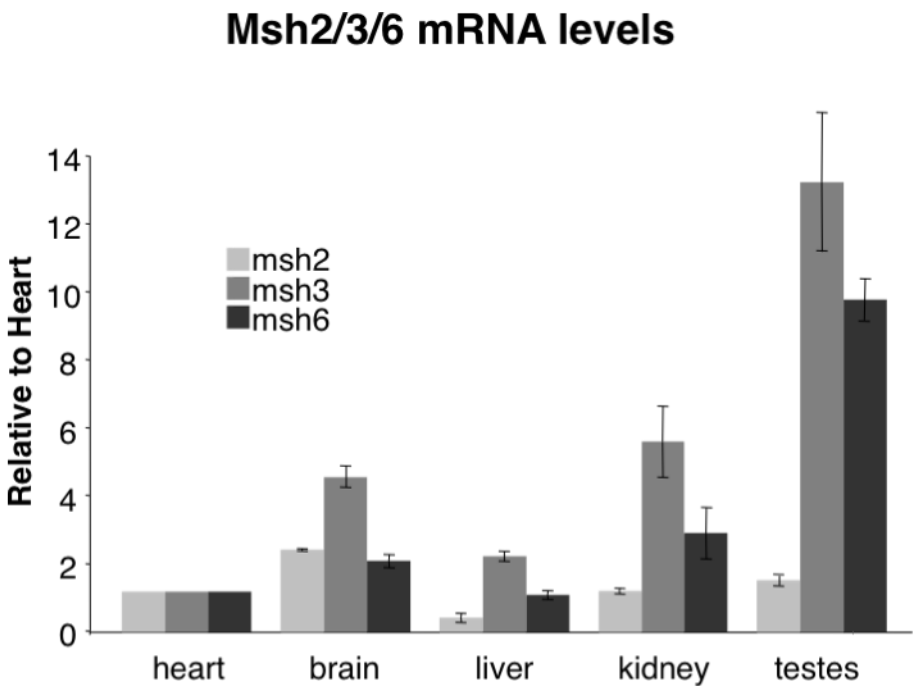
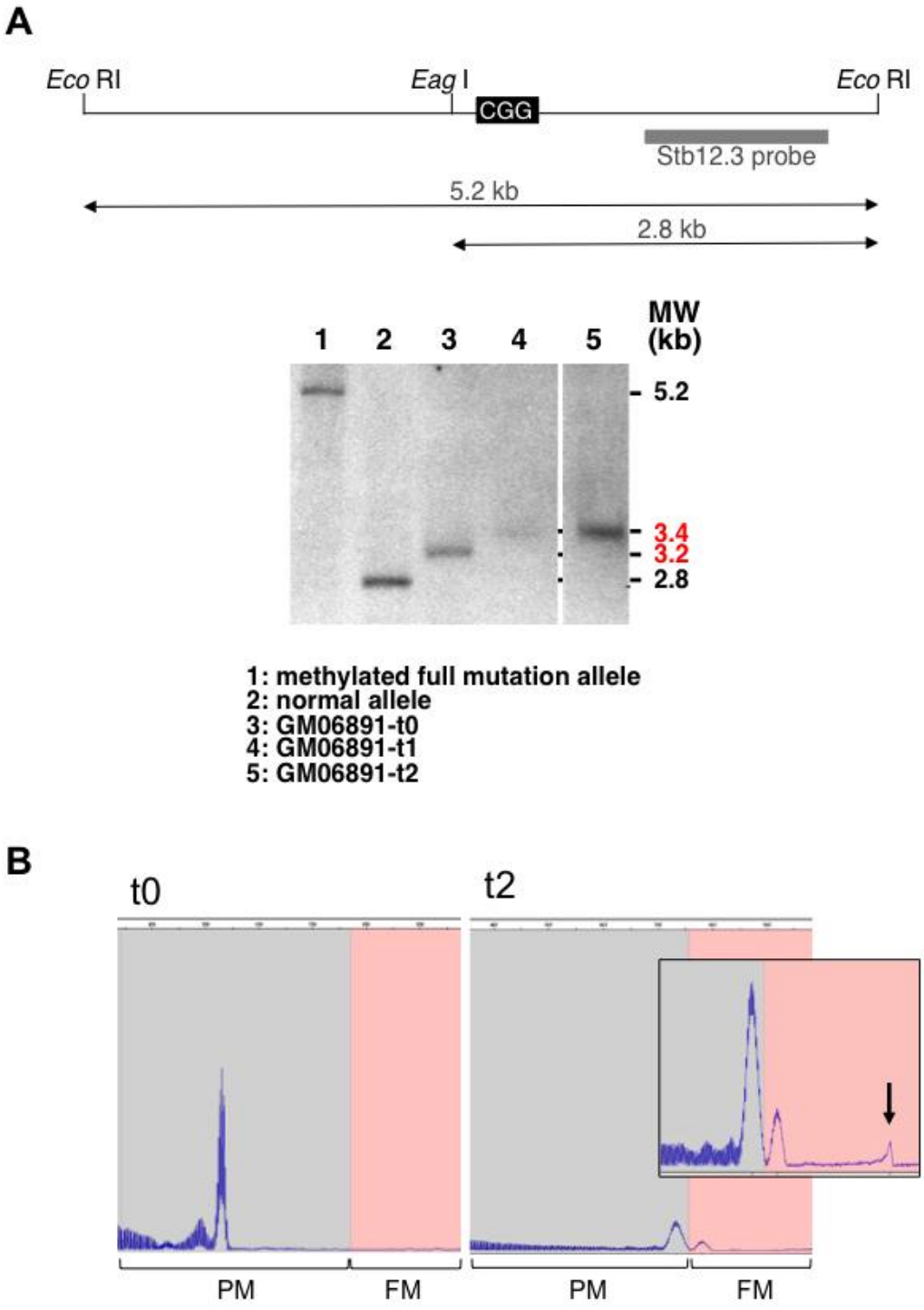
