

Saila Laakso^a, Sanna Toiviainen-Salo^a, Heli Viljakainen^a, Jaakko Perheentupa^a, Outi Mäkitie^{a, b}

^aUniversity Hospital and University of Helsinki, Helsinki, Finland; ^bFolkhälsan Research Center, Helsinki, Finland

saila.laakso@helsinki.fi

BACKGROUND

Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED or APS1) is an autosomal recessive disorder characterized by chronic candidiasis and autoimmune destruction of endocrine organs. Hypoparathyroidism, adrenocortical failure and hypogonadism are the most common endocrinopathies, which together with their treatment may impact bone health. However, very little is known about the long-term skeletal health in patients with APECED.

METHODS

The cross-sectional cohort consisted of 29 Finnish patients (21 females) who were examined clinically, their bone mineral densities (BMD) were measured with dual-energy x-ray absorptiometry (Lunar Prodigy Advance DXA) and radiographic abnormalities were evaluated with spinal x-ray imaging. Fracture history was collected with questionnaire.

RESULTS

CHARACTERISTICS AND FRACTURE HISTORY

The patients' mean age was 35 years (range 12-71, 3 females younger than 18 yrs, Table 1). 26 patients had hypoparathyroidism, 26 patients had adrenocortical failure and 17 patients had hypogonadism. Altogether 16 (55%) patients had had 1-5 fractures and seven of them had had low-impact fractures. Spinal compression abnormalities were evident in four (14%) patients. Ten (34%) patients had scoliosis.

Characteristic Mean (range)	29 patients with APECED	7 patients with low- impact fracture history	4 patients with vertebral abnormalities
Age (yrs)	35 (12-71)	43 (31-51)	44 (31-63)
Sex (female/male)	21/8	5/2	4/0
Hypoparathyroidism, n (%)	26 (90%)	7 (100%)	4 (100%)
Adrenocortical failure, n (%)	26 (90%)	7 (100%)	4 (100%)
Hypogonadism, n (%)	17 (59%)	6 (86%)	4 (100%)

TABLE 1. Characteristics and fracture history of 29 Finnish patients with APECED.

OBJECTIVE AND HYPOTHESIS

Aim of the study was to determine the prevalence of, and risk factors for, osteopenia and osteoporosis in children and adults with APECED.

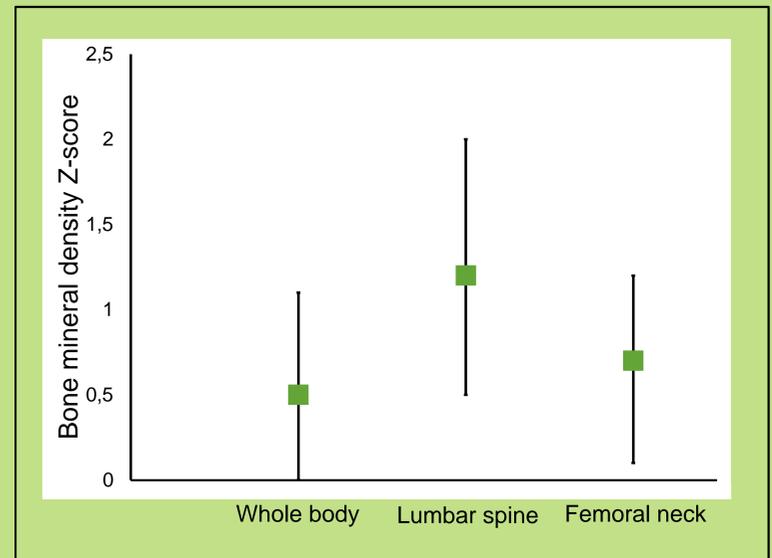
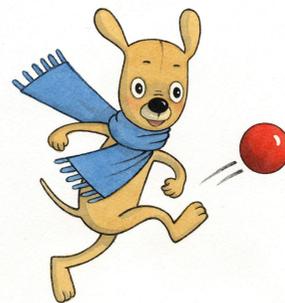


FIGURE 1. Bone mineral density Z-scores for whole body, lumbar spine, and femoral neck in 29 patients with APECED. Values are expressed as means with 95% confidence intervals.

BONE MINERAL DENSITY

The mean BMD Z-scores (95% CI) were +0.5 (-0.0 – +1.1) for whole body, +1.2 (+0.5 – +2.0) for lumbar spine, and +0.7 (+0.1 – +1.2) for femoral neck. Osteopenia was present in 20%, but none of the patients had osteoporosis.



CONCLUSIONS

Patients with APECED are not at high risk for low BMD, but based on fracture prevalence their bone structure may be altered and warrants further studies.

DISCLOSURE STATEMENT

The authors have nothing to disclose.

