

CLINICAL CASE ROUNDS IN CHILD AND ADOLESCENT PSYCHIATRY

Commentary: Rapid-onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): Remember Your ABCs (Airway, Breathing, Circulation)**Debra E. Weese-Mayer MD^{1,2}; Casey M. Rand BS²; Diego Ize-Ludlow MD³**

Rapid-onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD) is a devastating disorder that manifests in seemingly normal children, most typically between two and nine years of age. Because of the unique constellation of symptoms comprising the ROHHAD phenotype and inadequate public and medical awareness of the entity, diagnosis of affected children is often delayed and intervention is repeatedly incomplete and unsustainable. Extensive experience with ROHHAD patients in the Center for Autonomic Medicine in Pediatrics (CAMP) at Ann & Robert H. Lurie Children's Hospital of Chicago and the International ROHHAD Research Consortium informs us that children without early and adequate support of their hypoventilation (providing an adequate airway and breathing support) and anticipatory management of the ROHHAD phenotype (with particular attention to fluid balance and circulation) are most likely to have significant behavioral issues, deterioration, and heightened risk for sudden death.

The authors of this published case of a 14 year old teenager, described ten years after initial rapid-onset obesity, should be commended for discussing ROHHAD from a psychiatry perspective. Many aspects of the ROHHAD disease burden seem insurmountable to caregivers, but the psychiatric symptoms and mood issues are described as the most devastating. Indeed, at times these symptoms may be so overwhelming that the other potentially life-threatening features of the ROHHAD phenotype escape attention. Consequently, it is imperative that our psychiatry colleagues become experts in the diagnosis and care of children with

ROHHAD and that other specialists increase their awareness of the psychiatric phenotype because ROHHAD necessitates integrated care across *all* disciplines.

Repeated mention of "exacerbation" in the published case report suggests an incomplete knowledge of the typical ROHHAD phenotype. To our knowledge, children with ROHHAD do not have "exacerbations." Rather, there is an "unfolding" of the clinical features with advancing age such that vigilant care is needed to anticipate the "next" features in the constellation of findings that comprise the unique ROHHAD phenotype. The endocrinologist needs to move past consideration of exogenous obesity when interviewing an otherwise slim family to note the elevated prolactin levels, then at subsequent evaluations features of adipic hypernatremia and still later hypothyroidism. The respiratory physiologist or a sleep expert might note the obstructive sleep apnea but they must be anticipating the alveolar hypoventilation that is unveiled after treatment of the obstructive sleep apnea. The autonomic expert might notice symptoms of autonomic dysregulation including the hypothermia, vasomotor insufficiency, and bradycardia, but they need to be watching for orthostatic intolerance and sequelae of unnoticed volume depletion. And in the event of a tumor of neural crest origin (ganglioneuroma or ganglioneuroblastoma), the oncologist needs to be attentive to the history of rapid weight gain so that prompt referral to a Center of Excellence for ROHHAD can be made. Taken together, specialists in all disciplines must take the lead from the general pediatrician who will be the first to note the dramatic rapid-onset weight gain.

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Centralization of each child's care is essential to provide meticulous anticipatory management including prospective evaluation at two to three month intervals including physiologic monitoring in the laboratory and the home evaluating for the respiratory deterioration during sleep and wakefulness. Collaborative efforts on the part of all physicians providing complementary care to the child with initial features of the ROHHAD phenotype will optimize that child's outcome. But diagnosis is only the first step in caring for the child with ROHHAD. Without aggressive attention to the child's airway, breathing, and circulation, there is potential for irreversible deterioration in the child with ROHHAD. In the published case, desperation must have been sensed such that diaphragm pacers were surgically implanted in a child with no tracheostomy despite poor compliance with bi-level mask ventilation. Taken together, these features indicate that adequate airway and ventilatory support have not been achieved. Ideally, ventilatory management should be proactive and targeted to the child's specific needs to optimize life support. Sustained compliance with ventilatory recommendations is essential for providing safety. Without continuous attention to airway, breathing, and circulation in the child with ROHHAD, it is not possible to interpret the effect of the proposed medications on the described insomnia, severe anxiety, auditory hallucinations, and disabling

fears, and to maintain focus on neurocognitive outcome as we continue to work toward identification of a genetic basis and potential intervention strategies to decrease disease burden for these extraordinary children with ROHHAD.

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