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Early Recognition of Signs of Hypercoagulability to Prevent Adverse Health Outcomes

Omar Elsemary

College of Medicine, California North state University College of Medicine, Elk Grove, USA.

Abstract

Factor V Leiden is a genetic disorder that results in the elimination of the cleavage site on the clotting factor V and Va. This increases the risk for clotting events, such as a stroke, in affected individuals. Factor V Leiden is often overlooked due to a lack of screening measures. Our case report outlines a patient who suffered from a stroke with no abnormalities on diagnostic testing until further genetic testing revealed Factor V Leiden.

Keywords: hypercoagulability; stroke; pregnancy

Introduction

Strokes are extremely prevalent in the United States with the CDC estimating that about 800,000 people experience one every year [1]. The majority of these strokes are ischemic strokes and often leave individuals chronically disabled. Strokes commonly occur in the elderly and their incidence doubles with each decade of life after the age of 45 [2]. However, strokes do still affect younger adults, particularly those with genetic predispositions to thrombotic events. One such group that is at risk due to a genetic predisposition are those with Factor V Leiden.

Factor V Leiden is a mutation in which the amino acid arginine at the 506th position is substituted for glutamine [3]. This causes factor V to be resistant to degradation by protein C, ultimately leading to hypercoagulability. The increased risk of thrombosis in individuals with Factor V Leiden is also associated with increased risk of stroke with a much earlier incidence compared to the normal population. Although this presentation is uncommon, it is extremely dangerous and highlights the importance of genetic testing and preventive care, particularly in those with early warning signs. As such, this case report focuses on a young, obese but otherwise healthy female who experienced an ischemic stroke with the potential underlying cause of Factor V Leiden.

Case Presentation

A 39-year-old female with a BMI of 36 presented with right-sided upper and lower limb weakness along with slurring of speech. The patient was previously in good health with no major diagnoses. However, she does have a history of two spontaneous losses of pregnancy at ages 34 and 36. She states that her symptoms began at about 11:00 am and she presented roughly three hours later. She has not had a similar experience in the past and there is no family history of similar episodes. A CT scan was done upon admission, however there were no visible abnormalities. Complete blood count was also normal. A blood test revealed that she was positive for the Factor V Leiden mutation. The administration of tissue plasminogen activator (tPa) led to an almost complete reversal of the patient's symptoms.

Discussion

Strokes are the second leading cause of death in the world with a variety of risk factors which are important to understand [4]. Two particularly important risk factors for ischemic strokes are the presence of hyperlipidaemia and atrial fibrillation [5]. While many of these risk factors are relatively common and have been studied extensively, the presence of Factor V Leiden is a major risk factor that may often be overlooked.

Although strokes generally occur in elderly individuals with already existing risk factors, patients with Factor V Leiden are at a significantly higher risk of experiencing

strokes earlier in life. This is extremely important to consider, for individuals with Factor V Leiden are often unaware they have this mutation and may otherwise be healthy although they are at risk for a life changing thrombotic event. When associated with other risk factors, a Factor V Leiden mutation can significantly increase the probability of stroke. In the case of our patient, possessing the Factor V Leiden mutation alongside the risk factor of obesity put her at increased risk of stroke.

The increased risk of stroke due to Factor V Leiden calls for genetic testing to be done at an early age. It is estimated that about 5% of Caucasians and 1% of African Americans carry the Factor V Leiden mutation [6]. This is a significant portion of the population, and genetic testing will allow for early intervention and lifestyle modifications. Furthermore, carriers of this mutation may be completely healthy with no significant medical record as is the case with our patient. However, there may be some warning signs that patients may be carriers of Factor V Leiden such as recurrent pregnancy loss. Recurrent loss of pregnancy should raise red flags for thrombophilia and call for genetic testing [7]. Factor V Leiden is associated with a three-fold increase in risk for pregnancy loss, making it extremely important to consider in patients who have repeatedly lost pregnancies [8]. This exemplifies the importance of genetic testing, for it could be difficult to catch such mutations until it may be too late.

Conclusion

Our case focuses on a 39-year-old Caucasian female that was diagnosed with Factor V Leiden mutation upon presentation of stroke symptoms. Strokes become more common among individuals with each decade of life, however, a young patient with Factor V Leiden mutation could be prevented if diagnosed early. This calls attention to genetic/hypermobility testing on infants to ensure early prevention of risks associated with Factor V Leiden mutation and other hypercoagulability disorders.

References

1. Stroke facts Centers for Disease Control and Prevention, 2021.
2. Ovbiagele B, Nguyen-Huynh MN. (2011). Stroke epidemiology: advancing our understanding of disease mechanism and therapy. *Neurotherapeutics*, 8:319-329.
3. Albagoush SA, Koya S, Chakraborty RK, et al. (2021). Factor V Leiden Mutation. StatPearls Publishing, Treasure Island (FL).
4. Donkor ES. (2018). Stroke in the 21st century: A snapshot of the burden, epidemiology, and quality of life. *Stroke Res Treat*.
5. Boehme AK, Esenwa C, Elkind MS. (2017). Stroke Risk Factors, Genetics, and Prevention. *Circ Res.* 120:472-495.
6. De Stefano V, Chiusolo P, Paciaroni K, Leone G. (1998). Epidemiology of factor V Leiden: clinical implications. *Semin Thromb Hemost*, 24:367-379.
7. Ebrahimzadeh-Vesal R, Azam R, Ghazarian A, et al. (2014). Recurrent pregnancy loss in a subject with heterozygote factor V Leiden mutation; a case report. *Rep Biochem Mol Biol*, 2:98-102.
8. Martinelli I, Taioli E, Cetin I, et al. (2000). Mutations in coagulation factors in women with unexplained late fetal loss. *N Engl J Med*, 5:1015-1018.

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