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CGG REPEAT NUMBER AT THE FRAXA AND FRAXE LOCI DOES NOT SEEM TO BE INVOLVED IN PARKINSON DISEASE

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Carriers of expanded or intermediate alleles of the fragile X mental retardation (*FMR1*) and the fragile X mental retardation 2 (*FMR2*) have been associated with Parkinson's disease (PD). To determine whether these genes are risk factors in our PD population, two hundred and thirty patients (132 men and 98 women) and 227 control subjects (129 men and 98 women) with similar age and sex distribution, but without history of neurological diseases, were recruited from southern Spain and screened for the size of CGG repeats at the FRAXA and FRAXE loci. Diagnosis of idiopathic PD was made using the criteria of the United Kingdom PD Society Brain Bank. Clinical data including familial history of PD were collected. Genomic DNA was amplified by radioactive PCR for the FRAXA locus and size was estimated using a standard curve. The FRAXE locus was analyzed with an ABI 3130 DNA fluorescent-sequencer. FRAXA and FRAXE allele size were analyzed from control and PD patients to check whether the two groups come from the same population. The results show that there is no statistical difference in the size of CGG repeats at the FRAXA locus between control and PD patients. Particularly, the frequency of intermediate alleles, considered from 41 to 55 repeats, was not statistically different in both groups. Two small premutations, confirmed by Southern blot, were found in PD patients but were absent in the control group. The FRAXE locus showed the same distribution in control and PD patients and no intermediate alleles were found in our study. These results suggest that intermediate alleles at the FRAXA or FRAXE loci are not involved in the pathogenesis of PD in the PD population studied here.