Maternal Congenital Central Hypoventilation Syndrome in Pregnancy: A Case Report

Abstract

Background: Congenital Central Hypoventilation Syndrome is an exceedingly rare condition which may predispose to compromised placental function and intrauterine growth restriction. Labor is best managed in an intensive care unit. Congenital Central Hypoventilation Syndrome (CCHS) is an exceedingly rare autosomal dominant condition with about 160-180 cases diagnosed worldwide. It is characterized by adequate ventilation while awake and alveolar hypoventilation during sleep and if untreated the affected individual may survive only one to two months of age. Given the extreme rarity of the condition ideal antepartum and intrapartum management is largely undefined.

Case: A 28 year-old primigravida with CCHS was followed from 16 weeks gestational age through delivery and post-partum course. Amniocentesis confirmed that the fetus was not affected. During the antepartum course there was a progressive lag in fetal growth and placental weight was found to be much less than the third percentile. At night mechanical ventilation was utilized and a normal spontaneous vaginal delivery was achieved in the intensive care unit.

Conclusion: CCHS is an exceedingly rare diagnosis. During pregnancy multispecialty interaction is critical for both maternal and fetal wellbeing. Fetal and placental growth is likely impacted by a hypoxic environment even with nightly mechanical ventilation. Amniocentesis and PHOX2B gene testing should be highly considered even if termination is not an option as it allows critical preparation for the newborn caregiver and other hospital personnel.

Keywords

Congenital Central Hypoventilation Syndrome (CCHS); Intensive Care Unit (ICU); Rapid Eye Movement (REM) Sleep; Non-Rapid Eye Movement (Non-REM) Sleep; PHOX2B gene