

the index patient (ages 6, 12 and 14 years) were heterozygous for all 3 mutations and had also bone disease (either fractures, low BMD, or both). The father (41 years old) was homozygous for the COL-1A1 mutation and heterozygous for the CRTAP and LRP5 mutation. He had already several osteoporotic vertebral fractures associated with very low BMD (T-Score -4). The mother was not available for examination. One aunt (56 years) had no mutations and no bone disease, and another aunt (60 years) had only the COL-1A1 mutation and osteoporosis with vertebral fractures. The grandmother (paternal) had also osteoporosis and was heterozygous for the COL-1A1 and the CRTAP mutations.

**Conclusion:** The described mutations in the COL-1A1 and LRP5 genes are associated with decreased BMD, but do not fully explain the juvenile osteoporosis. It appears likely, that the new variant c.540G>A: G/A (p.Met180Ile) in the CRTAP gene is associated with juvenile osteoporosis or with a mild form of OI.

#### P1179

##### OSTEOPROTEGERIN AND BMD IN POSTMENOPAUSAL WOMEN WITH PRIMARY HYPERPARATHYROIDISM

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**Objective:** to study relationship between osteoprotegerin (OPG) and BMD in postmenopausal women with primary hyperparathyroidism (PHPT) compared postmenopausal women without PHPT.

**Methods:** We studied 30 postmenopausal women with PHPT, average age 62.5±6.12 years. The control group were 16 postmenopausal women without PHPT, mean age 59.7±6.28 years in physiological menopause. Examination: total calcium, phosphorus, albumin, creatinine, PTG, OPG, vitamin D, BMD measurements in the lumbar spine, femoral neck, total hip, and distal 1/3 radius by DXA.

**Results:** There were no differences in the age, years (U=138.5, p=0.052), height, m (U=216.5, p=0.99), weight, kg (U=202.5, p=0.72), BMI kg/m<sup>2</sup> (U=196.5, p=0.61), duration of menopause in both group, years (U=146.5, p=0.08). Osteoporosis of axial skeleton was founded in 43%, osteopenia in 33% in postmenopausal women with PHPT (T-score<sub>L1-L4</sub>=-2.1 (-2.9 - -0.9), T-score<sub>Right femur total</sub>=-1.1 (-1.7 - -0.2)), and osteoporosis was not founded in the control group, osteopenia in 12.5% (T-score<sub>L1-L4</sub>=-0.1 (-0.8 - 0.7), T-score<sub>Right femur total</sub>=-0.1 (-0.5 - 1)). Significant differences was detected in the BMD in postmenopausal women with PHPT compared postmenopausal women without PHPT (U<sub>L1-L4</sub>=57.0, p=0.0001; U<sub>Right femur total</sub>=88.5, p=0.0015). We detected correlation between OPG level and axial skeleton BMD: r<sub>s L1-L4</sub>=0.5, p<0.05, r<sub>s Right femur total</sub>=0.5, p<0.05.

**Conclusion:** The results of the study confirmed increased risk of osteoporosis in postmenopausal women with PHPT compared control group. The results may indicate association between OPG level and axial skeleton BMD.

#### P1180

##### OTHER ENTHESITIS THAN ACHILLES' IN PSORIATIC ARTHRITIS: AN ULTRASONOGRAPHIC STUDY

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**Objective:** Enthesal involvement is a frequent and distinctive feature of psoriatic arthritis (PsA), frequently underdiagnosed using only clinical tools. Over the last decade, ultrasonography (US) has been increasingly used in rheumatology, for assessing articular and periarticular involvement in patients with chronic arthritis and is lately gaining favor in the evaluation of enthesitis in patients with spondyloarthritis. Subclinical enthesitis detected using MSUS, particularly with a power Doppler signal inside, has proved to be of a predictive value for the development of structural damage in patients with psoriatic arthritis. The aim of this study is to find and to describe sonographic structural and inflammatory changes in other entheses than of Achilles in patients recently being diagnosed with psoriatic arthritis (PsA), patients with established PsA, according to currently used criteria and patients with other non-inflammatory chronic diseases.

**Methods:** The study included 10 patients with early PsA (ePsA), 30 with established PsA (PsA) (according to CASPAR criteria) and 20 patients with no inflammatory joint disease, as the control group were enrolled in the study. All patients underwent, besides clinical and paraclinical evaluation, an US examination of the following enthesal sites: common extensor and flexor tendon at the insertion at the lateral and medial humeral epicondyle; proximal and distal patellar tendon entheses.

**Results:** There was a rather poor relationship between clinical based enthesitis involvement and US findings, including loss of normal fibrillar echogenicity, hypoechoic swelling of the tendon insertion, effusions, bursitis, and increased blood flow detectable with a PD signal. Loss of fibrillar echogenicity was more frequent in the ePsA group, when compared to control group (p<0.01), and the presence of enthesophytes at the level of the elbow entheses, was more frequently identified in the PsA group, in comparison to ePsA and control group (p<0.05; p<0.01). Doppler signal showed high prevalence in the psoriatic patients, compared to controls, but with no difference in between psoriatic groups.

**Conclusion:** Structural ultrasound changes and PD in entheses are common in both new and established PsA. The present study confirms that US allows detecting structural and inflammatory abnormalities of entheses in PsA patients. Only the clinical examination is not enough for a certain diagnosis, precisely in the early stages.

#### P1181

##### MINERAL DENSITY OF BONE IN CHILDREN WITH DISPLASIA SYNDROME OF CONNECTIVE TISSUE

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**Objective:** To assess the state of mineral bone density (MBD) during connective tissue dysplasia (CTD) in children living in the city of Semey. We studied the condition of MBD in 84 children aged 1 to 14 years (mean age - 7.5±0.6 years), including 46 boys (54.8%) and 38 (45.2%) - girls. The control group consisted of 80 children without signs of CTD (mean age - 7.2±0.5 years), including 46 boys (57.1%) and 34 girls (42.9%). In the studied groups of children, there were no fractures in the anamnesis.

**Methods:** All children were assessed the blood content of total calcium, magnesium, phosphorus (reagents BioSystems S.A. [QSC according to ENISO 13485 and ENISO 9001 standards] were used). BMD was examined on the SONOST-2000 apparatus (OsteoSys Co., Ltd, Korean Republic). The BMD is compared with reference values from healthy youth of the same age, sex, and race/ethnicity to calculate a z-score, the number of SDs from the expected mean. At z-score, <-2.0 SD was considered as a state of "bone density reduction for a given age" (z-score that is > 2 SDs below expected (<-2.0) is referred to as "low for age").

**Results:** In children with CTD, the microelements in the blood were lower than in the control group and below normal. So, Ca (n 2.15-2.58 mmol/l) in children with CTD was 2.11±0.12 and in the control group - 2.43±0.10 (p=0.05). Mg (n 0.7-0.98 mmol/l) in children with CTD - 0.63±0.02 and 0.77±0.03, respectively (p<0.05). The content of P (n 1.29-2.25

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