Title	X-chromosome trinucleotide repeats: effects on brain structure (British Human Genetics Conference, Abstract 4.12)
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Citation	Journal of Medical Genetics, 2000, v. 37 n. 1, p. S58
Issued Date	2000
URL	http://hdl.handle.net/10722/45315
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4.12

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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