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X-chromosome trinucleotide repeats: Effect on brain structure

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Fragile X Syndrome (FraX), the most common form of inherited mental
retardation, is caused by expansion of CGG trinucleotide repeats. Healthy
subjects have <50 repeats, premutation carriers have 50-200 and FraX
individuals have >200 repeats. In this study, we hypothesised that
expanded CGG trinucleotide repeats have significant effects on brain
morphometry. We studied 12 normal IQ male controls, 10 normal IQ male
premutation FraX carriers, and 5 male FraX individuals. Magnetic
Resonance Imaging (MRI) scans were obtained on a 1.5 Tesla MRI
scanner. Group differences in grey matter volume were mapped onto a
standardised brain image using SMART software. Compared to controls,
premutation FraX carriers had significant (p<0.001) decreases in grey
matter volumes in right side cerebellum, middle temporal gyrus, post central
gyrus, parahippocampal gyrus and hippocampus. Also, FraX full mutation
subjects, compared to premutation FraX carriers had significantly (p<0.001)
smaller grey matter volume in the right thalamus, mid temporal gyrus and
cerebellum and left parahippocampal gyrus. The FraX showed significant
(p<0.001) increases of grey matter densities located in right side medial
frontal gyrus, cerebellum and postcentral gyrus, and in left cingulate gyrus,
superior frontal gyrus and superior occipital gyrus. Thus, X chromosome
CGG trinucleotide repeats affect brain grey matter volume in FraX
premutation carriers and FraX individuals in a regionally specific nature. Our
results may have important implications for our understanding of the effects
of expanded trinucleotide repeats on brain structure in other triplet repeat
disorders.

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