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Case Report

Stigma, Fear, and Acceptance: Three Phases of the Fibromuscular Dysplasia Experience

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Abstract

Fibromuscular Dysplasia (FMD) is an arteriopathy that can affect any vascular territory, though most often affects the renal and carotid arterial beds. Symptoms are consistent with the vascular bed affected and are often attributed to other conditions, leading to delays in diagnosis. We present a case of a middle-aged woman who presented to the emergency department (ED) with altered mental status and confusion, with an ED discharge diagnosis of altered mental state secondary to depression and alcohol abuse. During follow-up testing, she was diagnosed with hypertensive encephalopathy and, subsequently, FMD.

The true prevalence of FMD is unknown, although current estimates vary from 3-4%. Like other "uncommon" disorders, appropriate diagnoses are often delayed or missed altogether. This delay to diagnosis, in combination with non-specific symptoms and providers' unfamiliarity with FMD, leads to frustration for many patients. A recent FMD qualitative publication revealed patients' concerns about physical symptoms along with greater frequency of anxiety and depression. Importantly, these concerns decreased over time.

Through the work of many researchers, many health care institutions have created specialty centers for diagnosing and treating patients with FMD. There is now a strong community of patient support, partially due to the efforts of organizations like The Fibromuscular Society of America (fmdsa.org) and the U.S. Registry for FMD. This case illustrates that progress has been made in increasing the awareness of FMD and highlights that a lot of work still needs to be done.

ABBREVIATIONS

FMD: Fibromuscular Dysplasia; HTN: Hypertension; BP: Blood Pressure; ED: Emergency Department; CT: Computerized Tomography Scan; TIA: Transient Ischemic Attack; MI: Myocardial Infarction; BMI: Body Mass Index; NIHSS: National Institute of Health Stroke Scale.

INTRODUCTION

The vascular disorder, fibromuscular dysplasia (FMD), recently thought to be uncommon, is an arteriopathy that can affect any vascular bed, but most often manifests in the renal, carotid, and vertebral arteries [1,2]. Secondary sequelae of FMD such as hypertension (HTN), pulsatile tinnitus, transient ischemic attacks (TIAs) and stroke are often initial events that facilitate a diagnosis. In contrast, other patients are diagnosed incidentally during unrelated imaging studies, frequently among renal donors [1,2]. While the true prevalence of this condition remains largely unknown, it is estimated to be between 3% and 4% [2,3], affecting middle-aged women more than men, 9:1 [2]. Mean age at diagnosis is 52 years. Delays between symptom presentation and diagnosis, especially prevalent when the presentation is less severe, range from 4 to 9 years [2]. During these diagnostic delays, symptoms are often misinterpreted as primary conditions (such as HTN, headaches, stomach pain) or malingering [4]. The experience of being diagnosed and living with FMD poses another layer of complexity to managing patients afflicted by this condition. Herein we report a case of a delayed diagnosis of FMD in a patient with acute hypertensive encephalopathy, who was initially referred to psychology instead of receiving proper FMD treatment.

CASE PRESENTATION

Joan, a 43-year-old female, presented to a free clinic requesting a refill on her blood pressure medication, lisinopril. She explained that she has a rare condition "that you may not be familiar with," fibromuscular dysplasia. She was aware that most of her healthcare providers are unfamiliar with this condition. She used to have health insurance, but she subsequently lost it following her diagnosis. She is now out of her blood pressure medication and is afraid of having a stroke.

At this visit, Joan was 5'8", weighed 135 pounds, with a BMI of 20.6. She reported being a lifetime non-smoker, that she

walks her dog for 30 minutes daily, and does yoga. She has a past medical history of alcohol abuse in her 20's, but no other chronic illnesses. She currently is involved in a long distance relationship, recently moved and started a new job, and her mother passed away during the prior year.

The year prior, Joan recalls that she had been ill with stomach cramps and diarrhea. She was seen and treated in a clinic by a provider who noted her blood pressure (BP) to be 152/83. She was told at that time to "watch it." One month later, she experienced similar symptoms. She missed work for two days. Her coworkers became increasingly concerned when she quit answering the phone and went to check on her. Joan remembers recognizing her friends when they arrived, but was unable to say who they were. She was confused, lightheaded, and felt like she was fighting to stay conscious. Her friends drove her to a local emergency department (ED).

