

Abstract

Achondroplasia (ACH) is the most common form of skeletal dysplasia characterized by a rhizomelic short stature. Radiological skeletal findings in pediatric and adult patients with ACH include short long bones, a relatively longer fibula compared to the tibia, a narrow lumbar interpedicular distance, and a hypoplastic iliac wing. Nonetheless, the characteristics of skeletal growth during the neonatal and infantile periods have scarcely been explored. Therefore, this retrospective study aimed to analyze the radiological skeletal growth during the neonatal and infantile periods in 41 Japanese patients with genetically confirmed ACH. The length of long bones in the upper and lower limbs and the lumbar interpedicular distances at L1 and L4 were measured. These parameters showed significant positive correlations with age. The upper segment-to-lower segment ratio in the lower limbs resembled the data of healthy controls from previous reports. The L1/L4 and fibula/tibia ratios increased with age, suggesting that some representative skeletal phenotypes of ACH were less distinct during the neonatal and infantile periods. In conclusion, for the first time, this study radiologically characterized skeletal growth during the neonatal and infantile periods of patients with genetically confirmed ACH.