**Polysomnographic features in patients affected by congenital central hypoventilation syndrome (CCHS)**

Janis S, Vari MS, Romanello S, Ottonello GC, Baglietto MG

Child Neuropsychiatry Unit, Genoa University

*Correspondence to:*Dr Giancarlo Ottonello. [giancarloottonello@gmail.com](mailto:giancarloottonello@gmail.com)

**Objectives**

CCHS is a rare neurocristopathy characterized by abnormal automatic ventilation control and absent response to hypocapnia and hypoxia. Early diagnosis and evaluation of the disease severity may help to define treatment strategies in order to optimize ventilation and improve outcome. The aim of the study is to analyze the sleep patterns in patients affected by CCHS and correlate these findings with disease severity, evolution and treatment.

**Methods**

The cohort included 9 patients (5 females and 4 males), age 1-17 years old, 4 with tracheostomy, and 5 on non-invasive ventilation (NIV). OPTILEX GX520 system was used for polysomnography; data were authomatically analysed, revised and graphically elaborated by the neurologist and statistician, and compared with published data.

**Results**

In the 3 patients in the range of age from 1 to 5 years sleep macrostructures is preserved. In the 3 patients in the range of age from 6 to 12 years, with higher interindividual variability, abnormal values compared to healthy subjects matched for age were found.

In 4 patients in the first and second range of age with preserved macrostructure, Arousal Index (AI) was calculated and correlated to the ongoing treatment. In patients with tracheostomy AI was increased and greater than in patients on NIV

**Conclusions**

Matching the sleep macrostructure with normal sleep macrostructure for age we observed that, in absence of environmental influence and with a good clinical management, both with NIV and tracheostomy, sleep organization appear to be better preserved in younger patients. This observation needs to be confirmed by further study on larger cohort of case.

**References**:

1) Congenital central hypoventilation syndrome: genotype-phenotype correlation in parents of affected children carrying a PHOX2B expansion mutation. [Parodi S](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Parodi%20S%22%5BAuthor%5D), [Vollono C](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Vollono%20C%22%5BAuthor%5D), [Baglietto MP](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Baglietto%20MP%22%5BAuthor%5D), [Balestri M](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Balestri%20M%22%5BAuthor%5D), [Di Duca M](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Di%20Duca%20M%22%5BAuthor%5D), [Landri PA](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Landri%20PA%22%5BAuthor%5D), [Ceccherini I](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Ceccherini%20I%22%5BAuthor%5D), [Ottonello G](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Ottonello%20G%22%5BAuthor%5D), [Cilio MR](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Cilio%20MR%22%5BAuthor%5D). [Clin Genet.](javascript:AL_get(this,%20'jour',%20'Clin%20Genet.');) 2010 Sep;78(3):289-93. Epub 2010 Feb 11.

2) Sleep classification according to AASM and Rechtschaffen and Kales: effects on sleep scoring parameters of children and adolescents. [Novelli L](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Novelli%20L%22%5BAuthor%5D), [Ferri R](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Ferri%20R%22%5BAuthor%5D), [Bruni O](http://www.ncbi.nlm.nih.gov/pubmed?term=%22Bruni%20O%22%5BAuthor%5D). [J Sleep Res.](javascript:AL_get(this,%20'jour',%20'J%20Sleep%20Res.');) 2010 Mar;19(1 Pt 2):238-47. Epub 2009 Nov 11