

[Open Peer Review on Qeios](#)

Ondine syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Ondine syndrome. ORPHA:661*

Congenital central hypoventilation syndrome (CCHS) is a rare disease due to a severely impaired central autonomic control of breathing and dysfunction of the autonomous nervous system. The incidence is estimated to be at 1 of 200 000 livebirths. A heterozygous mutation of PHOX-2B gene is found in 90% of the patients. Association with a Hirschsprung's disease is observed in 16% of the cases. Despite a high mortality rate and a lifelong dependence to mechanical ventilation, the long-term outcome of CCHS should be ultimately improved by multidisciplinary and coordinated follow-up of the patients.