Computer-assisted diagnosis of dyschondrosteosis based on skeletal X-ray geometry.

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Background
Bone X-rays provide the main diagnostic parameters for chondrodysplasia, including common dyschondrosteosis (DC). Skeleton is usually studied piece by piece by visual analysis in search of characteristic signs. The phenotypic spectrum of DC is large. Indeed, children who have seemingly idiopathic short stature (ISS) may have subtle forms of DC that can be unrecognized.

Objective and hypothesis
Provide a user-friendly computer-assisted program that facilitates the identification of subtle forms of DC within a population of children with ISS

Patients and Methods
The program was trained on age-matched 10 patients with typical DC (SHOX mutations), and 10 patients with a diagnosis of ISS. Thereafter, it was tested on 54 patients with ISS in whom skeletal X-rays were considered normal by visual inspection.

84 points were placed on various key points of 6 radiographic images (ilia, ischia, lumbar spine, forearm, hand, leg), giving 39 measures. Angles and distances were drawn between points of the same skeletal piece and precisely quantified. Angles and distances from different regions were modelled.

Results
The more specific and more sensitive parameters for the diagnosis of DC seemed to be the largest distance measured between radius and ulna, carpal angle, length of 4th metacarpal and its relationship with length of 4th proximal and distal phalanx. Interpeduncular distance of lumbar spine. Our computerized program was able to detect 21 children with subtle forms of DC among the 54 children considered to have ISS.

Conclusions
In field conditions, it may be useful to paediatricians to identify subtle forms of DC among children with ISS. Our computerized quantitative analysis of skeletal morphology may be helpful in this respect.

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