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Commentary

Insights of Neurodevelopmental Disorders

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DESCRIPTION

Rett syndrome is a neurodevelopmental condition that mostly affects females, with mutations in the transcription regulation gene MECP2 being the most common cause. Normal carrier females, females with mild learning disabilities and Angel man syndrome-like features, and males with Klinefelter syndrome or Rett syndrome-like features, fatal neonatal encephalopathy, and familial X-linked mental retardation with or without motor abnormalities have all been found to have mutations in MECP2. As a result, molecular testing should be explored for a wide range of people. As a result, Rett syndrome is still considered a clinical diagnosis. Three recent developments are also discussed in this article: (1) the identification of significant gallbladder dysfunction, particularly in those under the age of 20; (2) a clinical trial of folate and betaine, which showed no objective improvement but did result in a subjective increase in attention and interaction; and (3) measurement of cerebrospinal fluid folate levels in a large cohort, which showed normal values, indicating no need for supplementation.

Despite the fact that Rett syndrome has been recognized in the Western Hemisphere for the past 20 years, Andreas Rett was the first to describe it in 1965. Bengt Hagberg was examining females with Rett syndrome clinical characteristics in Sweden at the same time. The pattern of a neurodegenerative sickness was initially described by Rett and others. However, as we've learned more about Rett syndrome, we've begun to see it as a paradigm for neurodevelopmental problems. The next sections offer the foundation for this viewpoint.

Despite the fact that Rett and Hagberg were both studying female Rett syndrome patients at the same time, they did not have any direct contact and did not discuss their findings for nearly 20 years.

Rett syndrome has a different temporal profile than other neurodegenerative diseases. Following a period of developmental stagnation and then regression, sociability increases substantially, with intense eye look and eye pointing resulting in a meaningful communication system between parents or other caregivers and people with Rett syndrome in many cases. Motor skills are generally preserved in late adolescence or early adulthood, despite a progressive slowdown or loss.

The morphology of the brain in Rett syndrome is quite distinct. The weight of the brain is consistently lower than it should be given its age. Reducement appears to be selective or more pronounced in frontal and temporal regions, as well as deep grey matter, according to volumetric magnetic resonance imaging. Melanin levels in pigmented nuclei like the substantial nigra are lower. Neurons are smaller than normal, with fewer dendritic barbarizations, when viewed under a microscope. The lack of any discernible illness pattern, that is, no indication of nerve cell loss or degeneration, as would be expected in a neurodegenerative ailment, is also notable. Other neurodevelopmental illnesses, interestingly, share a similar pathophysiology. Dendritic branches and spines are decreased in Down syndrome from infancy onwards.

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