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Parkinsonian Motor Features in Adults with 22q11.2DS

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Background: Parkinsonian motor features including bradykinesia, rigidity, and rest tremor have emerged as later onset manifestations of 22q11.2 deletion syndrome (22q11.2DS). However, prevalence of these features and their relationship to antipsychotic treatment and other factors are yet to be determined. We investigated the lifetime prevalence of parkinsonian motor features in adults with 22q11.2DS through history from patients and family members, review of medical records, and direct assessments using standardized scales. **Methods:** Between July 2015 and April 2017, 92 adults (median age 26, range 17-65, years; n=45 female; n=29 currently receiving antipsychotic treatment) with a molecularly confirmed 22q11.2 deletion entered the study. We assessed motor features using the Movement Disorder Society-sponsored revision of the Unified Parkinson's Disease Rating Scale (MDS-UPDRS, part III) and standard criteria for the presence or absence of parkinsonian features (n=88). We used non-parametric tests to investigate the association between MDS-UPDRS motor scores (bradykinesia, rigidity and rest tremor) and factors that could affect motor functioning, after excluding those with Parkinson's disease. **Results:** Overall, 66.3% (n=61) of patients had one or more lifetime motor feature. Lifetime prevalences were: 18.5% for bradykinesia, 23.9% for rigidity, and 56.5% for tremor; 14.8% of patients presented with resting tremor at examination. Overall, 66.3% (n=61) of patients had one or more motor feature. Two patients had clinically confirmed Parkinson's disease. Increasing age was significantly correlated with higher bradykinesia scores in those receiving antipsychotic medication ($r=0.42$, $p=0.032$; n=26), but did not reach statistical significance in those without antipsychotic medication ($r=0.25$, $p=0.059$; n=60). The only significant sex difference identified was that bradykinesia MDS-UPDRS scores were higher in females than males for those not on antipsychotic medication ($p=0.021$). **Conclusions:** The findings of this study suggest that parkinsonian motor features are common manifestations in adults with 22q11.2DS. In addition, in this relatively young sample, the results suggest that as patients age, or with antipsychotic medications, bradykinesia may become more apparent. Whether these increased motor dysfunction scores represent evidence of prodromal Parkinson's disease or other neurodegenerative processes will require further prospective studies.