Objective. To evaluate osteocyte-secreted proteins and molecular-genetic markers in relation to bone mineral density in women with estrogen deficit.

Materials and Methods. 110 women with amenorrhea and 236 postmenopausal women living in Russian Federation enrolled in the cross-sectional study. Levels of reproductive hormones, osteocyte-secreted proteins were determined, as well as DXA. SNPs were genotyped by PCR for TNFSF11 (RANKL) (rs9594738, rs9594759), TNFRSF11B (OPG) (rs3102735), SOST (rs1107748), LEPR (rs1805094, rs8179183).

Results. Low bone mineral density (BMD) (Z-score <= -2.0) in L1-L4 was found in 33.6% of women with amenorrhea (23.2% in POI, 47.5% in hypogonadotropic amenorrhea, 45.5% in gonadal dysgenesis, 46 XX); in femoral neck - in 8.9%. Sclerostin (Scl) in women with amenorrhea was lower than in postmenopausal women (p=0.02), osteoprotegerin (Opg) in POI was higher in normal vs. low BMD (p=0.02), RANKL in gonadal dysgenesis was lower in normal vs. low BMD (p=0.04). There was a moderate positive correlation of Scl to the age of beginning of amenorrhea (p=0.04) and a moderate negative correlation of Opg to the duration of amenorrhea (p=0.01). In postmenopausal women (p<0.05): T/T+T/C genotypes in RANKL (rs9594738, rs9594759) were associated with 2-fold increase in L1-L4 osteoporosis risk; C/C genotype in OPG (rs3102735) - with 17-fold increase in distal radius fracture risk. In women with amenorrhea (p<0.05): T/T genotype in SOST (rs1107748) - with 3-fold increase in risk of low BMD in femoral neck; C/C genotype in LEPR (rs1805094, rs8179183) - with 3-fold increase in risk of low BMD in L1-L4.

Conclusion. The age of beginning of estrogen deficit and its duration influence the concentration of osteocyte-secreted proteins and BMD. Several SNPs of genes coding these proteins have shown to influence risks of osteoporosis and fracture in different skeletal sites.