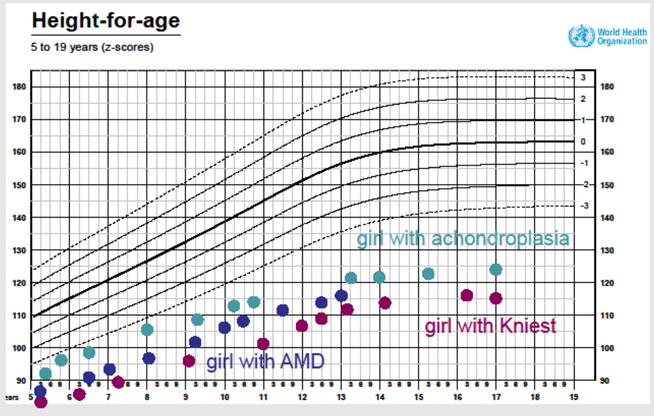
Achondroplasia height reference as a background matrix when following children with extreme short stature

Andrea Merker[1], Luitgard Neumeyer[1], Thomas Hertel[2], Lars Hagenäs[1,3]

[1] Department of Women's and Children's Health, Karolinska Institutet, [2] Odense University Hospital, [3] Pediatric Endocrinology Unit, Astrid Lindgrens Children Hospital

Problem when using normal population reference

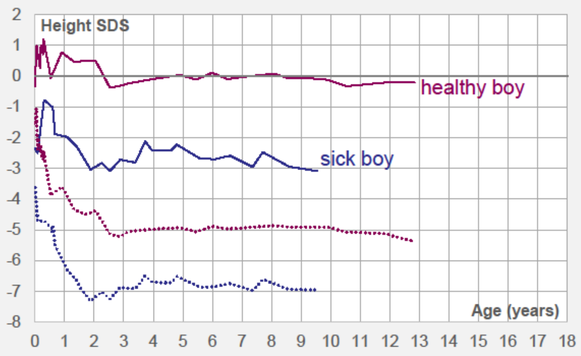


Conclusion

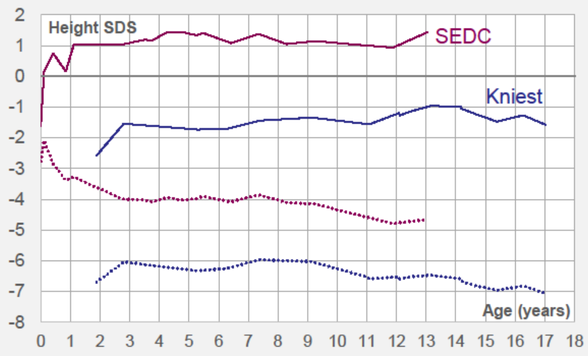
The presented examples show clearly the usability of a short stature reference as well as of expressing growth patterns in SDS format when following children with severe growth retardation.
Since many extreme short stature conditions are rare, using a standard height matrix would help in gaining knowledge on syndrome-specific growth patterns, in terms of both magnitude and tempo.
Especially pubertal growth component is often not known.

Height SDS expressed both relative to WHO (dotted line) & Achondroplasia reference (full line)

