Precocious puberty: a single academic center experience

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Introduction

Delayed and precocious puberty both represent rich veins for the identification of genes that regulate the timing of sexual maturation in humans. The purpose of this work is to identify novel genes that regulate puberty by taking advantage of the special genetic features of the Finns and the Finnish health care registries. Herein, we describe the diagnoses underlying PP in a single academic center with special attention in the identification of familial cases.

Patients and methods

We performed an ICD-10 code-based inquiry to the electronic patient records of the Children’s Hospital, Helsinki University Central Hospital, in order to identify patients evaluated for precocious puberty between 2001 and 2014. The following ICD-10 codes were used: E22.80, E28, E28.8, E28.9, E29, E29.0, E29.8, E29.9, E30, E30.1, E30.8, E30.9, and Q78.1 (Figure 1). The girls and boys presenting with clinical signs of puberty before age 8 years and 9 years, respectively, and with menarche at age 9.3 years or younger [representing the mean -3SD of healthy Finnish girls (2)], were included and classified into diagnostic subgroups (Table 1).

Results

• Girls (n=164) presented with PP ~10 times more frequently than boys (n=17).
• In both sexes, PP was most frequently GnRH-dependent.
• In girls presenting at age 6 years or older, GnRH-dependent PP was usually idiopathic, whereas younger girls predominantly had a pathological brain MRI finding or a disease that predisposes to PP (Figures 2A and 3).
• In boys, a pathological underlying cause was identified in 47% of cases with GnRH-dependent PP (Figure 2B).

Conclusions

• Almost one third of patients with idiopathic CPP had a positive family history of early puberty. These families are currently being enrolled to our genetic study.
• MRI scan was not performed to all patients with PP. This may reflect the implementation of national guidelines that allow tolerance in the cut-off limits if either of the parents had been an early maturer.
• As much as 14% (95% CI, 7%-27%) of 7-8-year-old girls with GnRH-dependent PP had pathological brain MRI findings lending credence to the use of current cut off age limit of PP (5).
• Our study confirmed that a boy with CPP, needs thorough endocrine examination
• Although thelarche is predominant in girls under 3-years-of-age, every third girl has an organic etiology behind the PP
• In support of previous findings (6), adoption emerged as a negative predictor of an organic cause for PP

References