



038: Vitamin B12 status in adults with 22q11.2 deletion syndrome

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Background: 4-5% of Canadian adults have vitamin B12 deficiency. Treatment of this deficiency is important as it can have lasting effects on neurocognitive function. As neurocognitive features are common in adults with 22q11.2 Deletion Syndrome (22q11.2DS), understanding the prevalence of B12 deficiency in this population will be an important part of preventing and managing these treatable conditions. **Methods:** Through the course of regular clinical assessment, vitamin B12 levels were collected for adults with 22q11.2DS. Vitamin B12 deficiency was defined as <148 pmol/L, marginal levels as ≥148 but <222 pmol/L, and above normal as >652 pmol/L. In individuals with levels <222 pmol/L, vitamin B12 levels post-3 months of treatment with supplemental B12 (1000 mcg/daily) were also collected, where available. **Results:** Of 180 consecutive adults (median age 28 years, n=100 females) with 22q11.2DS who were screened for vitamin B12 levels, n=10 (5.6%) had vitamin B12 deficiency, with a further 31 (17.2%) having marginal levels; n=10 (5.6%) had above normal levels. Of the 41 individuals with marginal levels or lower, 17 who underwent treatment with supplemental B12 had 3 months post-treatment B12 levels available. Of these, 10 (59%) achieved levels >222 pmol/L within 3 months, while 4 (24%) remained deficient or marginal, and 3 (18%) developed above normal levels. **Conclusions:** The results indicate that just under one in every four adults with 22q11.2DS may have marginal or deficient vitamin B12 levels, underscoring the importance of regular screening. Understanding the predictors of vitamin B12 deficiency in 22q11.2DS will be helpful for developing more targeted screening and management strategies for this population. Early identification of this readily treatable vitamin deficiency could help optimize neurocognitive functioning, a worthy goal for these patients.