Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease

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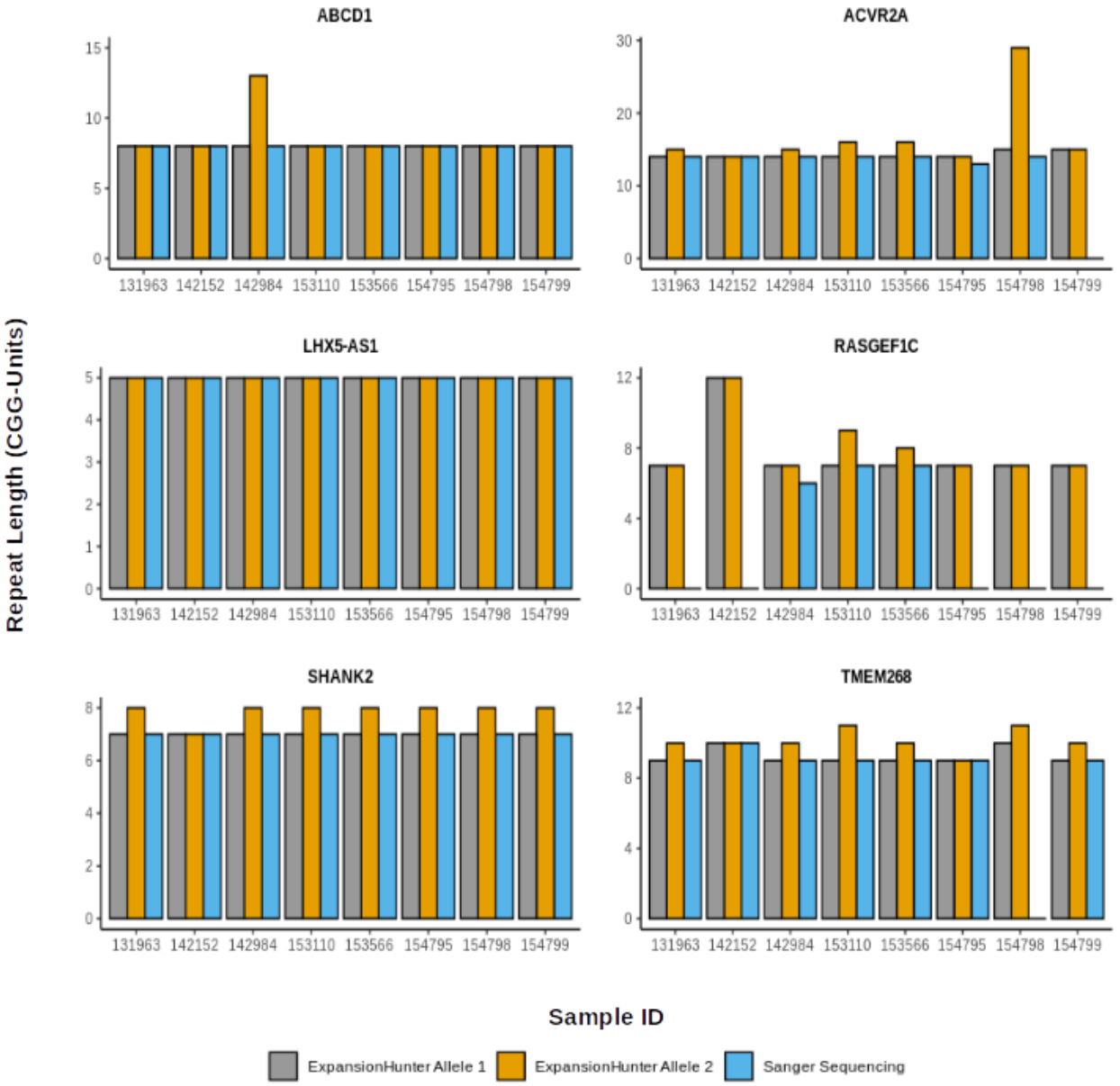
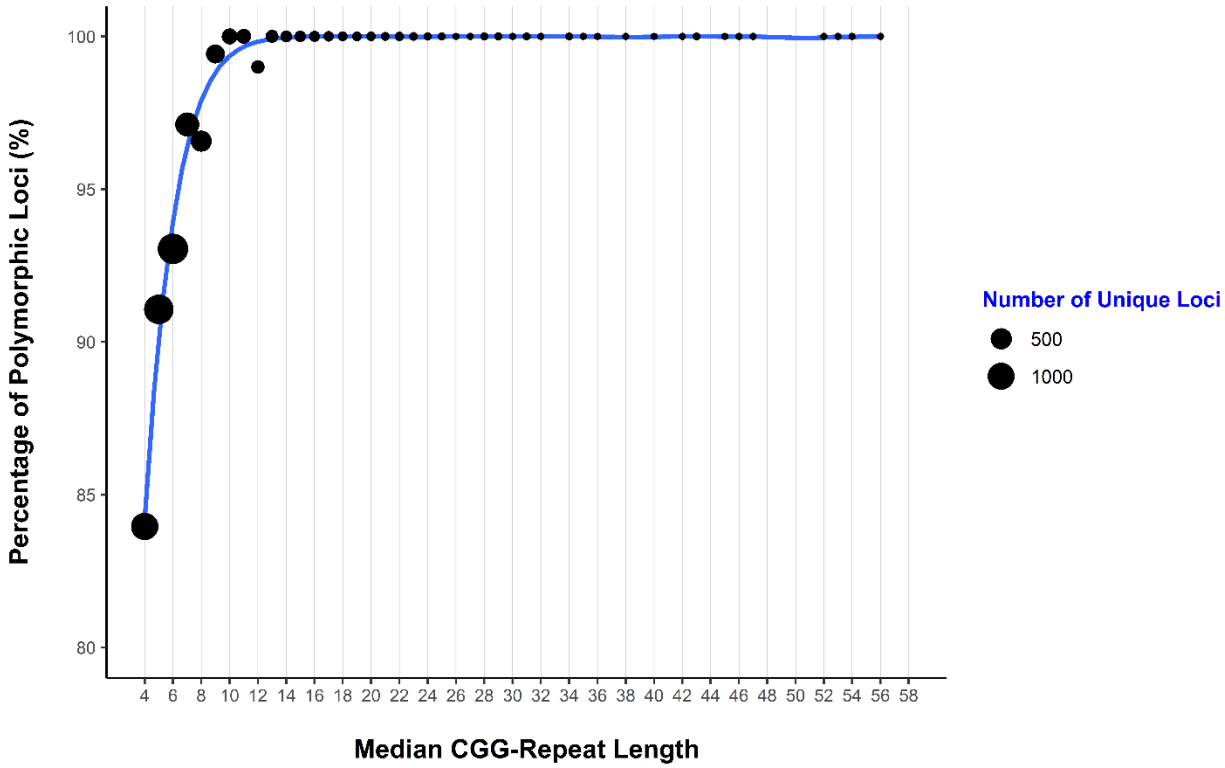
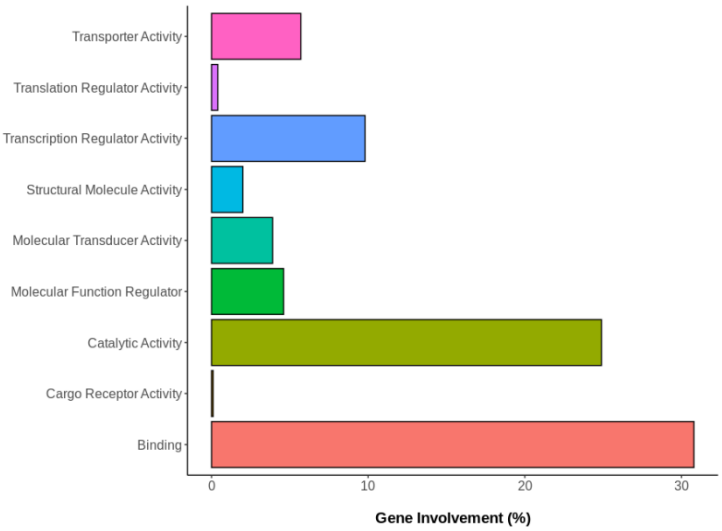


