GENETICS AND GENODERMATOSES

AN ANALYSIS OF OSTEOPOROSIS AND OSTEOPENIA IN EPIDERMOLYSIS BULLOSA

M Kang [1] - J Chen [1] - M Radjenovic [2] - A Yang [2] - G Feng [3] - D Murrell [2]

University Of New South Wales, Faculty Of Medicine, Sydney, Australia [1] - St George Hospital, Dermatology, Sydney, Australia [2] - University Of Sydney, School Of Mathematics And Statistics, Sydney, Australia [3]

Background: Epidermolysis Bullosa (EB) patients have multiple risk factors of osteoporosis like decreased physical activity and malnutrition. Presently, there is limited literature describing the prevalence of osteopenia or osteoporosis in EB, particularly in the adult cohort and in the less severe EB types, namely EB Simplex (EBS).

Objective: To describe the occurrence of osteopenia or osteoporosis in EB patients from the Australasian Epidermolysis Bullosa Registry (AEBR)

Materials and Methods: 72 patients underwent a dual energy X-ray absorptiometry scan. Patients' bone mineral density (BMD) T-and Z-scores, EB Disease Activity and Scarring Index (EBDASI), Quality Of Life in EB (QOLEB) scores were obtained.

Results: T-scores of Recessive Dystrophic EB (RDEB) and Junctional EB (JEB) patients were significantly lower than the osteoporosis and osteopenia diagnostic T-scores of -2.5 and -1, respectively. Z-scores of RDEB patients were significantly lower than the normal value of -2. EBDASI and QOLEB were inversely correlated with Z-scores. The prevalence of osteoporosis in adults with RDEB and JEB were 75% with the rest having osteopenia. The prevalence of osteopenia in adults with EBS and dominant dystrophic EB were 50% and 33%, respectively, with the rest having normal diagnoses.

Conclusions: This is the largest and the first study of cohort of adult patients with milder forms of EB (EBS and DDEB). Although the T-scores in these groups were not statistically lower than the osteopenia diagnostic T-score, high prevalence of osteopenia was observed. Patients should be assessed with holistic disease severity tools like QOLEB and EBDASI. Patients with high EBDASI (Activity≥20, Damage≥54, total≥75) and QOLEB (≥18) scores should receive early screening and prophylaxis irrespective of EB type. Although the above cut-off scores can be used as guidance, as osteoporosis occurs in a continuous spectrum, despite the dogmatic cut-off values used for its diagnosis, individualised patient fracture risk should be concurrently evaluated.