CONGENITAL CENTRAL HYPOVENTILATION SYNDROME : A CLINICAL OVERVIEW OF 8 CHILDREN FOLLOWED BY AN ITALIAN PEDIATRIC PALLIATIVE CARE CENTER

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**Background.** Congenital central hypoventilation syndrome (CCHS) is a genetic disorder usually presenting with reduced physiological response to elevated CO2, loss of respiratory drive, rapidly progressing hypoxiemia and hypercarbia. Therefore patients have a lifelong dependence on mechanical ventilation. Disease associated conditions concern anatomical and functional abnormalities of the autonomic nervous system.

**Methods.** Pediatric Palliative Care (PPC) Center of Padua University has followed children with CCHS since 2003, providing continuous care. We retrospectively reviewed charts and medical records to obtain an overview of relevant clinical problems in children with CCHS during follow-up.

**Results.** Data concerned 8 patients (4m/4f), mean age 6.4 years old. All of them have genetic confirmation of disease. All patients underwent tracheostomy in the first months of life and use mechanical ventilation during sleeping and during respiratory infections; LTBS was performed annually; no child required re-intervention during the follow-up. Tracheostomy tube had been changed monthly at home by families. Hospital admissions due to ventilatory management difficulties were limited.

All patients presented associated disorders (see table). No one had diagnosed neural crest tumor during the follow-up. All children were annually submitted to a complete neurological evaluation: 6 patients showed a moderate to severe psychomotor retardation, particularly for the aspect of language production. At neurological imaging (MRI) 1 patient showed rostral brain nonspecific injuries.

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| N | Sex | Age | Genetic mutation | Tracheostomy (age) | Cardiac | Neurological | Oculistic | Other |
| 1 | m | 7.2 | 20/27+sm | 3 mths | No | Seizures | Strabismus, visual defect | Emotional spasm, diabetes insipidus, abdominal pain  |
| 2 | m | 4.4 | 20/26 | 36 days | No | Seizures (related to hypoglycemia) | Pupillary asymmetry, visual defect | Profuse sweating, hypoglycemia episodes |
| 3 | f | 7.3 | 20/32 | 2 mths | Cardiac asystole, pacemaker positioned | Headhache | Pupillary asymmetry | Neonatal hypoglycemia, abdominal pain,diarrhea, profuse sweating  |
| 4 | f | 7.0 | 20/29 | 4 mths | No | Enuresis,suspected absence epilepsy | Pupillary asymmetry | Constipation |
| 5 | m | 8.4 | nd | 2 mths | No | Seizures with and without hypoglicemia | No | Hypoglicemia episodes |
| 6 | f | 11 | nd | nd | cardiac asystoles, PM positioned | Seizures | Strabismus | Hyperbilirubinemia |
| 6 | f | 11 | nd | nd | cardiac asystoles, PM positioned | Seizures | Strabismus | Hyperbilirubinemia |
| 7 | f | 2.6 | 20/26 | 1 mths | No | No | No | Profuse sweating |
| 8 | m |  | 20/27 | 40 days | Long pauses at Holter | No | Pupillary asymmetry, reduced papillary light response | Hirschsprung disease,profuse sweating, emotional spasms |

**Conclusions.** The immediate concern for children with CCHS is the loss of respiratory drive, which imposes the prompt introduction and continuation of ventilation support. Furthermore they need a multidisciplinary complex management. In our experience most relevant aspects in the follow up have been: cardiac rhythm abnormalities detection; seizure diagnosis and treatment; endocrine-metabolic abnormalities; gastroenterological anatomical and functional problems management. Oculistic abnormalities and psychomotor retardation should also be considered because they can severely affect daily life.