

Letter to the Editor

DOI: 10.22114/ajem.v0i0.316

A Letter on “The Current Status of Genes and Genetic Testing in Emergency Medicine: A Narrative Review”

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Published online: 2019-12-10

On August 25, 2019, you published an article titled “The Current Status of Genes and Genetic Testing in Emergency Medicine: A Narrative Review” (1). I recently read the article and want to both commend the authors for including information on Vascular Ehlers-Danlos Syndrome (vEDS) and expand upon the information provided. Thank you for publishing this article.

vEDS is an uncommon connective tissue condition resulting from a mutation in the COL3A1 gene, which directs the production of collagen III in the body. As a result of mutations in the COL3A1 gene, patients with vEDS present with a high risk of arterial ruptures, dissections, aneurysms, and carotid-cavernous sinus fistulas, as well as rupture of hollow organs (such as the bowels) and lung collapses. These complications can be spontaneous in nature, and oftentimes present with no trauma. In the third trimester of pregnancy, females with vEDS are also prone to uterine rupture without prior C-section. Along with the high risk of these complications, the identification of and intervention for these emergencies can be different from a patient without a connective tissue disorder. Invasive imaging techniques like catheter arteriography, colonoscopy, and endoscopy are potentially life threatening to individuals with vEDS due to the fragility of the tissue. Therefore, it is imperative both the treating physician and triage nurse be aware of the condition upon arrival to the emergency department (ED).

Individuals with vEDS are advised upon diagnosis to carry an emergency information card or letter from their physician with them containing information about vEDS in case of a need to visit the ED. The patient who presents with sudden onset of severe pain may be experiencing a life-threatening event. How this information is handled once in the ED could mean life or death for a patient with vEDS experiencing an emergency.

Here are some first-hand experiences of patients with vEDS on their experiences in the ED:

“With my carotid dissection, the ED docs didn’t know how to handle it so I was transferred to another hospital where the doctor on call was from France. This was before my diagnosis with vEDS, but the physician recognized vEDS immediately given my current state, medical history, and my mother’s medical history. She was the first to consider vEDS and because of her awareness of it, she took extra precautions during my embolization procedure to handle my arteries with TLC. It took two procedures to complete, but I am 100% convinced that the reason I survived is she had prior awareness of vEDS complications.”

“I have a family member who didn’t have the vEDS diagnosis. They were released by the hospital and then had a fatal event.”

“In my twenties, I was having lots of pain around my gallbladder, front & back area, so my GI said to go to the ED since it was Friday afternoon. I always take my medication list, surgery list, etc. Fortunately, the hospitals have my info too, but it is not always updated. When I got there, I explained I have vEDS and multiple aneurysms already. They got me back as soon as possible and ran me through the CT machine. Didn’t turn out to be my gallbladder at all, but a new aneurysm in my abdominal iliac artery.”

“When I was not yet diagnosed with vEDS, I had an arterial dissection in my heart and post-partum heart attack at 25. Multiple EDs told me that it was a very bad flu. I was left home vomiting, not being able to chew, could not go to the bathroom, tongue turned green, left like this for 10 days until my family doctor noticed severe elevation of my red blood cells...I was finally diagnosed with vEDS years later. Upon my diagnosis, I had a file with all of my history. When I delivered the second time, I had the same symptoms as the heart attack, but this time we were prepared. In the middle of the night we drove to the best hospital downtown, I had my binder, they took me in immediately and did the appropriate scans ASAP. It turned out to be two

blood clots (one to each lung) and they discovered a splenic aneurysm.”

I encourage emergency medicine physicians and nurses to familiarize themselves with vEDS, and other connective tissue disorders like Loeys-Dietz Syndrome and Marfan Syndrome, that can result in life-threatening aortic and arterial dissections and ruptures. Knowing the signs of these symptoms and having knowledge on how to treat a patient presenting with an emergency related to these conditions may save the patient’s life.

You can find more information on vEDS at TheVEDSMovement.org.

Thank you so much for publishing this article. I look

forward to seeing more articles like it in the future.

ACKNOWLEDGEMENT

None.

AUTHOR CONTRIBUTION

The author passed four criteria for authorship contribution based on recommendations of the International Committee of Medical Journal Editors.

CONFLICT OF INTEREST

None declared.

FUNDING

None declared.

REFERENCES

1. Aghamir SMK, Ebrahimi M, Khatami F. The Current Status of Genes and Genetic Testing in Emergency Medicine: A Narrative Review. *Adv J Emerg Med.* 2020;4(1):e10.