Severe OSA in a morbidly obese child – do not forget the geneticist!

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Obesity is an epidemic affecting up to 20% of children in Israel. Monogenic obesity accounts for a small proportion of these children. The eceptor (MC4R) most common of these are mutations in the melanocortin 4 r with an estimated allelic prevalence of 1:2000.

Herein we describe a 17-old female that presented with excessive weight gain from early-year childhood. She related complications including-developed early onset obesity temic hypertension, insulin resistance, sys recurrent cholecystitis, and abnormalities of nocturnal ventilation with chronic CO_2 retention. She was subsequently diagnosed with C271R MC4R mutation. At age 11, she weighed 159kg and was referred for sleeve phy (PSG) at that time was positive for nocturnalgastrectomy. Polysomnogra hypoxemia, but no sleep level-apnea (AHI 0.7/hour) and she was prescribed bi positive pressure ventilation, 24/4cmH_2O (BPAP) with supplemental up-oxygen. Shortly thereafter, she was lost to follow.

she had regained 6At age 1 surgery (166 kg). She was complaining of excessive-all the weight lost post daytime sleepiness (EDS) and had nocturnal hypoxemia on home monitoring. A PSG performed while on her BPAP device demonstrated severe residual OSA with an AHI moderate -hour. Echocardiogram revealed mild/13.5 of pulmonary hypertension. BPAP pressures were increased to 28/7cmH_2O with 3 LPM of supplemental oxygen, which improved symptoms and normalized nocturnal arterial blood gases.

This report emphasizes the o consider genetic diagnoses in the evaluation of OSA in obese childrenneed t referred for PSGs. We suggest that the genetic diagnosis should guide the aggressiveness of therapy of both obesity and OSA. Furthermore, it stresses the with repeat pressure assessments importance of close follow up.