Introduction

Arthrogryposis Multiplex Congenita type II is autosomal dominant, characterized by multiple congenital contractures in the limbs without a primary neurological deficit. The most frequently observed clinical features are triangular face, palpebral fissures facing downwards, clarity in nasolabial folds, small mouth, high palate, adherent ear lobes, short stature, camptodactyly, ulnar deviations in the fingers, vertical talus and/or talipes equinovarus.

Case

A male patient who applied to the hospital complaint of his short stature was born with term 3000 gr and 50 cm. At the time of birth it was learnt that the arm, left leg, hand and neck had a curvature and limitation of movement and was diagnosed with Arthrogryposis Multiplex Congenita. In his physical examination 5 years 10 months, stature age: 2 years 8 months, bone age: 3 years 6 months, weight:12.7kg(<3p), height: 92.1cm(-4.49 SD) and prepubertal. In existence were triangular face, epicantus, micrognati, facial asymmetry, clearness in nasolabial folds, narrow mouth opening, high palate, long filtrum, ulnar deviation in fingers, abduction limitations in thumbs, tarsal fusion, short neck, asymmetry in shoulders and scoliosis. Hand, shoulder, elbow and neck joints were limited due to contractures. In his laboratory TSH: 2.4 mIU/ml, sT4: 0.55 ng/dl, IGF-1: <15 ng/ml (<-3SD),IGFBP3: 489 ng/ml (<-3SD) was observed. LT4 therapy was initiated for his central hypothyroidism after ACTH: 18.3 pg/ml, cortisol: 8.3ug/dl. The rate of elongation in 3 months after euthyroidism was 0.5 cm and IGF-1: <15 ng/ml (<-3SD) and IGFBP3: 570 ng/ml (<-3 SD). In the growth hormone stimulation tests results were in L-DOPA peak 0.66, in clonidine peak 0.84. The growth hormone treatment that was started on the patient and a 3cm prolongation in 3months was observed. The genetic analysis results is awaiting.

Conclusion

Half of the Arthrogryposis Multiplex Type II cases are caused by mutation in troponin I(TNN12), troponin T (TNNT3) and embryonic myosin (MYH3) genes. In spite of short stature of syndrome features, only 2 cases of Panhypopituitarism have previously been reported in available literature.