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Poster presentation

The behavioural neurogenetics of Fragile X syndrome: a model of gene-brain behaviour relationships

Marjaneh Akbarzadeh*[1], Reza Akbarzadeh[2] and Reyhaneh Akbarzadeh[1]

Address: [1]Department of Psychology, Russian University of Economics and Culture, Russian Federation and [2]Mashhad University of Medical Sciences, Mashhad, Russian Federation

* Corresponding author

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Background

Fragile X syndrome is a neurodevelopmental disorder that caused by a dynamic mutation on the X chromosome and displays a remarkable pattern of inheritance.

Fragile X syndrome a single-gene disorder that has become a well-characterized model for studying behavioural neurogenetics dysfunction in childhood.

Materials and methods

The purpose of this article is to summarize key advances made in understanding the variation in brain structure and function, and biological and environmental factors that influence developmental and cognitive outcomes of children with fragile X syndrome.

Results

The syndrome is the most important X-linked etiology of mental retardation and developmental disability currently known. The physical, cognitive and behavioural features of individuals with fragile X syndrome depend on gender (females have two X chromosomes, one active and one inactive) and the molecular status of the mutation (permutation, full mutation or mosaic). Features of the behavioural profile of individuals with fragile X syndrome include hypersensitivity to stimuli, overarousability, inattention, hyperactivity and (mostly in men) explosive and aggressive behaviour to others or self. Accumulating evidence also indicates that male and female carriers of the fragile X genetic abnormality demonstrate a relatively specific pattern of psychiatric disturbance. Other anxiety disorders such as social anxiety, depression, impulse control disorder and mood disorders are the most common psychiatric disorders diagnosed in individuals with fragile X syndrome.

Discussion

Analyzing gene-brain-behavior linkages in childhood neurodevelopmental disorders, a research approach called behavioural neurogenetics, has provided new insights into understanding how both genetic and environmental factors contribute to complex variations in typical and atypical human development. Our research and that of others demonstrates that neurobehavioural and neurocognition, genetics, and neuroanatomy are all different views of the same intriguing biological puzzle; a puzzle that today is rapidly emerging into a more complete picture of the intricate linkages among gene, brain, and behavior in developing children.