hierarchies, and signal propagation schemes that challenge current paradigms of endocrine regulation. These findings provide an essential framework for developing targeted endocrine profiling approaches and understanding system-wide hormone dysregulation in pathological states.

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P795

JOINT2950

Respiratory and sleep disorders in children with prader willi syndrome: prevalence and characterization

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Introduction

Prader Willi Syndrome (PWS) has a broad clinical spectrum whereas respiratory disorders are major causes of morbidity and mortality across the lifespan. The approach must include close monitoring to minimize the risks.

Aim

To describe respiratory and sleep patterns in a cohort of PWS children.

Materials and Methods

Retrospective evaluation of respiratory and sleep patterns in a cohort of 25 pediatric PWS patients admitted at Ricardo Gutierrez Children hospital from 2002 to 2024. Variables analyzed were body mass index (BMI kg/m^2), pituitary function (TSH, free T4, cortisol), recombinant human growth hormone treatment (rhGH 1 mg/m^2/d), tonsils volume; obstructive sleep apneas (OSA), central apneas (CSA), nocturnal hypoventilation by Polysomnography (PSG) and daytime sleepiness by multiple sleep latency test (MSLT).

Results

Median age at admission was 3.3yr (0.4 to 13.6yr), 52% boys. The genetic mechanism was the deletion of 15q11.2-q13 chromosomal region (60%) and maternal uniparental disomy (40%). Hypothyroidism was diagnosed in 21% and none had adrenal insufficiency until their last examination. Basal median BMI was 2.35 SDS (-1.9 to 11 SDS). Median time between diagnosis and respiratory evaluation was 4.9yr (0.8 to 18.9yr). Anamnesis revealed snoring in 56% and clinical examination showed tonsillar hypertrophy in 83% of the cases. Twenty-one patients were evaluated prior to rhGH indication: 18/21 (86%) had OSA, 3 CSA and 1 nocturnal hypoventilation. Ulterior indications were only clinical follow up in 8/21, tonsillectomy in 5/21, tonsillectomy and non-invasive mechanical ventilation in 1. Seven patients were suitable to begin rhGH treatment whereas 4 patients required specific interventions before rhGH indication. Three patients were already under rhGH at first respiratory evaluation: 1 had mild OSA. None of the patients who underwent tonsillectomy developed velopharyngeal insufficiency post-surgery. Sleep disorders were diagnosed in 5/25: narcolepsy in 3 cases and cataplexy in 2. Individualized medical interventions were implemented. Six patients under rhGH developed respiratory disorders during follow-up (1 to 4 yr): 5 tonsillar hypertrophy and OSA, and 1 CSA concomitant with respiratory infection. Median ΔBMI was 0.01 (-0.37 to 2.86 SDS). In patients under rhGH there was no significant correlation between the occurrence of respiratory events and ΔBMI.

Conclusion

Respiratory and sleep disorders were a prevalent comorbidity in this cohort of PWS children and detected only by careful anamnesis and specific tests performed by trained specialists. Regular examination of respiratory and sleep disorders before and during rhGH treatment should be included in the multidisciplinary approach of PWS.

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JOINT605

Evaluation of the relationship between metabolic-associated fatty liver disease and sarcopenic obesity in patients with diabetes

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Objective

To assess the relationship between metabolic-associated fatty liver disease (MAFLD) in patients with diabetes and sarcopenic obesity (SO) indices.

Materials and Methods

The study included 92 patients with diabetes (45 men and 47 women). The average age of patients was 55.90 ± 14.18 years and duration of diabetes was 16.80 ± 9.34 years. Muscle strength (MS) was assessed using hand dynamometry measuring arm strength. Muscle function (MF) was assessed using the 4-meter walking speed test. Muscle mass (MM) and fat mass (FM) were assessed using dual-energy X-ray absorptiometry (DXA) ("lunAR prodigy") as the ratio of appendicular muscle mass (AMM) to height (AMM/m^2), AMM to weight (AMM/weight), FM to height (FM/m^2). The body mass index (BMI), waist circumference (WC), and glycated hemoglobin (HbA1c) were determined. The presence of MAFLD was determined based on the results of ultrasound examination of the liver and calculated diagnostic indices (ST-index, FLI). Statistical processing was performed using the statistical program «Statistica 10.0».

Results

In patients with diabetes, 86% of cases taking into account the ST- index and 96% of cases taking into account the FLI were found to have SO. FLI is positively associated with the age of patients ($r_s = 0.46$; $p < 0.05$), BMI ($r_s = 0.92$; $p < 0.05$), WC ($r_s = 0.90$; $p < 0.05$), FM/m2 ($r_s = 0.69$; $p < 0.05$), AMM/m2 ($r_s = 0.46$; $p < 0.05$) and negatively associated with MS ($r_s = -0.34$; $p < 0.05$), MF ($r_s = -0.32$; $p < 0.05$), AMM/weight ($r_s = -0.64$; $p < 0.05$). ST- index is positively correlated with patients' age ($r_s = 0.68$; $p < 0.05$), BMI ($r_s = 0.88$; $p < 0.05$), WC ($r_s = 0.92$; $p < 0.05$), FM/m^2 ($r_s = 0.70$; $p < 0.05$), AMM/m^2 ($r_s = 0.34$; $p < 0.05$) and negatively correlated with MS ($rs = -0.33$; $p < 0.05$), MF ($r_s = -0.45$; $p < 0.05$), AMM/weight ($r_s = -0.62$; $p < 0.05$). FLI and ST-index are not correlated with the duration of diabetes and the level of Hb1c ($p > 0.05$).

Conclusion

In patients with MAFLD and diabetes, 86% to 96% of cases show SO associated with the patient's age and independent of the duration of diabetes and the Hb1c level. MAFLD in diabete is associated with a change in body composition due to a decrease in MM and an increase in FM, the degree of decrease in MS and MF.

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JOINT3328

Decoding clinical heterogeneity in ornithine transcarbamylase deficiency: novel mutations and therapeutic outcomes from a longitudinal cohort study

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Objective

To characterize the clinical spectrum, genetic landscape, and long-term therapeutic efficacy in pediatric-onset ornithine transcarbamylase deficiency (OTCD).

Methods

A retrospective longitudinal cohort study of 7 OTCD patients (3 males, 4 females) with confirmed biochemical/genetic diagnoses and ≥24-month follow-up (median 38 months, range 16-41). Comprehensive analyses integrated metabolic profiling, next-generation sequencing, and multidisciplinary outcomes assessment (neurological, hepatic, developmental).

Results Clinical presentation

Age at diagnosis ranged from 10 days to 7.3 years. Hyperammonemic crises (71.4%, 5/7) manifested as recurrent vomiting (100%), encephalopathy (60%), and seizures (40%), with 42.9% (3/7) exhibiting pre-existing neurodevelopmental delays. **Genetic landscape:**Seven distinct *OTC* mutations identified, including two novel pathogenic variants (c.241T>C, c.490T>G; ACMG Class IV) unreported in global databases. **Therapeutic trajectories:**Medical therapy cohort ($n = 2$): Intermittent hyperammonemia (45-78 μmol/l) persisted in patients receiving sodium phenylbutyrate or arginine/citrulline, correlating with

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