Fragile x syndrome - symptoms and investigation

Title

Complications and investigations of Fragile X syndrome

Introduction

Fragile X syndrome is considered the most common heritable form of mental retardation, it affects approximately 1 in 4000 males and 1 in 8000 female it is an X linked disease caused by mutation in the” FMR1 gene “ This mutation in dynamic it varies in length and so in severity from generation to generation The syndrome is more severely affecting boys It is associated with by prominent or long ears This abnormal morphology suggests abnormal auditory function.. The mutational mechanism, combined with the location of this gene on the X chromosome, leads to remarkable inheritance patterns in which the relevant alleles are passed from intellectually normal men through their unaffected daughters and then to affected sons

Aim of Work

Study the complications and achieve accurate investigation of fragile X syndrome

Methods

Meta-analysis randomized controlled trials studies were revised of Fragile x analysis patients either complicated with hemiplegia or not , and Systematic review and several case control studies were revised about hearing assessment using auditory evoked potential or auditory stimuli on children with fragile X syndrome were identified based on systematic searches of 5 electronic databases including Pubmed database , in addition to Searching on internet on site of “ national fragile x foundation” and site ” European

Journal of Human Genetics", which provide me with many information that helped me in proving the role of chromosome Karyotyping and comparative genomic hybridization (CGH) microarray in investigation of Fragile X syndrome

Results

According to Above Methods , Sample of Children with Fragile X syndrome showed that Seizures are reported to occur in 10 to 20 percent of cases with a full mutation FXS 1 and are typically of the complex partial type 4.

About 25 – 50 % suggested a high incidence of autistic behavior , most of them were in the mild to moderate autistic range while few suggested to have severe autism . Also, another screening studies were done and revealed that the sensitivity to develop autism in children with FXS is very high.

in one cross sectional study, Participants (aged 10-23 years) included 17 males and females with FXS and 16 males and females serving as controls. Patients with FXS showed decreased functional connectivity in the salience, precuneus, left executive control, language, and visuospatial networks compared with controls. Decreased fALFF in the bilateral insular, precuneus, and anterior cingulate cortices also was found in patients with FXS compared with control participants, it is evident that FXS is a neurogenetic disorder characterized by a cognitive and behavioral phenotype resembling features of autism spectrum disorder. But no cases develop a more serious neurologic disorder such as hemiplegia as one of its complications

In control studies which is done on sample of children with Fragile X syndrome after chromosome Karyotyping We found a break, or weakness, on the long arm of the X chromosome. it was helpful, it was not always accurate as, those with premutations did not have the typical “ Fragile X” chromosome as seen under the microscope.

Also, using comparative genomic hybridization (CGH) micro array is very useful as Comparative genomic hybridization (CGH) micro array is a genetic test to detect any small deletions and duplications of the subtelomers of chromosomes

Several case control studies was performed using auditory evoked potential One of them was made on 10 girls with fragile X syndrome and another age matched typically developing controls aimed at performing an auditory temporal discrimination task in a 3T scanner proved that there is significantly great brain activation in the left lateralized network

On the other hand a case control study was mad on 23 with FXS and 21 controls by using auditory evoked potential and it proved that AEP of fragile X syndrome patients is within the normal limits

Conclusion

From above studies methods and their results, it was found that there is many complications of fragile X syndrome as:

Seizure, which is reported to occur in 10 to 20 percent of cases with a full mutation FXS high percentage of Fragile X syndrome patients develop autism .

There are also evidence of developing other developmental disorders

But, no case has been reported to develop hemiplegia so, the fragile x patient is not prone to be hemiplegic.

Above studies also showed the accurate investigations for FXS
as chromosome Karyotyping help in diagnosis of Fragile X syndrome I recommend it in this case to show the affected gene but , it was not always accurate

hearing assessment is recommended to diagnose fragile X syndrome as most of these children have hearing difficulties Although the absence of these difficulties does not rule out the disease

(CGH) micro array is recommend in to investigate case with fragile X chromosome. As it can detect specific abnormalities in a chromosome.

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Keywords :-

FragileXsyndrome – autism- epilepsy- CGH-chromosome karyotyping - auditory evoked potential -hemiplegia