Evaluation of Bone Microarchitecture with High Resolution-pQCT in Patients with Thalassemia

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Thalassemia is a genetic disorder of hemoglobin synthesis. Due to improved blood transfusion and chelation therapy, survival has been increased with the consequence of complications like osteoporosis not seen during childhood and adolescence. The obvious shortcomings of conventional BMD methods like dual energy x-ray absorptiometry (DXA), can be overcome by simultaneously assessing the microarchitecture of the bone using high-resolution peripheral quantitative computed tomography (HR-pQCT), which may improve the estimation of the fracture risk in patients with thalassemia. In 17 regularly transfused patients (age: 13 - 43 y, 9/17 female) with beta-thalassemia major (n = 10), -intermedia (n = 6), and CDA-II (n = 1), the BMD of lumbar spine (LS) and total hip was measured by DXA (Hologic QDR1000+, Bedford, USA). Age, gender and ethnic specific BMD Z-scores were calculated. In addition, we assessed the volumetric BMD and the trabecular architecture of the non-dominant distal radius and tibia by HR-pQCT (XtremeCT®, SCANCO Medical AG, Bassersdorf, Schweiz). Liver iron concentration and endocrinological parameters were also determined. In 15/17 patients low BMD values (LS Z-score range: -1.1 to -3.1) measured by DXA were significantly correlated with total volumetric density (range: 91 - 388 mg/cm³, p = 0.002) measured by HR-pQCT at the distal radius. In 6/17 patients (> 28 y), all with latent hypogonadism, the spongiosa was porous or nearly dissolved. Patients with hypogonadism (n = 9) were significantly different from normals with respect to radial trabecular inhomogeneity parameter TbSp SD (p = 0.02), but not to LS Z-score. Patients with fractures (n = 5) had lower total densities (p = 0.02) and trabecular TbSp SD (p = 0.02) at the tibia and started blood transfusions at a higher age (p = 0.023). However, Z-scores did not reflect the fracture risk in this patient group (p = 0.11). Liver iron was mainly correlated with tibial TbSp SD (Rs = 0.54, p = 0.025).

In patients with thalassemia BMD Z-scores seem to underestimate fracture risk because a normal cortical thickness and density may conceal a porous trabecular structure. Endocrinological failures, especially hypogonadism, were responsible for the pathological microarchitecture of distal radius and tibia, while bone marrow expansion as in thalassemia intermedia and liver iron concentration seem to play a minor role. These initial results from bone microarchitecture measurements in thalassemia should be confirmed in a larger sample of patients with greater age range.