Clinical Case: Worsening Cough and Respiratory Failure as a presentation of Chiari Type 1 Malformation

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A ten-year-old female with a history of two prior lifetime seizures, obesity, and recent rapid weight gain presented with a cyanotic coughing fit to an outside hospital where she was found to have likely acute on chronic hypoxemic and hypercapnic respiratory failure. She was initially treated for severe asthma exacerbation - but further workup was concerning for sleep-disordered breathing requiring initiation of BiPAP, dysphagia, and presumed aspiration pneumonia. After stabilization and NG tube placement, she was transferred to our center for neurologic evaluation. On further history, her parents described four months of worsening cough, voice change, and progressive dysphagia with liquids and solids. Recent rapid weight gain was confirmed on our review of PMD growth charts. Her exam was notable for a nasal-sounding voice, significant oral secretion burden, left trochlear nerve palsy, left upgoing plantar response, and right-sided dysmetria with normal reflexes. Barium swallow study showed gross aspiration across all textures and severe pharyngeal weakness. Endocrinology was consulted due to concern for possible ROHHAD syndrome, however pituitary function testing was normal. Neurology was immediately consulted due to our clinical concern for subacute descending paralysis in the context of multi-focal bulbar weakness and upper motor neuron signs on exam. They recommended MRI Brain and myasthenia auto-antibody testing. Serum AChR-binding antibody and Anti-MuSK antibody resulted negative. MRI Brain demonstrated Chiari type 1 malformation with cervicothoracic syrinx. Neurosurgery was consulted, and she underwent suboccipital craniectomy and expansile duraplasty. She required discharge with G-tube feeds and overnight BiPAP, and at two week follow-up was noted to have improved dysphagia with tolerance of soft solids.

Discussion:

1. While Chiari malformations are congenital, symptomatology often doesn't develop until adolescence or adulthood. Thus, they should remain on the differential for acute neurologic presentations in older children.
2. This patient's progressive bulbar symptoms may have correlated with extension of her cervicothoracic syrinx over time.
3. Obesity has been associated with Chiari malformations in adults. Our patient's recent rapid weight gain (in the setting of severe dysphagia and likely decreased caloric intake) could have been secondary to hypothalamic obesity due to her Chiari.

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