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.

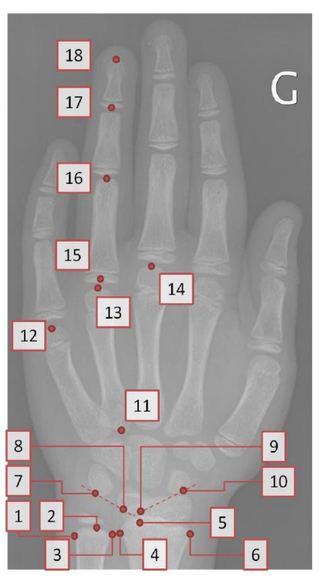
Forearm (front)

1. Lateral edge of the ulna at the largest distance between radius and ulna
2. Medial edge of the ulna at the largest distance between radius and ulna
3. Medial edge of the radius at the largest distance between radius and ulna
4. Lateral edge of the radius at the largest distance between radius and ulna

Larger distance measured between Radius and ulna

Hand (front)

1. Lateralmost point of the ulnar metaphysis
2. Distalmost point of the ulnar metaphysis
3. Medialmost point of the ulnar metaphysis
4. Medialmost point of the radial metaphysis
5. Distalmost point of the radial metaphysis
6. Lateralmost point of the radial metaphysis
7. Proximal edge of the triquetral bone (on the tangent to the proximal edges of the triquetral and the lunate bones)
8. Proximal edge of the lunate bone (on the tangent to the proximal edges of the triquetral and the lunate bones)
9. Proximal edge of the lunate bone (on the tangent to the proximal edges of the lunate and the scaphoid bones)
10. Proximal edge of the scaphoid bone (on the tangent to the proximal edges of the lunate and the scaphoid bones)
11. Proximal extremity of the 4th metacarpal
12. Distal extremity of the 5th metacarpal (including the head)
13. Distal extremity of the 4th metacarpal (including the head)
14. Distal extremity of the 3rd metacarpal (including the head)
15. Proximal extremity of the 4th proximal phalanx (including the epiphysis)
16. Distal extremity of the 4th proximal phalanx
17. Proximal extremity of the 4th distal phalanx (including the epiphysis)
18. Distal extremity of the 4th distal phalanx

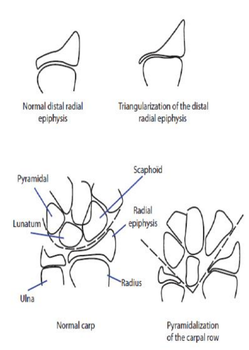


M4 < Ph1+Ph3

(N. M4 = Ph1+Ph3 ± 2 mm)

Carpal angle:

(N. 131,5° ± 14,4°)

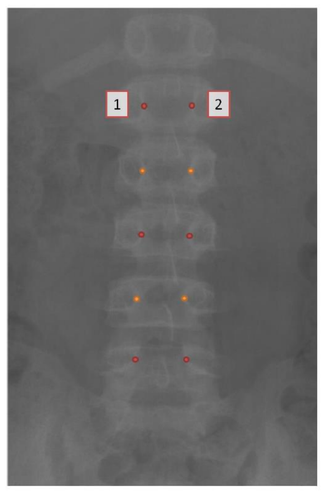


Carpal angle and lenght of 4th metarcarpal.
Relationship with lenght of 4th proximal and distal phalanx

Lumbar rachis (front)

1. L1: medialmost point of the right pedicle
2. L1: medialmost point of the left pedicle

Interpeduncular distance of lumbar spine



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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