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Endocrine Alterations in Adults with 22q11.2DS

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Background: Endocrine disorders, including hypoparathyroidism, hypocalcaemia, hypomagnesemia and hypothyroidism, are more common in 22q11.2 deletion syndrome (22q11.2DS) than in the general population. However, there are few systematic studies on endocrine alterations in 22q11.2DS involving adults and/or a healthy comparison group. **Methods:** We assessed serum parathyroid hormone (PTH), pH-corrected ionized calcium, magnesium, and thyroid-stimulating hormone (TSH) levels in 92 adults with 22q11.2DS (median age 26.0, range 17-65 years; 48.9% female) and 27 healthy controls (median age 26.0, range 18-59 years; 55.6% female). Twenty-five subjects (n=23 22q11.2DS) with a history of hypothyroidism were excluded from TSH analyses. The PTH level was considered 0.3 pmol/L for four subjects (n=3 22q11.2DS) with levels <0.3 pmol/L. Non-parametric tests were used to examine the association between PTH, calcium, magnesium, and TSH values with age and sex. **Results:** Adults with 22q11.2DS had significantly lower median (range) PTH (2.95 (0.3-9.0) vs 4.80 (0.3-7.0) pmol/L, p=0.011), calcium (1.14 (0.91-1.25) vs 1.21 (1.15-1.2) mmol/L, p=0.000), and magnesium (0.77 (0.60-0.97) vs 0.85 (0.74-0.97) mmol/L, p=0.000), and significantly higher TSH (2.13 (0.78-17.98) vs 1.29 (0.47-3.29) mIU/L, p=0.000) levels, than controls. Age correlated negatively with calcium (r=-0.32, p=0.002) and positively with TSH (r=0.29, p=0.017) levels in 22q11.2DS subjects. No significant correlations between age and serum blood values were found in controls. Within the 22q11.2DS and control groups, PTH, calcium, magnesium, and TSH values were not different between males and females. **Conclusions:** The findings of this study suggest that the distributions of PTH, calcium, and magnesium serum levels are shifted to the left and the distribution of TSH serum levels to the right, in adults with 22q11.2DS compared to healthy controls. In addition, older age was associated with lower calcium and higher TSH levels. The results support the importance of the current international guidelines that recommend regular monitoring of endocrine function in adults with 22q11.2DS. Further, we propose considering a low threshold for magnesium supplementation in adults with 22q11.2DS, given increased risk for hypomagnesemia in this population.