CORRESPONDENCE

Anthropometric evaluation of children with *SHOX* mutations can be used as indication for genetic studies in children of short stature

In their recent article, Rappold *et al*[1] investigated the presence of *SHOX* defects in a large cohort of 1608 children of short stature. Of the total number of *SHOX* mutations/deletions identified, 58% were found in 55 children with Leri–Weill dyschondrosteosis (LWD) and 2.2% in 1534 cases considered to have idiopathic short stature. The authors created an evidence-based scoring system based on the clinical features of 68 patients with *SHOX* defects to identify the most appropriate children for *SHOX* gene testing. The following criteria were used: arm span:height ratio <96.5%, sitting height:height ratio >55.5%, body mass index >50th centile and the presence of cubitus valgus, short forearm, bowing of the forearm, appearance of muscular hypertrophy and/or dislocation of the ulna. This scoring system had some limitations, such as a low positive predictive value (11%) when using the lower cutoff (score of 4) and a lower sensitivity (61%) when using the upper score (score of 7, out of a maximum of 24).

To select those likely to have mutations in the *SHOX* gene from a population of children with short stature, previous studies have already suggested a limbs:trunk ratio, ((calculated subischial leg length + arm span)/sitting height)[2] and sitting height:height ratio (SH:H), expressed as standard deviation score (SDS) for age and sex.[3] Rappold *et al.*[1] analyzed the SH:H ratio as absolute values, even though their cohort represented a wide age range, and age is known to strongly influence this ratio.[4]

It would be useful if Rappold *et al.* were to report the limbs:trunk ratio proposed by Binder *et al.*[2] and SH:H ratio expressed as SDS[4] in this large cohort of patients with *SHOX* mutations. These parameters could also improve the proposed score system.

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References

1 **Rappold G**, Blum WF, Shavrikova EP, Crowe BJ, Roeth R, Quigley CA, Ross JL, Niesler B. Genotypes and phenotypes in children with short stature: clinical indicators of SHOX haploinsufficiency. *J Med Genet* 2007;**44**:306–13.
2 **Binder G**, Ranke MB, Martin DD. Auxology is a valuable instrument for the clinical diagnosis of SHOX haploinsufficiency in school-age children with unexplained short stature. *J Clin Endocrinol Metab* 2003;**88**:4891–6.
3 **Jorge AA**, Souza SC, Nishi MY, Billerbeck AE, Liborio DC, Kim CA, Arnhold IJ, Mendonca BB. SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. *Clin Endocrinol (Oxf)* 2007;**66**:130–5.
4 **Gerver WJM**, Bruin R. Paediatric morphometrics. *A reference manual*. 2nd edn. Universitaire Pers Maastricht, Maastricht, 2001.