复旦大学附属儿科医院

Multicenter study of early screening and prevention of Prader-Willi Syndrome

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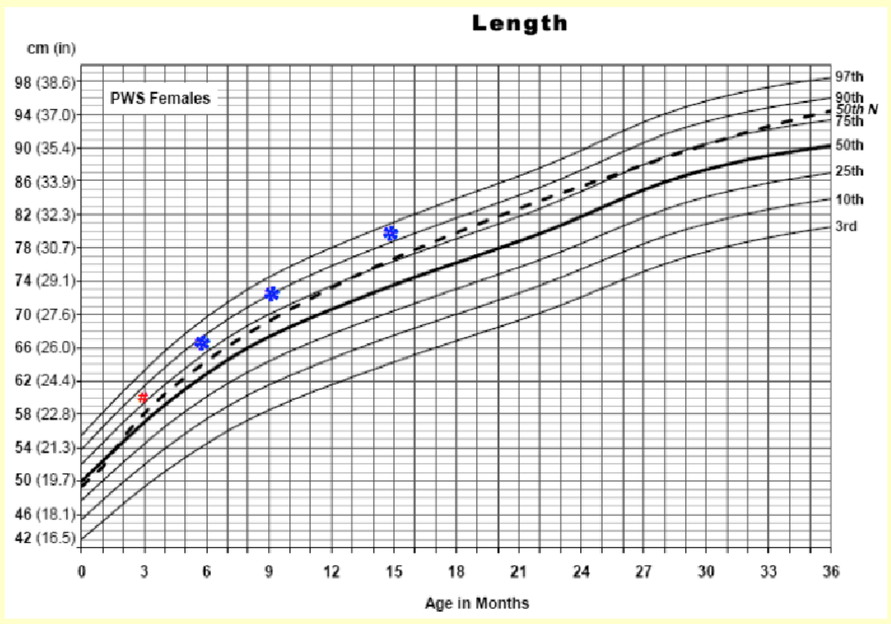
OBJECTIVES

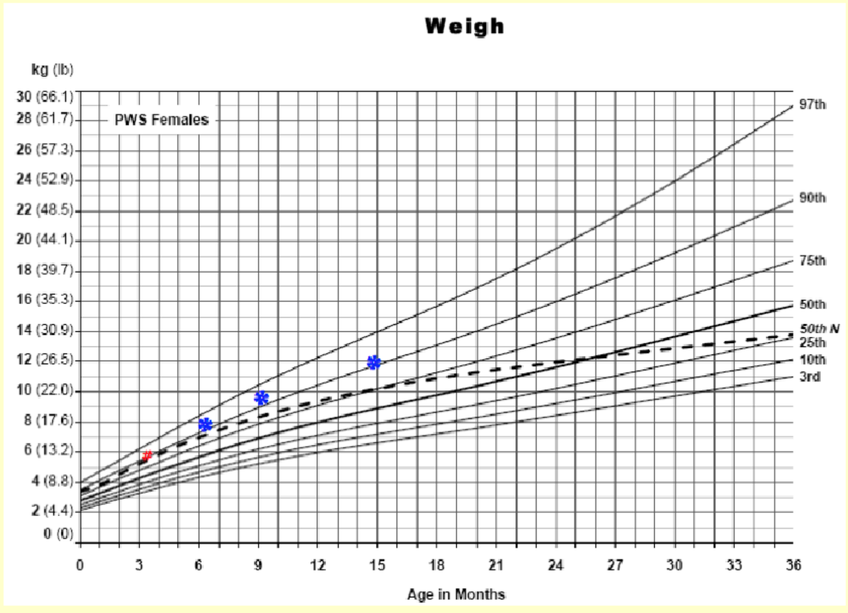
The current diagnostic criteria for Prader-Willi syndrome (PWS) is proposed by Holm *et.al.* in 1993. Although the criteria are widely accepted, it was challenging to be implemented in Chinese population.

The present study collected PWS cases from 12 centers across China. By analyzing the clinical manifestation during early infancy, we aimed to provide data for clinical characteristics, screening strategy and effect of growth hormone (GH) treatment in Chinese PWS patients.

METHODS

We screened 63 suspected PWS cases in 12 centers from May-2012 to Aug-2013 using MS-PCR. Patients diagnosed by MS-PCR further underwent analysis by MS-MPLA and STR to identify PWS genetic markers. Data on patients' history, clinical manifestation, anthropometrics and clinical biochemistry test before/after GH treatment were collected for analysis.





Height and weight curves before and after the intervention of rhGH for one case

RESULTS

Among our enrolled subjects, 16 were confirmed by MS-PCR. Further analysis using MS-MLPA and STR analysis showed that 13 were associated with paternal deletion while the rest 3 were maternal uniparental disomy (mUPD). Among the 16 diagnosed PWS, 13 were delivered at full term, 1 were preterm birth, 2 postterms, 4 delivered vaginally, 12 delivered by cesarean section. Fetal distress was diagnosed in 10 cases while abnormal fetal position found in 5 cases. All patients had reduced fetal movement, hypotonia and infant feeding difficulties. Characteristic facial appearance was found in 6 cases when 13 showed hypogonadism, 8 had hypopigmentation. There were 4 patients received rhGH treatment. When we found patients treated with GH had improved physical development, no difference was found in thyroid function, plasma IGF-1 levels, fasting blood glucose, fasting insulin levels and blood lipid levels.

CONCLUSIONS

PWS might account for 25% of infants with idiopathic hypotonia and infant feeding difficulties. Screening using MS-PCR in suspected cases is critical to identify PWS patients. Hypogonadism and hypopigmentation are important clues for diagnosis. GH treatment during infancy can improve physical development in PWS patients, however how to improve cognitive development and function of endocrine system in PWS patients requires future studies.

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