

Abstract 32

FRAGILE X SYNDROME DIAGNOSIS EXPERIENCES AMONG FAMILIES OF AFRICAN AMERICAN ETHNICITY

Krista Charen, Julia K. Rohr, Emily G. Allen, Stephanie L. Sherman
Department of Human Genetics, Emory University, Atlanta, USA

A timely diagnosis of fragile X syndrome (FXS) is important so that those with FXS can begin treatments and therapies. Other family members can also be educated and undergo genetic testing to determine their risk for any of the fragile X-associated disorders. Previous studies have evaluated FXS diagnosis experiences, predominately among those as self-reported Caucasian. Understanding the diagnosis experiences of other ethnic/racial groups is important to identify possible differential barriers. Semi-structured, qualitative interviews were analyzed for common themes. Participants consisted of women self-reported as African American who have at least one child diagnosed with FXS. A comparison group consisting of Caucasian women matched on age, income and education also completed the interviews. Although data collection is still in progress, we will present preliminary findings. Participants reported missing developmental milestones as the first sign that something might be wrong with their child. Many reported first receiving a diagnosis of a condition other than FXS, such as ADHD. The most common barrier to receiving a FXS diagnosis was lack of knowledge by the pediatrician. Half of the African American participants had to bring information about FXS to their doctor and ask for the test. Half of the African American participants also mentioned that their child did not physically resemble the typical description of FXS. The comparison group did not mention this experience. Participants reported the internet as their main source of information about FXS. Other avenues of information included physicians, parent support groups, and research groups. All participants demonstrated a proactive approach when dealing with the healthcare industry. Education programs targeting pediatricians about FXS are needed, as the time from first symptoms to receiving a diagnosis of FXS continues to be too long for families of both ethnic origins. For many, the internet seems to be an effective resource in educating families.