Autonomic disturbance in Rett syndrome

Sarojini Budden MD, FRCP C, FAAP

Portland, Oregon

The function of MECP-2 gene contributing to regulation and activity of other genes is being established in Rett syndrome. It plays an important role in brain development and maintenance of cortical and subcortical connections resulting in known anatomical, physiological, biochemical, functional, and growth disturbances.

Hypo-function of the monoamines; noradrenaline, serotonin and nor-epinephrine is present as early as 36 weeks of gestation. Deficient brain-derived growth factor (BDNF) has been shown to occur and serotonin receptors are increased in the brainstem suggesting defective serotonin supply or activity. Throughout early childhood, levels of glutamate are raised and cortical receptors are increased, decreasing later in life. This period of increased excitatory neurotransmitter level coincides with a regressive, hyperactive period in the child. Other chemical changes such as low substance P, transient increase in lactic acid, pyruvate, alanine and ammonia have been demonstrated suggesting possible mitochondrial dysfunction.

These anatomical, physiological and biochemical changes have a profound effect on growth, neuro-development, physiological functions and behavior. Understanding the underpinnings of physiological, cognitive and behavioral disturbances and growth facilitates a clinician's approach to management of Rett Disorder.

Ref:

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