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Exploring Factors That May Contribute to the Underdiagnosis of Hereditary Transthyretin Amyloidosis in African American Patients

Authors : Kelsi Hagerty, Ami Rosen, Aaliyah Heyward, Nadia Ali, Emily Brown, Erin Demo, Yue Guan, Modele Ogunniyi, Brianna McDaniels, Alanna Morris, Kunal Bhatt

Abstract : Hereditary transthyretin amyloidosis (hATTR) is a progressive, multi-systemic, and life-threatening disease caused by a disruption in the TTR protein that delivers thyroxine and retinol to the liver. This disruption causes the protein to misfold into amyloid fibrils, leading to the accumulation of the amyloid fibrils in the heart, nerves, and GI tract. Over 130 variants in the TTR gene are known to cause hATTR. The Val122Ile variant is the most common in the United States and is seen almost exclusively in people of African descent. TTR variants are inherited in an autosomal dominant fashion and have incomplete penetrance and variable expressivity. Individuals with hATTR may exhibit symptoms from as early as 30 years to as late as 80 years of age. hATTR is characterized by a wide range of clinical symptoms such as cardiomyopathy, neuropathy, carpal tunnel syndrome, and GI complications. Without treatment, hATTR leads to progressive disease and can ultimately lead to heart failure. hATTR disproportionately affects individuals of African descent; the estimated prevalence of hATTR among Black individuals in the US is 3.4%. Unfortunately, hATTR is often underdiagnosed and misdiagnosed because many symptoms of the disease overlap with other cardiac conditions. Due to the progressive nature of the disease, multi-systemic manifestations that can lead to a shortened lifespan, and the availability of free genetic testing and promising FDA-approved therapies that enhance treatability, early identification of individuals with a pathogenic hATTR variant is important, as this can significantly impact medical management for patients and their relatives. Furthermore, recent literature suggests that TTR genetic testing should be performed in all patients with suspicion of TTR-related cardiomyopathy, regardless of age, and that follow-up with genetic counseling services is recommended. Relatives of patients with hATTR benefit from genetic testing because testing can identify carriers early and allow relatives to receive regular screening and management. Despite the striking prevalence of hATTR among Black individuals, hATTR remains underdiagnosed in this patient population, and germline genetic testing for hATTR in Black individuals seems to be underrepresented, though the reasons for this have not yet been brought to light. Historically, Black patients experience a number of barriers to seeking healthcare that has been hypothesized to perpetuate the underdiagnosis of hATTR, such as lack of access and mistrust of healthcare professionals. Prior research has described a myriad of factors that shape an individual's decision about whether to pursue presymptomatic genetic testing for a familial pathogenic variant, such as family closeness and communication, family dynamics, and a desire to inform other family members about potential health risks. This study explores these factors through 10 in-depth interviews with patients with hATTR about what factors may be contributing to the underdiagnosis of hATTR in the Black population. Participants were selected from the Emory University Amyloidosis clinic based on having a molecular diagnosis of hATTR. Interviews were recorded and transcribed verbatim, then coded using MAXQDA software. Thematic analysis was completed to draw commonalities between participants. Upon preliminary analysis, several themes have emerged. Barriers identified include i) Misdiagnosis and a prolonged diagnostic odyssey, ii) Family communication and dynamics surrounding health issues, iii) Perceptions of healthcare and one's own health risks, and iv) The need for more intimate provider-patient relationships and communication. Overall, this study gleaned valuable insight from members of the Black community about possible factors contributing to the underdiagnosis of hATTR, as well as potential solutions to go about resolving this issue.

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