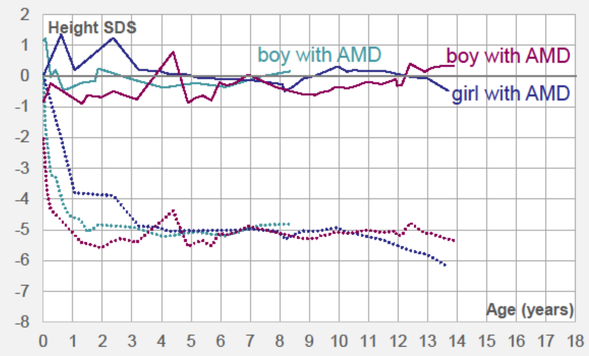
2 boys with achondroplasia



2 girls with COL2 mutation

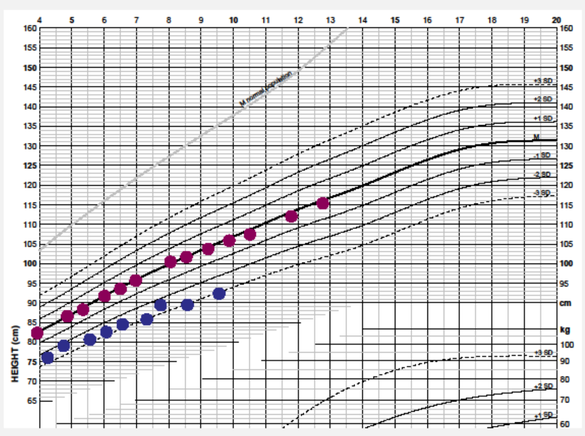


3 children with acromesomelic dysplasia Maroteaux

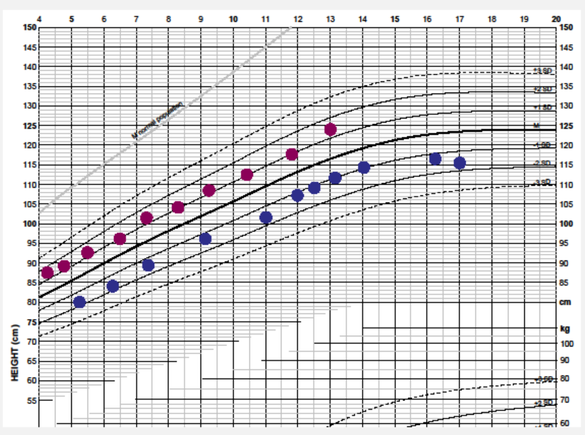


Same examples in achondroplasia chart, from 4 years

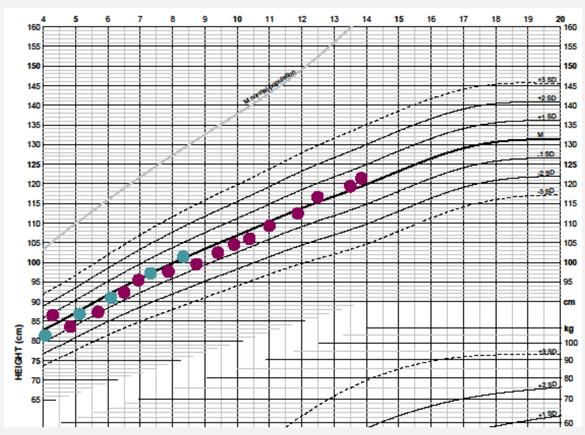
both boys with achondroplasia



both girls with COL2 mutation



both boys with acromesomelic dysplasia Maroteaux



Introduction

There is a shortage of clinically useful growth charts for following growth development in conditions with extreme short stature. At the same time, it is not possible to construct syndrome-specific growth charts for many of these conditions due to low prevalence and perhaps also often a great inter-individual variability of the growth defect within a certain syndrome.

Aim

The aim of this project was to evaluate growth patterns of children with severe growth retardation using our newly constructed achondroplasia reference as a short stature reference.

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Method

Longitudinal height measurements from about 100 children and adolescents with defined skeletal dysplasia (here illustrated: achondroplasia, SEDC and Kniest syndrome as well as acromesomelic dysplasia) were expressed in standard deviation scores (SDS) both relative to the WHO and the achondroplasia (Neumeyer et al., unpublished) reference.

Results

Height in several syndromes followed within the normal range (±2 SD) of the used short stature standard.

Height development in acromesomelic dysplasia Maroteaux for instance was almost identical to the achondroplasia mean.

Expressing height development in a SDS curve format is an easily understandable way to illustrate growth patterns.

Karolinska Institutet
Andrea Merker
PhD student • Department of Women's and Children's Health
171 76 Stockholm
E-mail: andrea.merker@ki.se

Karolinska Institutet

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