Rett syndrome – Unmasking complex cardio-respiratory dysfunction and possible treatment.

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Rett syndrome (RTT) is a rare X-linked dominant neurodevelopmental disorder affecting 1:10.000 girls and a few boys. Mutations in the MECP2 gene on the Xq28 are found in around 90%. Neuronal immaturity and insufficient control of the central autonomic nervous system cause cascades of signs and symptoms leading to extreme clinical challenges. Breathing dysrhythmia, insufficient control of blood gases, unstable blood pressures and heart rate derange the body's homeostasis. Rolling of eye-balls and unsteadiness due to abnormal spontaneus brainstem activation (ASBA) can easily be mistaken for epileptic fits. Oropharyngeal dysfunction and gastrointestinal dysmotility cause swallowing difficulties, aspiration, gastrooesophageal reflux, oesophagitis and constipation. Cold discoloured feet, mood swings, agitation, fear and difficulty to relax and concentrate are common. Extrapyramidal signs as dystonia with orthopaedic deformities are present including incoordination of thoughts and movements. These complex clinical challenges require multi-disciplinary care.

At the Swedish Rett Center we have identified three unique Cardiorespiratory phenotypes in RTT by use of the Neuroscope: The Forceful, Feeble and Apneustic phenotypes, each requireing unique treatment and linked to specific clinical risks. Forceful breathers may develop respiratory alkalosis, apneas and asphyxia due to loss of carbon dioxide. A rebreathing mask with carefully adjusted deadspace is on trial. Feeble breathers may become asphyxic with the feeling of strangulation due to sudden falls in oxygen and need respiratory support. Apneustic and Feeble breathers may fail to restart breathing after artificial ventilation. Correct treatment requires prior knowledge of Cardiorespiratory phenotypes.

Unique clinical managements of each Cardiorespiratory phenotype and lifelong comprehensive management on an individual basis are of great importance for the quality of life and longevity in the RTT disorder.

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