consultation was able to further identify significant life stressors that they believed could be associated with her symptoms. Most of her conflicts stemmed from her parents three year history of divorce and subsequent remarriage, as well as the academic and social stresses her parents and she herself had. She has many friends, is comfortable with her weight (BMI 19.3), and even notes that she wants to increase her weight if that could help her with her athletics. Psychiatry ruled out suicidal ideations and intragynic causes of her dizziness and syncope.

CT and subsequent MRI illustrated mixed radiolucent/radiopaque lesions in the left temporal bone and right lateral maxilla. The inner ear anatomy appeared normal and there was no narrowing of the external auditory canal. A bone scan similarly showed these two lesions as well as a lack of lesions elsewhere.

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Figure 1 & 2: Coronal CT images showing bony involvement of the left temporal, right maxilla, and bilateral sphenoid/zygoma.
After biopsy, microscopy confirmed the diagnosis of fibrous dysplasia.

Based on her lack of vertigo and lack of cranial nerve involvement, surgical correction was deferred. With advice from psychiatry, the team and patient agreed to initiate a several month trial of individual and family therapy sessions as well as an overall decrease in extracurricular involvement. We also hope to work with her school to better accommodate her shortened attendance ability so that she will not fall behind in her education, which would further compound the academic and social stresses she is experiencing.

**DISCUSSION**

A female teenager presenting with a chronic history of nausea, vomiting, dizziness, syncope, and significant social/academic stressors initially causes one to suspect hypovolemia and electrolyte disturbances possibly associated with an eating disorder. Once an eating disorder was ruled out, through a thorough history and physical in addition to her basic metabolic panel values all within reference ranges, our team still focused on two suspicious aspects of our patient. One of these was her family dynamic: divorced parents who apparently continually emphasize academic and athletic perfection and openly argued with the other regarding their daughter’s history of symptoms. Through the consultation of our pediatric psychiatry department, we were able to suspect that some of her symptoms, while unintentional, might be related to the external stressors in her life. Psychiatry did not suspect her to have a factitious disorder.

Another issue was her bilateral maxillofacial discomfort. This symptom of maxillofacial pain is noted in less than half of those with a maxillofacial FD. Our patients noted that her orthodontist dismissed her discomfort, maybe appropriately, as being due to her active orthodontic corrections but a further investigation might have allowed earlier detection and possible avoidance of the syncopal episodes and better anticipation of inability to maintain school attendance.

One of the major concerns for patients of any age groups is how such a lesion might progress. According to more than 25 years of research at the NIH, it appears that MFD does not progress to PFD and neither progress to malignant osteosarcoma. Additionally, recent clinical guidelines published in 2011 noted that “malignant change to osteosarcoma or other forms of sarcoma has been reported to occur in less than 1% of cases of FD”. Such information is especially important in a patient such as ours due to an already high level of family stress that we are trying to alleviate.

Another important aspect of FD is that it typically is able to be identified early in the disease process. A 2007 study showed that in 90% of FD lesions are present prior to 15 years of age. This study also showed 90% of craniofacial FD lesions can be detected via bone scan by 3.4 years old, and usually there will be no new FD lesion occurring at this region once the patient is older than 10 years.

The treatment for FD is one that still deserves much research. While some studies have shown bisphosphonate therapy can alleviate the child’s pain, it produces no appreciable radiographic or histomorphometric changes in the FD lesions. Such treatment options have been questioned due to a possible link between bisphosphonate use and osteonecrosis of the jaw. At present the long term effects of bisphosphonates on the growing skeleton are not known. Interestingly FD in adults is more likely to cause significant pain (severity 4.1/10, and 2.8/10, respectively) and the pain that does occur in kids is usually undertreated (44% vs. 26%). With this information it should be noted that while 67% of the patients in this study did complain of pain, the study did not find a correlation between pain severity and skeletal disease burden. For our patient specifically, we believed that a trial of psychosocial therapy would be the least invasive and possibly most revealing based upon the emotional aspects of her disease burden. If our trial of individual and family therapy fails to alleviate her symptoms then we plan on revisiting her options of bisphosphonate therapy, surgical correction, or watchful waiting.

**CONCLUSION**

Fibrous dysplasia in children does not follow a pathognomonic presentation. Features such as chronic dizziness, nausea, vomiting, and syncope, especially in the absence of intentional weight loss, should raise the suspicion of a craniofacial lesion. A 2005 study illustrated how one of the most concerning aspects for the parents of patients with FD appears to be the fear that their child might have significant impairment with ambulation. This fear should be addressed early in the treatment dialogues. Additionally, the role that the child’s FD has on the emotional wellbeing of the parents should not be underestimated as they too can have significant emotional impairment. It is important to reassure the patient and the family that 75% of the disease usually has already been established by the time the child is 10 years of age. Furthermore, it has been shown that FD patients generally achieve a high level of social and emotional functioning. Knowledge of fibrous dysplasia, its natural history, proper diagnosis, and treatment will hopefully aid physicians and families by having awareness and a healthy level of suspicion in the pediatric population.

**REFERENCES**


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