

The Influence of Mobility on Bone Status in Subjects with Rett Syndrome: a 10-Year Longitudinal Study

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Low bone mass is a frequent complication of subjects with Rett syndrome. This study aimed to investigate the long-term influences of mobility on bone status in girls with Rett syndrome

In 47 girls with Rett syndrome, serum calcium, bone alkaline phosphatase, 25-hydroxyvitamin D and quantitative ultrasound (QUS) parameters at phalanxes by Bone Profiler-IGEA (amplitude dependent speed of sound: AD-SoS and bone transmission time: BTT) were measured at baseline and after 5 and 10 years. The subjects were divided into two groups: non ambulatory (n= 22) and ambulatory (n= 25).

At baseline both AD-SoS and BTT values were lower in non ambulatory with respect to ambulatory subjects, but the difference was not statistically significant. Non ambulatory subjects presented a significantly ($p<0.05$) later onset of age at menarche and lower birth weight with respect to the ambulatory subjects. BMI was significantly lower in non ambulatory subjects than in ambulatory subjects at each time point. At the 5-year follow up both ambulatory and non ambulatory Rett subjects presented a similar reduction in both AD-SoS and BTT. Also at 10-year follow up both non ambulatory and ambulatory subjects showed a significant reduction in AD-SoS (-4.7% $p<0.05$; and -3.4 % $p=n.s.$ respectively) and in BTT (-54% $p<0.05$; and -41% $p= 0.05$, respectively) with respect to baseline.

In conclusion this longitudinal study suggests that QUS parameters are markedly decreased in non ambulatory subjects and that nutritional status play a key role in the progressive deterioration of bone status.