**NIV in a 2 months girl with CCHS**

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Ondine’s Curse, also know as Congenital Central Hypoventilation Syndrome (CCHS) or Primary Alveolar Hypoventilation is a multisystem disorder of the central nervous system where, as the worst case, the breathing automatic control is absent or impaired. The prevalence is 1 out of 200,000 of the living born children population.

We report a case of an infant whose diagnosis was delayed by her mild clinical pattern and who is currently ventilated through a mask without tracheostomy.

Alessia was born on due time with a weight of Kg 3,050, APGAR score: 1’ 6; 5’ 8. She went under intubation shortly after following a very severe respiratory distress; she needed ventilation for 3 weeks, and then she was addressed to our Neonatology Intensive Care Unit because of the persistent dependence on mechanical ventilation.

A week later the admission, mechanical ventilation was stopped and she was supported by oxygen (FiO2 about 30%) and nasal-CPAP (6 cmH2O) for another 20 days. She was submitted to polysomnography with the evidence of severe respiratory insufficiency, severe OSAS but normal values of carbon dioxide [mean oxygen saturation (SaO2) 77%; obstructive desaturation index (ODI) 31.5; mixed-obstructive apnea-hypopnea index (MOAHI) 11.1].

After that, she was then taken by our Unit due to a persistent O2-dependence and severe apneas with desaturations. At 3 months of age we performed the genetic test, which identified heterozygous PHOX2B mutation, in particular the duplication of 18 nucleotides (c.724-741dup18) leading to expansion of the repeat tract from 20 to 26 alanines in exon 3. The baby had normal parameters if asleep; she started eating normally leaving the naso-gastric tube with a regular growing; her neurological state was analyzed and considered normal; holter-ECG was also normal; no clinical evidence of Hirschsprung disease, nor neural crest origin tumors were present and specific examinations were negative.

We decided, in agreement with parents, to support the baby by non-invasive ventilation with 2 different masks: a nasal mask during the night and nasal pillows during the day. Her cardio-respiratory parameters during sleep were normalized.

Alessia is now 5 months. She is at home and is regularly followed by her pediatrician with whom we are in strict contact. At the first follow up after one month she was normally grown in height and weight and her parameters were normal while asleep and during sleep hours in NIV.

We will check her every 4 months, whether clinical situation is stable.