INTERNATIONAL JOURNAL OF PEDIATRIC ENDOCRINOLOGY

POSTER PRESENTATION Open Access

Analysis of clinical and genetic features among 12 neonatal diabetes mellitus

Cao Bingyan*, Gong Chunxiu, Yang Jinkui, Cao Xi, Wu Di

From 7th APPES Biennial Scientific Meeting
Nusa Dua, Bali. 14-17 November 2012

Objectives

To analyze the clinical and genetic features among 12 neonatal diabetes mellitus (NDM).

Methods

Describe clinical features of 12 diagnosed NDM cases in my hospital between Jul. 2001 and Jun. 2012. KCNJ11, ABCC8 and INS gene were sequenced in all patients.

Results

The age at diagnosis was between 0.5 and 5 month, the median was 3 month. 7 cases (58.3%) were SGA. Infection occurred in 7 cases, 4 cases with convulsion and 8 cases with ketoacidosis. The mean HbA1c at diagnosis was 10.0% (7.4%~13.7%). Insulin treatment was started in all 12 patients, the initial dose was 1.0~1.2IU/kg/d, 6 cases were treated with glyburide after the acute phase, only one boy who diagnosed as DEND syndrome reached euglycemia. 2 cases stopped glyburide because of gastrointestinal adverse reaction. Among the 12 cases followed up, 7 had PNDM, 4 had TNDM, 1 case lost following, 2 cases died, one was the DEND syndrome patient died of DKA, the other died of hepatic and renal failure at the age of 1 year and 6 months. One had skeletal dysplasia and diagnosed as Wolcott-Rallison syndrome. The blood glucose of most patients was well controlled. KCNJ11c.175G>A (V59M) and KCNJ11c.601C>A (R201H) mutation were found in two patients.

Conclusion

The clinical expressions of NDM were atypical and easily missed diagnosis. Of all the NDM cases, 30% was TNDM and relieved automatically. Insulin was the best choice before genetic identification. The KCNJ11 mutation rate of PNDM was 28% in China.

Published: 3 October 2013

doi:10.1186/1687-9856-2013-S1-P5
Cite this article as: Bingyan *et al.*: Analysis of clinical and genetic features among 12 neonatal diabetes mellitus. *International Journal of Pediatric Endocrinology* 2013 **2013**(Suppl 1):P5.

Beijing Children's Hospital, Capital Medical University, Beijing, China