Pseudoachondroplasia

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Introduction

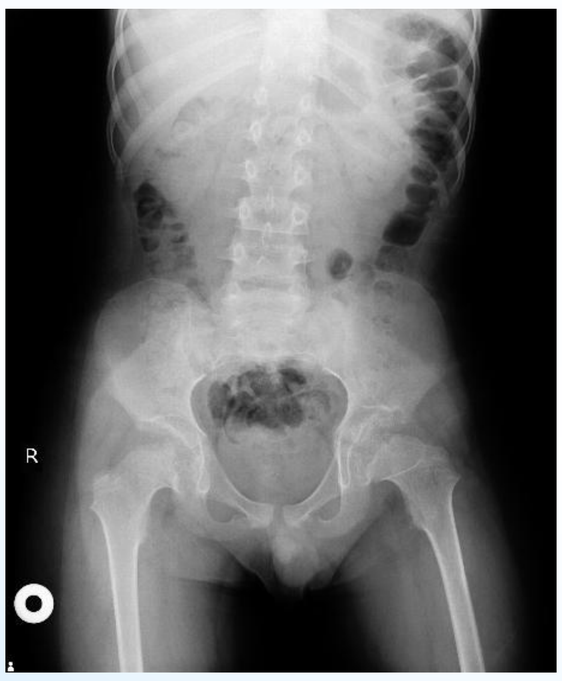
Pseudoacondroplasia is a short extremity dwarfism characterized by lifelong arthralgia and early onset osteoarthritis. At birth there is a normal height and face appearance. At the beginning of walking a swaying walking nature are signs that can be seen. Typically, a two-year old's short height becomes apparent and leads to a disproportionate short-limb appearance. In childhood, joint pain in the broad joints especially in the lower extremities is common. Degenerative joint disease is progressive.

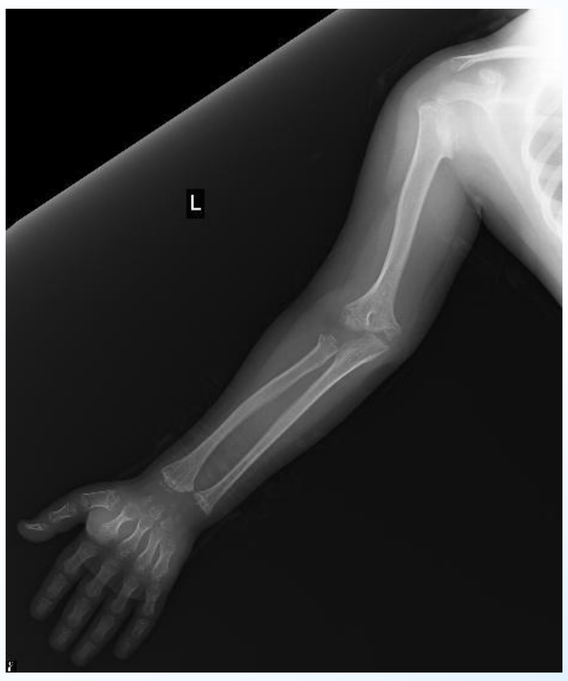
Case

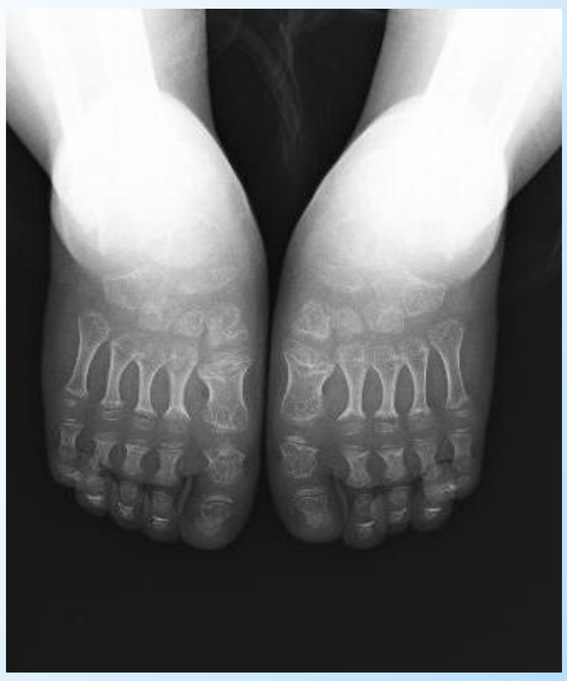
Ten-year and six-month old male patient admitted to our clinic due to short stature , his knee and hip pain Before admission ,he was diagnosed with achondroplasia and given growth hormone treatment, it was learned that after 3 months of growth hormone treatment, it was stopped because of Pertes disease. in his natal history he was a 36-week, 2800 gram born. The mother of patient showed similar characteristics. There were no other short stature history in the family. His chronological age: 10 years 6 months, height age: 5years 3months, bone age: 10 years, weight: 27.9kg(10p), height:102 cm (<3 p ; -4.65 SD), at physical examination, axilla(-), pubis stage 2, testis volume: 3/3ml. Upper arm: 21.5cm, forearm: 24.5cm, head-pubis: 61cm, pubis-heel : 50.8cm were measured. Brachydactyly, rhizomelia, scoliosis, sway walking , genu varum and lumbar lordosis were present. Previously performed FGFR3 gene analysis revealed normal. On the bone radiography of the patient, irregular epiphysis and metaphysis of long bones, delayed epiphyseal ossification, smallness on femoral head epiphysis, brachydactyly, short metacarpal bones, irregular carpal bones were detected and diagnosed as pseudoachondroplasia. The result of the COMP gene analysis is awaiting.

Conclusion

The diagnosis of Pseudoachondroplasia due to mutations in the cartilage oligomeric matrix protein (COMP), which is autosomal dominant, can be made based on clinical findings and radiological features. The appearance of normal craniofacial appearance, joint hyperlaxity and characteristic radiological findings is separated from achondroplasia. Pseudoacondroplasia should be kept in mind in cases with achondroplasia but with joint aches and hyperreactivity.







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