Opinion

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Myoclonus in Childhood-Onset Neurogenetic Disorders and Applied Behaviour Analysis

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Description

Many people who have developmental disabilities also have intellectual disabilities that are genetic. Because the underlying genetic etiology of these disorders predisposes them to a profile of atypical neurodevelopment, they are frequently referred to as "neurogenetic" disorders. A distinctive behavioral profile, or behavioral phenotype, emerges from this genetic etiology. A particular behavioral profile is uniquely associated with a particular neurogenetic disorder because the underlying genetic origin contributes to (or shapes) the behavioral phenotype. Probabilistic emergence of behavioral phenotypes is frequently influenced by interactions with the environment. Therefore, there is no direct correlation between a behavioral profile and a neurogenetic disorder. Instead, there is overlap between neurogenetic behavioral phenotypes and variability within syndromes, as well as a greater likelihood that an individual's profile will reflect one disorder over another. Environmental contingencies can be structured in therapeutic ways to address areas of relative difficulty in particular neurogenetic disorders, as evidenced by the complex, interactive nature of emerging behavioral phenotypes across the lifespan.

Social communication and interaction deficits and restricted, repetitive patterns of behavior, interests, or activities are hallmarks of autism spectrum disorder (ASD).Heller reported on six normally developing children who experienced a severe regression in skills between the ages of 3 and 4 that resulted in global impairments, including autistic features, decades before Kanner published his landmark paper describing autism. Heller gave the condition dementia infantilis the name childhood disintegrative disorder (CDD), which was included in the ICD-10 and DSM-IV. Normal development for at least the first two years of life and regression before age 10 in at least two of the following areas were used to define CDD: (1) language that is either expressive or receptive, 2) social skills or adaptive behavior, 3) control of the bowel or bladder, 4) play, and 5) motor skills.It has been hotly debated whether CDD is a separate condition or a late-onset variant of autism.In the DSM-5, CDD was included under the diagnosis ASD because there was little evidence to support its inclusion as a separate disorder.

Prevalence of CHD

However, there are significant phenotypic distinctions between CDD and other ASD types. The onset of CDD symptoms typically occurs between the ages of 3 and 4, whereas ASD symptoms typically begin at 2 years of age. CDD is characterized by regression, which is characteristically of later onset, more global in scope, and more severe in degree. In contrast, approximately a third of children with ASD experience a regression in skills, again typically by the age of two. Indeed, children with CDD typically have the worst outcomes compared to those with ASD, typically suffering from severe cognitive and communication impairments. In contrast to CDD, children with ASD who are diagnosed later than the typical age range typically have higher functioning children, which causes the delay in diagnosis. Early, subtle abnormalities are frequently noted in retrospect. A distinct prodrome is experienced by the majority of children with CDD, and it is characterized by episodes of anxiety and terror. CDD has not been consistently linked to any specific medical, environmental, or psychosocial factors.

The "heightened probability or likelihood that people with a given syndrome will exhibit certain behavioural and developmental sequeale relative to those without the syndrome" is the definition of the term "behavioural phenotype. "The behavioral characteristics of the affected children have typically been the focus of research on behavioral phenotypes. However, the concept of a phenotype must be extended beyond behavior because behavior is only one aspect of aetiology-related characteristics that affect these children and their families. Behavioural phenotypes should take into account not only the impact on others, like parents, but also non-behavioral aspects.

Types of Neurogenetic Disorders

It may be possible to consider the role of the neurogenetic conditions' indirect effects by considering their effects on affected children's families. A child may be predisposed to certain behaviors due to a specific genetic disorder; These behaviors, in turn, make the child more likely to have certain reactions or behaviors from other people, which may change the

child's behavior. Supporting the child and the family as a whole will likely require an understanding of this interaction.

The current investigation focuses on four of the most prevalent neurogenetic syndromes associated with learning and behavioral issues: fragile X syndrome (FXS).According to Greco et al., FXS is the most well-known cause of inherited intellectual disability.2006), and it has a strong connection to anxiety, Attention Deficit Hyperactivity Disorder (ADHD), and Autism Spectrum Disorder (ASD).According to Sherman (2002), the prevalence is estimated to be one in 4,000 males and one in 8,000 females. Hyperphagia, a variety of physical and behavioral characteristics, and cognitive impairment are all symptoms of PWS. Estimates of prevalence range from one in 10,000 to one in 45,000.According to Martens et al., WS is a condition that is linked to mild intellectual disability.2008), a number of different medical conditions, and a behavioral profile that puts them at a higher risk for problems paying attention, fears and phobias, and social withdrawal. It is estimated to affect 1 in 7,500 to 1 in 20,000 people.22qDEL is linked to a variety of congenital and late-onset diseases, such as major congenital heart disease, cognitive impairment, and an increased risk of attentional, anxiety, and schizophrenia difficulties.1 in 4000 people are thought to be affected.