A Unique Case of Subclinical Becker Muscular Dystrophy Due to a Single Exon 48 Deletion in the Dystrophin Gene

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Introduction

Duchenne and Becker Muscular Dystrophy are the two most common forms of muscular dystrophy. BMD, which is a type of dystrophinopathy, is generally considered to be less severe. Men with a marked exon 48 deletion in the dystrophin gene may present with a mild or subclinical type of BMD. Typically, neurological assessments remain normal and show good exercise tolerance.(1-3) In this case, we present a boy who was initially diagnosed with Bethlem Myopathy, but further testing revealed a single exon 48 deletion in the dystrophin gene causing BMD.

Case

A 13-year-old boy, born to consanguineous parents, experienced his first febrile seizure at the age of five. He had subsequent febrile seizures and elevated creatine kinase levels. At the age of eight, genetic testing was performed which showed heterozygosity for the COL6A3 and COL12A1 genes. This led to a preliminary diagnosis of Bethlem myopathy caused by compound heterozygous mutation. However, the patient's reported symptoms of difficulty in climbing stairs, occasional falls, **hypertrophy of the calves, chest pain** while running, and involuntary twitching of the face required a more comprehensive examination. The continued **elevation of CK levels** and the **absence of typical myopathy** findings **on EMG** confidently warranted re-evaluation. Upon molecular karyotype analysis, a pathogenic deletion in the P21 region -arr[GRCh38]Xp21.1 DMD- with a size of 77.2 Mb (14 markers) above the resolution threshold was discovered. The individual has a mild form of Becker Muscular Dystrophy with the potential for severe cardiomyopathy due to an exon 48 deletion. Despite the echocardiographic evaluation not showing any signs of cardiomyopathy, the individual experienced **dysrhythmia**. Yearly cardiac follow-up was performed and more frequent follow-up was planned.

Conclusions

Preventive cascade screening of relatives and newborn screening for DMD/BMD is crucial to obtain a definitive diagnosis by genetic testing, given the high sCPK levels and clinical details. Although an article has been reported that isolated deletion of exon 48 presents a mild phenotype and cardiomyopathy is not common, follow-up for cardiomyopathy is still important.(1,2) Conducting therapeutic trials for DMD/BMD is essential to avoid cardiac complications and prolong survival.

References

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