Perinatal features of Prader-Willi syndrome: a Chinese cohort

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Abstract

Background: Prader-Willi syndrome (PWS) is a rare complex genetic disorder caused by an absence of expression of imprinted genes on the paternally derived chromosome 15q11-q13 region. This study aimed to characterize the perinatal features in a cohort of Chinese individuals with PWS.

Methods: We analyzed anonymous data of 134 patients from the PWS Registry in China. Perinatal and neonatal presentations were analyzed, and compared between the two PWS genetic subtypes. We also compared the perinatal features of PWS patients with the general population and other previous reported large cohorts from France, UK and USA.

Results: This study included 134 patients with PWS (115 patients with 15q11-q13 deletion and 19 with maternal UPD). High mean maternal age was found in this cohort (30.5 vs. 26.7) comparing with the general population. 88.6% of mothers reported a decrease of fetal movements. 42.5% and 18.7% of mothers had polyhydramnios and oligohydramnios during pregnancy, respectively. 82.8% of the patients were born by caesarean section. 32.1% of neonates had birth asphyxia, 98.5% had hypotonia and 97.8% had weak cry or even no cry at neonatal period. Feeding difficulty existed in 99.3% of the infants, and 94.8% of them had failure to thrive. 69.4% of the infants ever used feeding tube during hospitalization. However, 97.8% of them discontinued tube feeding after discharged home. Maternal age, maternal pre-pregnancy weight and BMI were significantly higher in the UPD group (all $P<0.05$).

Conclusions: PWS should be highlighted for differential diagnosis for infants with following perinatal factors including polyhydramnios, intrauterine decreased fetal movements, cesarean section, low birth weight, feeding difficulty, hypotonia, and failure to thrive. Higher maternal age may be a risk factor for PWS, especially for UPD, and further studies for the mechanism of PWS are required.

Topic: Fat, Metabolic disease and obesity