GENETICS

FRAGILE X SYNDROME: WHAT A PEDIATRICIAN HAS TO KNOW?

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Abstract: *Fragile X syndrome is the most common cause of inherited intellectual disability caused due to a mutation in fragile X mental retardation 1 gene on the X chromosome. The clinical features of fragile X syndrome in affected males include severe intellectual disability, peculiar facial features, joint hypermobility, macroorchidism, seizures and neuropsychiatric abnormalities. Females with fragile X syndrome have milder intellectual disability and cognitive abnormalities because of one normal copy of the gene on X chromosome. Molecular diagnosis of this condition is possible by southern blot or triplet primed polymerase chain reaction and is essential for providing genetic counseling and prenatal diagnosis. This article is a brief review on clinical features, molecular diagnosis and genetic counseling issues in fragile X syndrome in pediatric practice.*

Keywords: *Intellectual disability, Fragile X, Southern blot, Prenatal diagnosis.*

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Points to Remember

- ***Fragile X syndrome is a common cause of inherited intellectual disability.***
- ***Males are severely affected than females.***
- ***Molecular diagnosis is essential for genetic counseling and providing prenatal diagnosis.***

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