

Abstract

Introduction: Achondroplasia is the most common bone dysplasia. It is caused by mutations in the FGFR3 gene, which is involved in regulation of proliferation and maturation of chondrocytes on the growth plates. Activating mutation results in impaired endochondral ossification and a wide range of symptoms: severe growth disorder with limb shortening, macrocephaly with risk of hydrocephalus, mid-facial dysplasia, sleep apnea, narrowing of the spinal canal, increased risk of respiratory complications, and airway inflammation. The current rapid technological development has led to a better understanding of the processes of epiphyseal growth cartilage, thus enabling the development of new drugs for the treatment of this disorder (modified CNP, soluble FGFR3, meclozine). Nevertheless, current treatment is primarily symptomatic. It is very important to have a growth patterns of patients with achondroplasia who have not been affected by any growth promoting treatment (prolongation surgery, growth hormone).

Objectives: The aim of this study is to evaluate the growth of Czech patients with achondroplasia, to compare our data with the world-wide used data by Horton et al. (1978) and the current data by del Pino et al. (2018). The next aim was to verify the accuracy of the multiplier method of final height prediction by Paley et al. (2005) in Czech patients with achondroplasia.

Material and Methods: Our group consists of 79 patients (49 boys, 30 girls) who were examined longitudinally at the Department of the Paediatric Clinic of Motol University Hospital and in the Ambulant centre for defects of locomotor apparatus and 7 patients (3 males, 4 females) who were measured only in adulthood. Data of 22 patients after growth promoting therapy were not included in the study. Only 9 patients (5 boys, 4 girls) had complete data from childhood to adulthood.

Mean values corresponding to Horton's age categories were obtained using the model of linear regression. Comparison with the Horton study was performed by using the confidence intervals.

Results: Growth curves of Czech boys do not differ significantly from Horton et al. (1978). In girls over 11 years of age, body height is significantly lower than in Horton's study. A similar trend is also indicated in boys. Differences in final height of adult patients are not significant. Significant differences were not found when comparing the upper and lower segment. Significant differences were not found between our and Argentine study by del Pino et al.

(2018). Between the final height and predicted body height by the multiplier method by Paley et al. (2005), we found clinically significant differences in most patients.

Conclusion: Growth data of Czech patients with achondroplasia are comparable to foreign, more representative studies. We prefer a graphical method of predicting the final height that takes into account the patient's entire growth curve, its trend, and the patient's clinical condition. The prediction of the lower limbs by the multiplication method can be used with correction to bone age and sexual maturation.

Keywords: achondroplasia, dwarfism, proportionality, final height prediction