## Ocular Anomalies Correlated With Genotype In Congenital Central Hypoventilation Syndrome (CCHS)

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Background and Purpose: Congenital Central Hypoventilation Syndrome (CCHS), ultra rare disease, ocular signs, first described by Goldberg, have to be recognized. Our study aims to precise clinical ocular phenotype and genetic mutations in order to correlate phenotype genotype and adapt treatment for a better patients’ outcome.

Methods: 43 patients with CCHS were recruited at CCHS Orphan Disease Center of Robert Debre Hospital. They underwent a complete ocular examination with an orthoptic evaluation. Patient’s genetic mutations were analyzed. Ocular abnormalities were quoted and classified and then compared to the genetic mutations of the patients.

Results: Ophthalmic examination allowed to individualize ocular clinical signs that were divided in anomalies of pupil, extraocular muscle, levator palpebrae, iris and eyeball. Strabismus and esophoria screening allowed an adapted treatment. Of the 43 patients, 40 presented moderate to severe ocular anomalies. In PHOX2B genetic mutations, 39 dysfunctions from 6 to 13 ALA duplication and 2 frame shift mutations were demonstrated.

Conclusions: In CCHS, ocular abnormalities have a high incidence. Ocular motility and intrinsic pupil anomalies are the most common ocular. The correlation with genetic mutation could allow an early re-education treatment of these children for a better outcome.