SciMedCentral

Archives of Paediatrics and Developmental Pathology

Perspective

Congenital Central Hypoventilation Syndrome

Mary O'Connor*

School of Nursing, Notre Dame of Maryland University, USA

***Corresponding author**

Mary O'Connor, School of Nursing, Notre Dame of Maryland University, Baltimore, Maryland, USA, Tel: 816 916 1526; Email: oconnormary@umkc.edu

Submitted: 10 October 2017

Accepted: 21 October 2017

Published: 24 October 2017

Copyright

© 2017 O'Connor

OPEN ACCESS

The aim of this manuscript is to inform providers of neonatal and infant care of the symptoms and Diagnosis of Congenital Central Hypoventilation Syndrome (CCHS). Hypoventilation, under breathing, is caused by a disorder in the bottom of the brain anterior to the back of the neck. Most CCHS individuals do not stop breathing all together; rather they fail to breathe deeply enough to support the body functions which are compatible with life. This apnea state result in a lack of oxygen uptake, failure to exhale sufficient carbon dioxide causes rapid accumulation of waste products which contributes to organ failure and the risk for sudden unexpected death. A low concentration of O_2 in red blood cells may cause hypoxia induced pulmonary hypertension ending in right sided heart failure leading to risk of death.

The central and autonomic nervous systems work together to control automatic body functions: blood pressure, blood levels of O_2 and CO_2, bladder and bowel control, heart rate and temperature. The inability to control breathing due to a central or autonomic nervous system injury or developmental variance is well known. The severity of this lack of breath control varies resulting in the need in mild cases for only a change of position to stimulate inhalation to cases with a severe need which would require continuous ventilation support. Some CCHS patients require ventilation support during hours of sleep [1].

Causes of CCHS and SIDS are being studied. In 1962 when babies ceased spontaneous respiration when asleep the term used was Online's Curse. Online in a German legend placed a course on her mortal husband who had been less than faithful. The curse removed the husband's automatic functions requiring that he needed to remember to breathe. If the husband went to sleep he stopped breathing. The current diagnostic term for infants who cease spontaneous respiration when asleep is CCHS.

There are rare germ line mutations of the PHOX2B gene which impairs the function of PHOX2b protein [2]. This is the only gene mutation known to cause CCHS. A PHOX2B mutation is present before birth: it has varied effects. Symptoms with this mutation are respiratory control failure in 90% of patients; central hypoventilation, recurrent pneumonia, hypotonia, temperature deregulation, cardiovascular abnormalities, growth impairment, development variances, ganglio neuroma, ganglio neuroblastoma, ophthalmic variances and bowel issues including gastro-esophageal reflux, feeding issues and constipation also occur with less frequency [3,4]. Some PHOX2B variant patients function at a normal or high level. In contrast some will experience academic challenges. Most PHOX2Bmutations are new occurrences, de novo. Between 5 to 10% of CCHS are from a mosaic unaffected parent [5].

Mutations can be present in all the cells including germ line reproductive cells of the parent yet the parent is unaffected and asymptomatic. Poly-alanine repeat expansion mutations (PARMS) occur in 90% of children with CCHS. Non-polyalanine mutations (NPARM) also occur. Newborns with CCHS have monotonous respiratory rate, hypoventilation, shallow breathing when awake and asleep exhibit autonomic nervous system deregulation. Late onset CCHS needs to be considered with cases of Sudden Infant Death Syndrome (SIDS) and in Sudden Unexplained Death of Childhood (SUDC).

CCHS is a rare diagnosis. Male and female occurrence rates are equal. In mild cases the diagnosis may be missed until later in life when after receiving anesthesia, anti-seizure medications or sedatives there is a subsequent respiratory arrest. By 2013 worldwide one thousand cases of CCHS were known. The prevalence within society, NICUS, ICUS and SIDS deaths remains unknown. Diagnosis and ventilation can help patients sleep safely and live.

REFERENCES

1. Carroll M, Patwari P, Kenny A, Brogdir C, Stewart T, Weese-Mayer D. Residual chemosenstivity to ventilator challenges in genotyped congenital central hypoventilation syndrome. J Appl Physiol. 2014; 116: 438-450.

2. Jennings L, Yu M, Rand C, Kravis N, Berry-Kravis E, Patwari P, et al. Variable human phenotype associated with novel deletions of the PHOX2B gene. Pediatric Pulmonol. 2012; 47: 153-161.

3. Rand C, Yu M, Jennings, Panesar K, Berry-Kravis E, Zhou L, et al. Germline mosaicism of PHOX2B mutation accounts for familial recurrence of congenital hypoventilation syndrome (CCHS). Am J Med Genet. 2012; 158A: 2297-2301.

4. Saiyed R, Rand C, Patwari P, Koliboski C, Stewart T, Peters P, et al. Altered temperature regulation in respiratory and autonomic disorders of infancy, childhood, and adulthood (RADICA). Am J Respiratory Critical Care Med. 2011; 183A: 6394.

5. Weese-Mayer D, Patwari P, Rand C, Diedrich A, Kuntz N, Berry-Kravis E. Congenital central hypoventilation syndrome (CCHS) and PHOX2B mutations. In: Robertson D, Biaggioni I, Burnstock G, Low P, Paton J. editors. Primer on the Autonomic Nervous System. Oxford, UK: Academic Press. 2012; 445-450.