Abstract

BACKGROUND: Marfan syndrome (MS) is an autosomal dominant condition of the connective tissue that involves the ocular, cardiovascular and musculoskeletal systems. MS is caused by mutations in the fibrillin-1 gene, leading to joint ligaments flaccidity, joint hypermobility and an overgrowth of the long bones. OBJECTIVES: The aim of the present study was to assess anthropometry, musculoskeletal alterations and the prevalence of physical therapy treatments among patients with MS. METHODS: Twenty-six patients were included in this study [17 females (age: 13.23±2.77 years; body mass 51.5±24-68 Kg; height 1.70±1.40-1.81 m; arm span: 1.73±0.12 m) and 9 males (age: 14.44±2.18; body mass: 61.0±42-72 Kg; height: 1.83±1.66-1.97 m; arm span: 1.93±0.13 m)]. Anthropometric measurements and musculoskeletal abnormalities were determined in a standardized fashion: pectus and scoliosis were assessed through radiography and angulation (ángulo) of the scoliosis curve using the Cobb method; arachnodactyly was assessed through the thumb sign and Walker-Murdoch test and dolichostenomelia was assessed by arm span in relation to height. Patients also responded to a questionnaire addressing participation in physical therapy. RESULTS: In comparison to values estimated for the Brazilian population, mass and height were greater among the patients with MS (females: p=0.001 e p<0.0005 e males p=0.019 e p=0.0001, respectively). The following musculoskeletal abnormalities were found: pectus in 3 patients (11%), pectus and scoliosis in 19 (73%), dolichostenomelia in 11 (42%) and arachnodactyly in 21 (80%). Eleven patients (42%) with MS had previously undergone physical therapy. CONCLUSIONS: Patients with MS exhibit altered musculoskeletal and anthropometry and have infrequent physical therapy treatment.

Keywords
Marfan Syndrome, anthropometry, musculoskeletal system, connective tissue, physical therapy.