In the ED, Joan's vital signs were: temperature 97.8 F, heart rate 80 bpm, respirations 16, BP 198/105, Pulse Ox 100%. She did not know the year, day, or when she had last eaten or had water. She denied any fever or chills, chest pain, shortness of breath, urinary symptoms, or being depressed. However, after her friends left the ED, she tearfully reported that she had been depressed because of her mother's death and the status of her current relationship. In the ED, she received negative results on an EKG, computed tomography of the head, chest x-ray, urinalysis, and 6-panel urine drug screen. She additionally tested negative for serum troponin, Acetaminophen, Ethanol, and salicylates. Her electrolytes, not surprisingly, were abnormal; likely related to her prolonged confused state (Sodium 132mMol/L Low; Chloride 93 mMpl/L Low; Glucose 111 mg/dL High; BUN 5.5 mg/dL Low; Bilirubin 1.8 mg/dL High; Lipase 53 Units/L Low; Magnesium 1.6 mg/dL Low). She also had an elevated white blood cell count of 13.9 thou/mcl and a hypochromic (MCHC 30.4 gm/d), microcytic (MCV 72 FL), anemia (Hgb 10.9 gm/dL). Her National Institute of Health Stroke Scale (NIHSS) score was 1. She was treated with intravenous labetolol and as her BP returned to normal she became less confused, less upset, and described feeling more like herself.

Joan was kept overnight and the treating physicians plan included: 1) a referral for depression, 2) correction of the hypomagnesemia, 3) monitoring of the leukocytosis, 4) and iron studies for the chronic microcytic anemia. Her official diagnoses were: altered mental state secondary to depression and alcohol abuse, alcohol dependency, starvation ketoacidosis, and chronic microcytic anemia. Upon discussing her diagnoses with the physician, she became upset and again denied drinking. She chose to leave against medical advice after speaking with a social worker, but before meeting with the psychologist.

Certainly, the role of the ED is to manage life-threatening acute issues and to facilitate connections with appropriate outpatient resources for long-term management. In this case, the assessment and management of the electrolyte and hematologic concerns were appropriate. Further, the ED relieved the immediate danger by reducing her BP, and given her negative non-contrast head CT and low NIHSS score, a cerebral vascular event was discounted. What is concerning was the lack of acknowledgement of the BP concern in the plan or discharge diagnoses. With no further diagnostic evaluation for the etiology of the elevated BP, it became incumbent upon the patient to pursue the issue. Not all patients will be as motivated to seek answers as Joan was. By not including HTN as a diagnosis, primary care providers might not have investigated it. This is one manner by which delays to FMD diagnosis may occur. Additionally, the general lack of knowledge about FMD and lack of primary care providers with FMD experience often result in missed opportunities to screen for FMD. When this happens, patients may become misdiagnosed in the interim, or only receive a diagnosis when a chance finding is made by a radiologist on a study for a different reason.

Despite being upset and feeling that the ED providers may have missed something, Joan followed the providers' advice to see a primary care doctor. Joan's primary care doctor ordered a renal duplex which revealed right renal artery FMD with velocities consistent with stenosis of greater than 60%. She was started on lisinopril and referred to a neurologist and a nephrologist. Their retrospective diagnoses were that her prior episode of altered mental state and confusion were secondary to a hypertensive encephalopathy.

FOLLOW UP

Now a year later, Joan remains stable without any further events. She takes her medication faithfully, continues to exercise regularly and watches her diet. She admits that it has not been easy, especially without health insurance. She spent several months fearing she might have a stroke. More recently, she has come to terms with her diagnosis and is living her life as usual. She knows that when she sees new healthcare providers she often has to educate them about her condition, and she is tolerant of her well-meaning friends and family who do not understand her diagnosis.

