**Neurodevelopment in Congenital Central Hypoventilation Syndrome Children**

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**Introduction**: Children with Congenital Central Hypoventilation Syndrome (CCHS) are at risk of developing neurocognitive and developmental impairments. Episodes of hypoxemia, seizures, cardiac pauses, level of ventilation support, repeated hospitalizations, are some of the factors which have been incriminated. Standardized neurodevelopmental assessment has not been used in a systematic way, although impairment of intellectual and psychomotor functioning seems to be rather common in CCHS.

**Objective**: Neurodevelopmental characterization of CCHS children currently followed at HPC.

**Methods**: Retrospective analysis of the clinical reports and of the neurodevelopmental evaluation of the seven CCHS children who are currently followed in the HPC.

**Results**: At present, seven CCHS children (4 F/3M) are followed in the HPC. Ages range from 3 to 12 years. Genetic testing revealed PHOX2B heterozygoty in six of them - 20/26(4), 20/27(1), exon 1 c.23dupA.pY8X mutation (1), and PHOX2B homozygoty in one (20/24). One child, a 10-year old boy (20/26), presents a severe mental retardation. He was a 31 week preterm with neonatal hypoxic ischemic encephalopathy, and he is on sleep ventilation through tracheostomy and has a colostomy for Hirschprung disease. Of the other children, five are on non-invasive ventilation (NIV) during sleep and one (20/26) is on sleep ventilation by tracheostomy. One child has a late-onset CCHS. Each of these six children had an neurodevelopmental assessment performed. The evaluation tools varied according to the child’s age - Weschsler Intelligence Scale for Children–Revised or Griffiths’ scale. Results were as follows: two 3-year old children (20/26 tracheostomized and exon 1c.23dupA.pY8X late-onset CCHS) have an intellectual functioning in the normal range (DQ>85). The other four revealed intelligence index lower (IQ 55-73) than the general population and all of these met criteria for Attention Deficit Hyperactivity Disorder (ADHD) – clinical DSM-IV and Conner’s questionnaires. The three eldest children, who attend normal school, present global learning disabilities and one child has a pervasive developmental disorder. All children on NIV manifest verbal articulation problems. All children have special education support.

**Comments and questions:**

*Most children have some kind of neurodevelopmental disability.*

*The two youngest are less impaired.* Did the tracheostomy in one protect the neurological development by avoiding so many episodes of hypoxia? Is the late-onset a less severe form? Did medical care improve in the latest cases?

*ADHD was a constant finding in the four eldest children.* Should it be expected in most children? Or does it depend on the adequate control of ventilation?

*Verbal articulation was affected in all children on NIV*. Is it secondary to CCHS? Or is the facial deformation with midface hypoplasia by mask pressure the main responsible factor?

**CCHS is associated to some degree of neurodevelopmental disability, the spectrum of which needs to be more clearly defined. Every effort should be undertaken to abolish possible cofactors in the establishment of neurocognitive, developmental and functional impairments.**

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