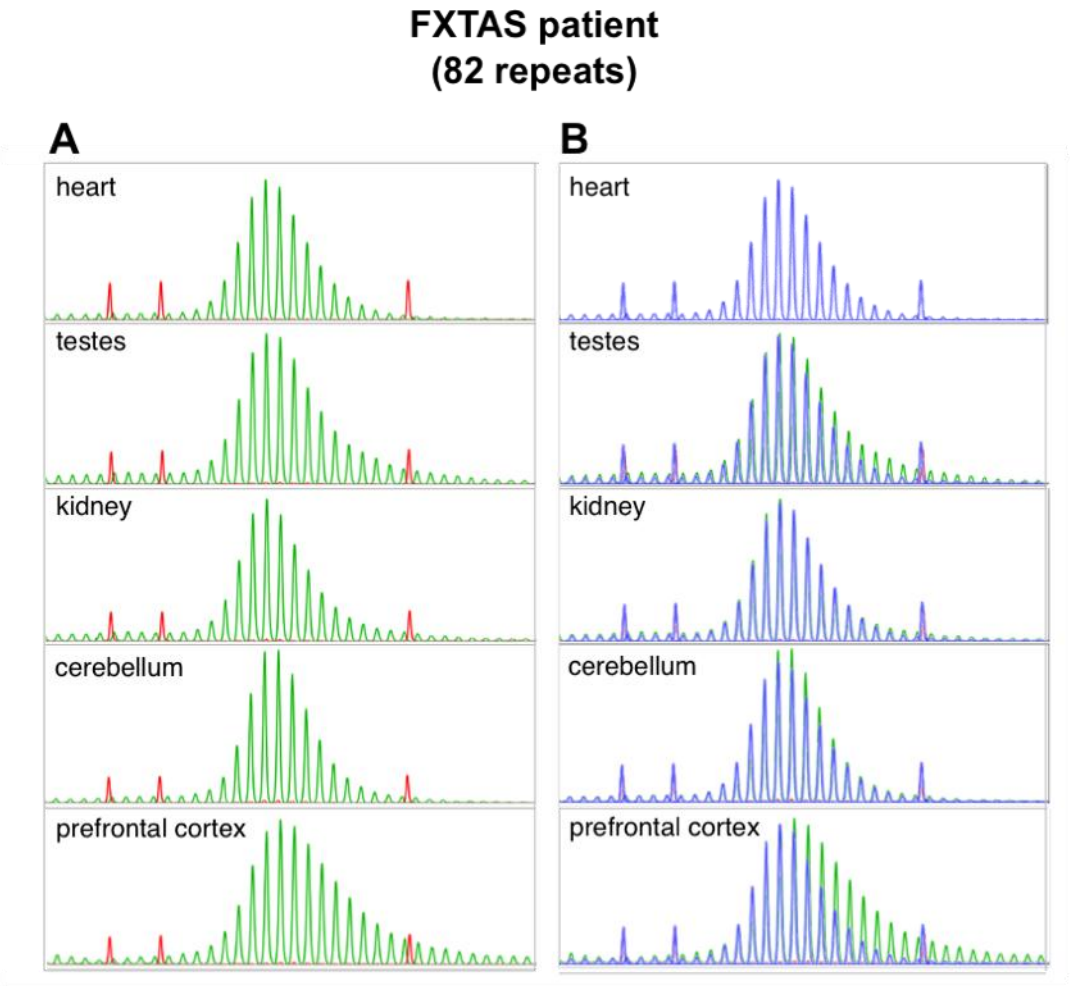