Joan was happy to have a provider who understood her condition, and she was provided a new prescription for lisinopril. She was referred back to nephrology for annual follow-up with a request to discuss a one-time head-to-pelvis CT imaging study to screen for aneurysms/dissections. Because FMD is a polyvascular disease, other arteries may also be affected. Though this screening is not a guideline, it has become the practice of several large FMD centers as a way to guide ongoing surveillance [10,11]. It is worth noting that presently, revascularization is controversial and highly dependent upon the the individuals presentation [2]. Additionally, Joan was unaware of the Fibromuscular Dysplasia Society of America (FMDSA). She was provided the web address for the FMDSA (www.fmdsa.org) and educated on the resources available to her from this organization. Among the resources available on the website are links to support groups and a Patient Toolbox. This toolbox contains downloadable materials such as an Emergency card, Medication card, a sample Letter to Practitioners, an arterial map, a FMD dictionary, and a Letter for Family and Friends of FMD patients. These resources were printed and given to the patient before she left. With these resources, Joan realized that many people were working to inform providers and patients about this uncommon disorder and that she was not alone.

DISCUSSION

Joan's story is like that of many patients diagnosed with FMD.

In fact, a recent qualitative study of the life experiences of 19 individuals diagnosed with FMD revealed five discrete themes that participants shared; symptom burden, worries and concerns, experience of health care, loss and change, and resilience [4]. Patients described physical symptoms such as fatigue, pain, pulsatile tinnitus and psychological symptoms of anxiety and depression. In fact, most patients reported frequent worries over the uncertainty that FMD brought into their lives. They expressed fear of disability more than fear of death, but also fear about how to continue to care for their families or if they would become a burden [4]. Another common theme described by participants in this study was their experiences with the health care system. Both from a structural and an emotional perspective, participants identified lack of, and lack of access to, knowledgeable providers as one of their biggest concerns. They also reported that their symptoms were often dismissed, leading to diagnostic delays and being labelled as a malingerer [4]. Yet, despite overwhelming reports from patients of how being diagnosed with FMD caused hardships (from feeling isolated to having to leave their careers), participants also seemed to develop a sense of resiliency. Participants noted that, over-time, they were able to get on with their lives and become strong [4].

In a quantitative follow-up study, Heidt and Bumpus [5] surveyed 72patients diagnosed with FMD using standardized depression (PHQ9), anxiety (GAD7) and somatoform disorder (PHQ15) tools [6,7]. Their objective was to ascertain what proportion of FMD patients experience depressive, physical, or anxiety type symptoms; if symptoms varied by age, gender, or race; and if scale scores differed by how long patients had lived with the diagnosis, the duration of diagnostic delays, or experiences of major vascular events (stroke, TIA, aneurysm, dissection) (Table 1).

Preliminary data indicates that rates of depression (21.7%) and anxiety (10.4%) were more frequent in the FMD population when compared to the general population (6.7% and 3.1% respectively) [8,9]. Neither the number of vascular beds affected nor the location(s) of the affected bed(s) had any influence on psychological symptoms. However, symptoms seemed to be exacerbated by diagnostic delays. Patients who had a delay to FMD diagnosis of 1 to 5 years had a higher incidence of depressive and somatoform symptoms (Figure 1). Further, patients diagnosed with FMD of the head or neck and patients who had experienced an FMD-related event (stroke, TIA, MI, aneurysm, dissection, subarachnoid hemorrhage, renal infarction, mesenteric ischemia) had higher rates of somatoform symptoms [5]. Yet, these preliminary findings do suggest that

Table 1: The Effects of FMD Location, Number of Vascular Beds Affected, and Major Adverse Events on Mental Health.

	FMD Location			Number of Vascular Beds Affected by FMD			History of Major FMD events[e]		
	Extracranial carotid, vertebral, or intracranial FMD (n=37)	Other vasculature affected by FMD (n=35)	p-value	Single vascular bed affected (n=45)	Two or more vascular beds affected (n=27)	p-value	History (n=38)	No history (n=34)	p-value
Screened positive for Depressive Syndrome[a]	6/36[d] (16.7%)	9/33 (27.3%)	0.38	10/44 (22.7%)	5/25 (20.0%)	1.0	8/36 (22.2%)	7/33 (21.2%)	1.0
Screened positive for Physical Symptoms[b]	15/28 (53.6%)	7/26 (26.9%)	0.057	14/34 (41.2%)	8/20 (40.0%)	1.0	16/28 (57.1%)	6/26 (23.1%)	0.014
Screened positive for Generalized Anxiety Disorder[c]	3/35 (8.6%)	4/32 (12.5%)	0.70	3/42 (7.1%)	4/25 (16.0%)	0.41	4/35 (11.4%)	3/32 (9.4%)	1.0

