ROHHAD syndrome: clinical case and literature review

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Background. Sleep-related breathing disorders are a potentially lethal conditions characterized by multiple episodes of sleep apnea. Study of these conditions seems to be relevant for pediatrics due to the difficulties of diagnosis and the lack of actual effective treatment strategies. This article describes the clinical case of the rare ROHHAD syndrome which represents a condition related to central sleep apnea.

Objective. To review the literature data on sleep-related breathing disorders and present our own clinical case of the rare ROHHAD syndrome.

Methods. More than 100 literature sources were reviewed. Presented the case of a 6-year-old child with sleep apnea hospitalized to the Mordovian Republic Clinical Hospital.

Results. ROHHAD syndrome is a very rare disease (about 100 cases are described worldwide). In most cases it presented as rapid obesity development for 3–5 years of age as well as hypoventilation, endocrine dysfunction, electrolyte disturbances. Autonomic nervous system dysfunction reveled in ROHHAD pediatric patients rarely. Own observation demonstrated a typical clinical presentation of ROHHAD syndrome. Boy D., 6 years 11 months was admitted to the pediatric department of the Children's republican clinical hospital with severe weakness, fatigue during physical exertion, obesity, shortness of breath at rest, orthopnea, inability to sleep on the back (sleeps with an elevated head end or on the right side); lower extremities weakness during walking and climbing upstairs during the last year, changes in walking and in behavioral reactions. Cyanotic attacks and tachycardia have appeared during sleep in previous six months before admission. Firstly, the condition manifested with sudden and rapidly progressive obesity. Ventilation disorders became clinically evident after carried over pneumonia. Episodes of sleep apnea lasted up to 20 seconds, were accompanied by cyanotic attacks and tachycardia. Multiple investigations were performed to exclude systemic connective tissue diseases, neurological disturbances and brain pathology, muscular tissue pathology and metabolic diseases. Finally, the diagnosis was confirmed by the polysomnography and genetic tests. The ventilation support during sleep was prescribed.

Conclusion. The rarity of the disease leads to a decreased awareness of pediatricians. The diagnosis is made by clinical picture and polysomnography, and timely diagnostics can increase the duration and improve the quality of life of patients.