Family History, a Crucial Element in Diagnosis of Fragile X Syndrome: A Case Report

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INTRODUCTION: Fragile X Syndrome (FXS) is an X-linked genetic disorder associated with a range of physical, cognitive, and behavioral features. The full mutation form typically involves cognitive delays, behavioral changes and characteristic physical markers.

PURPOSE: The purpose of this study was to give an overview of FXS, discuss hallmark signs and symptoms, and consider how a comprehensive family history can lead to earlier diagnosis.

CASE PRESENTATION: This case report discusses the diagnosis of a 2-year-old male patient with full mutation FXS and how a detailed family history provided important information for family members experiencing similar symptoms. The patient was initially evaluated for delayed developmental milestones, right eye deviation, and abnormal hand movements. Further questioning revealed relevant family history including one maternal male first cousin who was later diagnosed with full mutation FXS, and another being evaluated for delayed talking milestones.

DISCUSSION: Although FXS is one of the most common inherited causes of intellectual disability and simple genetic testing is available, on average there is an 18-month delay between family first expressing concerns regarding symptoms and a diagnosis being made. This delay is significant considering early interventions result in better outcomes. Genetic counseling is also important and may have an impact on reproductive planning in families with known family history.

CONCLUSION: Early intervention is advantageous in FXS. To achieve early diagnosis, providers must be familiar with symptoms of FXS and perform a thorough and relevant family history.