Neupane, Pritam & Manchanda, Shalini & Bodkin, Cynthia & Sigua, Ninotchka. (2019). 1017 Unusual clinical course of congenital central hypoventilation syndrome: is outgrowing the syndrome possible?. Sleep. 42. A409-A410. 10.1093/sleep/zsz069.1014.

Introduction

Congenital Central Hypoventilation Syndrome (CCHS) is a rare disease diagnosed mostly in neonatal or early childhood period. Typically, this condition requires lifelong ventilatory support. Many newer presentations of this syndrome have been described after the availability of the genetic probe for diagnosis. Here we describe a case where a patient no longer has the symptoms of CCHS in her twenties after a typical clinical course during childhood.

Report of case

A 29 years old female was seen in our adult sleep medicine clinic given issues with her noninvasive ventilation (NIV) machine. History was significant for respiratory symptoms of CCHS since infancy, however she was only diagnosed and treated with NIV at the age of seven. She had genetic probe analysis which showed polyalanine repeat mutations (PARMs) in paired-like homeobox 2B (PHOX2B) gene with 20 polyalanine repeat normal allele and a very large expanded allele. As a child, she was followed regularly in the pediatric clinic with sleep studies, imaging studies and blood work as per practice standards for CCHS. She had evidence of hypoventilation in all sleep studies and presented with clinical symptoms of headache, fatigue, and exercise intolerance whenever she missed using her NIV.

Over years, she had gained weight but had no sleep apnea symptoms. Upon presentation in the clinic, she has not used nocturnal NIV for 8 months due to issues with the machine. Surprisingly, she was completely asymptomatic, without any recurrence of her CCHS symptoms. Her serial polysomnogram showed continued improvement in her baseline end tidal CO2 and the severity of hypoventilation at night.

Conclusion

While respiratory arrest was believed to be the presenting feature of CCHS, it is now known that this syndrome has varied presentations. This is explained by reduced and variable penetrance of the autosomal dominant inheritance of this mutation. Spontaneous improvement or resolution of symptoms has not been reported for CCHS. Findings like this could alter our understanding of the natural history of this condition which otherwise is usually considered to be grim. Further analysis of specific molecular mechanisms in these unusual cases could improve our understanding of this fascinating disease.