Supp. Figure S1. Msh2, Msh3 and Msh6 mRNA levels in different organs of FX PM mice. Total RNA from different mouse organs was isolated as described in the Materials and methods and the RNA quantitated by RT-PCR using Taqman probe-primer pairs for *Msh2 Msh3* and *Msh6* (Applied Biosystems). The data shown is an average of the transcript levels in 3 animals.



Supp. Figure S2. Analysis of repeat size in passaged cultures of lymphoblastoid cells. Southern blotting and PCR analysis was carried out by the Institute for Basic Research in Developmental Disabilities (Staten Island, NY). A) Southern blotting was carried out according to standard procedures after digestion with *Eco RI* and the methylation sensitive enzyme *Eag I* as described elsewhere (Nolin, et al., 2008). The probe used, Stb12.3 (Oberle, et al., 1991) is located within

the 2.8 kb *Eco* RI-*Eag* I fragment. Normal males show a band of ~2.8 kb in this assay since the *Eag 1* site is unmethylated. Unmethylated full mutation alleles will produce one or more bands of >3.3 kb. Methylated full mutation alleles would be >5.7 kb since cleavage at the 5' *Eag I* site is blocked. The DNA isolated at t2 corresponds to an unmethylated allele of ~3.4 kb. B) PCR analysis was carried out using a commercially available kit for human Fragile X alleles (Asuragen, Inc, Austin, TX). Alleles in the premutation range fall into the grey shaded area, while full mutation alleles fall into the pink shaded area. The DNA isolated at t2 shows 2 PCR products, one corresponding to a large premutation allele of 194 repeats and a second one corresponding to a full mutation allele of >200 repeats. Expansion of the scale on the PCR profile indicates the possible presence of a second full mutation allele that is >280 repeats (indicated by the black arrow in the inset in Panel B).



Supp. Figure S3. Somatic expansion in a FXTAS patient with 82 repeats. A) GeneMapper profiles obtained for the repeat in the indicated tissue of the patient. B) the repeat profiles for different tissue in green, superimposed with the repeat profile from heart shown in blue.

Supp. References

Nolin SL, Ding XH, Houck GE, Brown WT, Dobkin C. 2008. Fragile X full mutation alleles composed of few alleles: implications for CGG repeat expansion. Am J Med Genet A 146A:60-5.

Oberle I, Rousseau F, Heitz D, Kretz C, Devys D, Hanauer A, Boue J, Bertheas M, Mandel J. 1991. Instability of a 550-base pair DNA segment and abnormal methylation in fragile X syndrome. Science 252:1097-102.