Supplementary Figure 1. Validation of select CGG-Repeats by PCR amplification and Sanger sequencing. Comparison of ExpansionHunter repeat length predictions with observed length of Sanger sequenced repeats. For homozygous alleles ExpansionHunter predictions were identical to sequenced repeats. For heterozygous alleles, only the size of smaller allele could be elucidated. However, these we all identical with ExpansionHunter reports.



Supplementary Figure 2. Relationship between median CGG-repeat length and repeat polymorphisms. As median repeat length increases so does the proportion of repeat loci with that median repeat length that are polymorphic. All STR loci with a median repeat length equal or greater than 12 CGG units were polymorphic among the tested cohort.

a)



b)

Supplementary Figure 3. Percentage of involvement of CGG repeat associated genes with GO defined terms. a) molecular functions and b) protein classes.

Supplementary Table 1. Summary of all CGG-repeat loci detected by ExpansionHunter and their corresponding attributes (*n*=6101).

See the attached txt file Supplementary Table 1.txt

Supplementary Table 2. Median repeat lengths observed among all polymorphic CGG repeat loci (*n*=5673) and the corresponding number of repeat loci that displayed each median repeat length.

See the attached txt file Supplementary Table 2.txt

Supplementary Table 3. Genes of interest. Genes associated with ID, AD, and CGG-repeats with their corresponding haploinsufficiency index, pLI score and cohort polymorphism rate (*n*=410).

See the attached txt file Supplementary Table 3.txt.

Supplementary Table 4. List of genes used genes used for routine screening for ID and related NDD (*n*=1295).

See the attached txt file Supplementary Table 4.txt.