**Central Congenital Hypoventilation Syndrome and paroxysmal events: not only epilepsy**

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**Background.** Central congenital hypoventilation syndrome (CCHS) is a genetic disease affecting respiratory control, with life-long dependence on mechanical ventilation. Affected patients can also present with autonomic nervous system anatomical and functional abnormalities. Seizures are described as a possible associated disorder in children with CCHS, likely because of hypoxemia episodes, but other paroxysmal events are often under diagnosed in these patients and confused with epileptic seizures.

**Methods.** PPC Center of Padua University has followed children with CCHS since 2003 providing multidisciplinary and continuous care. We retrospectively reviewed charts and computed database about patients with CCHS who presented seizures or seizure-like episodes.

**Results.** To date Center has followed 8 patients (4m/4f), mean age 6.4 years old. Seizures or seizure-like episodes concerned 6 children, with various presentation and diagnostic approach (see table).

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| Age and sex | Genetic mutation | Paroxysmal events | Investigations | Treatment |
| M, 7.2 years | 20/27 +sm | Breath-holding spells (at 2 yrs old) |  | Martial therapy |
| Focal motor seizures with secondary generalization (3-4 yrs old) | Intercritical EEG: negative. Critical EEG: alterations. | Valproic acid |
| Absence seizures (from 4 yrs old to date) | Intercritical EEG: alterations. Critical EEG: not available. | Vigabatrin |
| M, 4.4 years | 20/26 | Clonic movements, staring and breathing difficulties during sleep; hypotonia, unresponsiveness, sweating and cyanosis while awake (from 4 mths old) | Intercritical EEG: negative.  Critical EEG: alterations. Glycemic blood test: hypoglycemia during event. | Valproic acid: unsuccessful; diazoxide therapy successful also for seizures |
| F, 7.3 years | 20/32 | Sudden generalized hypotonia and unresponsiveness followed by sleeping for about 1 hour. Preceding severe sudden frontal headache (6-7 yrs old) | EEG always negative.  72h HolterECG negative.  7daysHolterECG: long sinus pauses | Pacemaker positioning with resolution of episodes of syncope. |
| M, 8.3 years | nd | Convulsion (9 months). | Intercritical EEG. Negative.  Critical EEG not available; Glycemic blood test: hypoglycemia during events. | Octreotide to control glycemia values. Valproic acid for seizures control. |
| F, 7 years | 20/29 | Suspected absence seizures | EEG negative | Follow-up |
| F, 11 years | nd | 4 episodes of generalized rigidity, head turning, eye revulsion, followed by cry and agitation (8 yrs old) | EEG negative;  Long sinus pauses at HolterECG | Pacemaker positioning with resolution of anoxic seizures |

**Conclusions.** Children with CCHS can present epileptic seizures often due to episodes of unrecognized hypoxiemia or hypoglycemia, but it is important to stress that paroxysmal events other than epileptic seizures can occur, such as lipothymia, syncope due to cardiac rhythm abnormalities. In particular some patients can present with anoxic seizures after prolonged sinus pauses.