Abbreviations:FMD: Fibromuscular dysplasia
[a]As assessed by the PHQ9; [b]As assessed by the PHQ15; [c]As assessed by the GAD7; [d]The denominator reflects the number of patients who completed each survey; [e]Stroke, transient ischemic attack, myocardial infarction, coronary revascularization, aneurysm, dissection, subarachnoid hemorrhage, renal infarction, or mesenteric ischemia

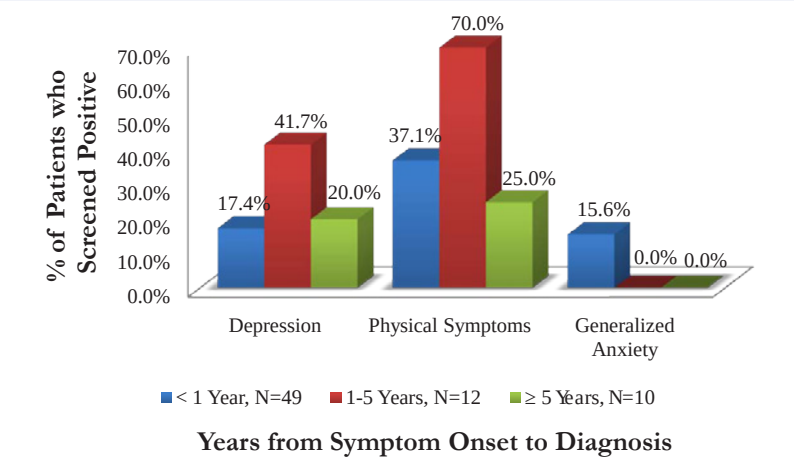

Figure 1 Rates of depressive syndrome, physical symptoms, and generalized anxiety by delay to FMD diagnosis.

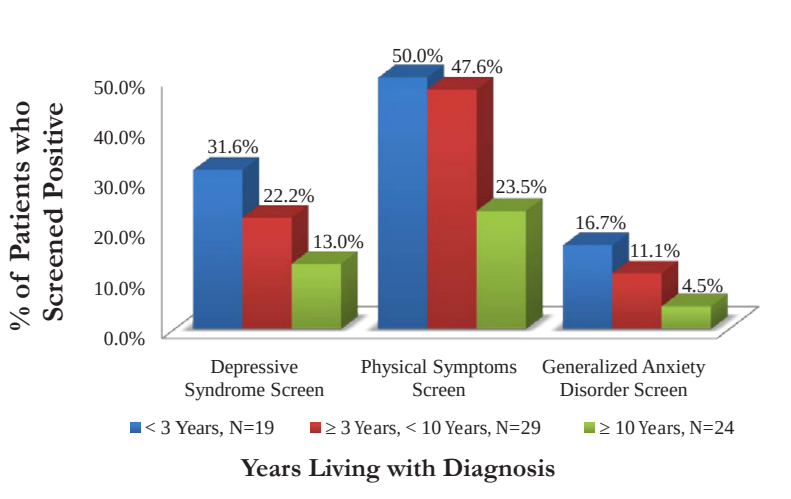

Figure 2 Rates of depressive syndrome, physical symptoms, and generalized anxiety by length of time post-FMD diagnosis.

symptoms of depression and anxiety, and somatoform symptoms are greatest in the first three years after diagnosis and appear to lessen over time (Figure 2).

Overall, the results of the FMD survey, in a small sample of FMD patients [5], supported the earlier qualitative findings of Bumpus, Kuck, Heidt, and Bluhm [4]. Moderate depressive symptoms, physical symptoms, and generalized anxiety disorders are not uncommon in the FMD population. However, perhaps the most interesting finding was the trend that depression, physical symptoms, and anxiety may decrease over time.

CONCLUSION

Through the work of many researchers, a number of health care institutions have created specialty centers for diagnosing and treating patients with FMD. There is now a strong community of patient support, partially due to the efforts of organizations like The Fibromuscular Society of America and the U.S. Registry for FMD. This case illustrates that progress has been made in increasing the awareness of FMD and highlights that a lot of work still needs to be done. In particular, not only is research aimed at understanding and managing the disease necessary, research related to provider awareness and access is paramount; as is translational research designed to enhance quality of life.

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