Etiology of severe short stature: 
Single center experience

Juho Kärkinen, BM, Päivi J. Miettinen, MD, PhD, Taneli Raivio, MD, PhD1 and Matti Hero, MD, PhD
1Faculty of Medicine/Physiology, University of Helsinki, Helsinki, Finland
2New Children’s Hospital, Pediatric Research Center, Helsinki University Hospital (HUH), Helsinki, Finland
3Stem Cells and Metabolism Research Program, Faculty of Medicine, University of Helsinki, Helsinki, Finland

Introduction

- Severe short stature (i.e. height less than -3 SDS), at the age of > 3 years, warrants diagnostic evaluation in specialized health care. In the absence of apparent underlying cause, targeted and eventually untargeted genetic studies have been proposed (1).
- Etiology of severe short stature is poorly characterized and no reports from population level exist.
- We describe the underlying etiology of severe short stature in children over 3 years of age in a tertiary center that serves as the primary referral center for the region’s well-child and school primary health care.

Results

- A pathological cause for short stature (i.e. condition other than ISS) was diagnosed with equal frequency in girls and boys (n=286 [76%] vs. n=289 [71%], P=NS)(Figure 1).
- Sex differences were evident in favor of girls in the frequency of syndromic causes (28 vs. 13 %, P < 0.0001 ) and in favor of boys in GHD (16 vs. 8 %, P < 0.001) respectively.
- The proportion of patients with skeletal dysplasias (P <0.0001) and syndromes (P <0.0001) increased, whereas GHD (P <0.01) and ISS (P <0.001) decreased, with increasing severity of short stature (Figure 2).
- Sitting height/height SDS was increased in ISS (0.5 SDS), GHD (0.9 SDS), SGA (1.2 SDS), and skeletal dysplasia (3.6 SDS median) groups (P<0.01)(Figure 3).

Conclusions

- In contrast to previous studies, severe short stature affected girls and boys equally.
- Pathological causes for severe short stature were found in more than two-thirds in both sexes.
- Unexplained cause was extremely rare in those with height < -4 SDS.
- Increased sitting height/height ratio suggests that growth plate-related pathology contributes to growth failure in some patients with GHD or SGA.
- Our results reflect the spectrum of growth disorders at the population level, as HUCH catchment area’s well-child care is well adhered to and employs Finnish growth screening rules.

References