The American Journal of Human Genetics, Volume *95*

Supplemental Data

Genome-wide Identification and Characterization of Fixed Human-Specific Regulatory Regions

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Figure S1

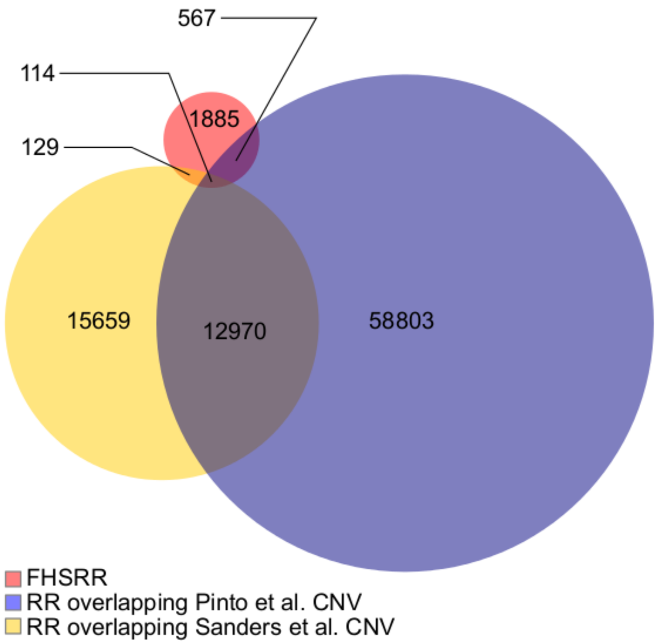

Figure S1: Overlap between FHSRR and Autism spectrum-related CNV regions. The Venn diagram shows the overlap between regulatory regions with the following properties: Red - FHSRRs; Blue - overlapping an autism-related CNV according to Pinto et al.[26]; Yellow - overlapping an autism-related CNV according to Sanders et al.[27]. The intersections are statistically significant: $P = 2.4 \cdot 10^{-4},\ 9.1 \cdot 10^{-21},\ 3.8 \cdot 10^{-3}$ respectively for the Pinto dataset, the Sanders dataset and their intersection (exact Fisher test). However such enrichment was present also in the neutral control, even when the latter was randomly scattered on the genome to prevent proximity with FHSRRs. Therefore the association of human-specific regions with autism-associated CNVs might be due to the higher propensity of human-specific regions to undergo copy number alterations rather than to their functional role.

Figure S2

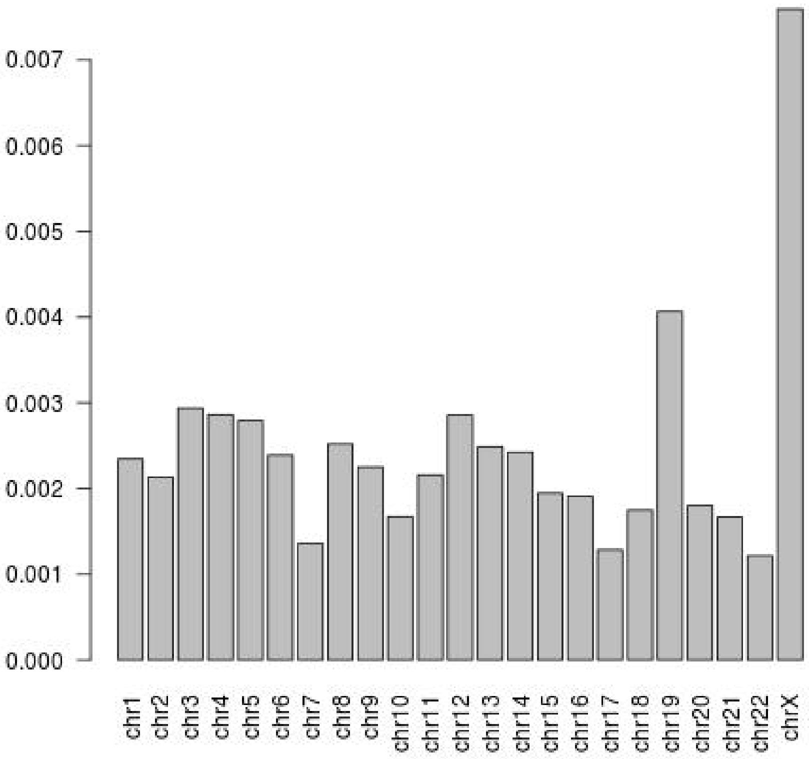

Figure S2: Ratio of genome size occupied by human-specific enhancers (weak and strong, all cell lines merged together) to total genome size occupied by enhancers in the chromosomes.

Figure S3

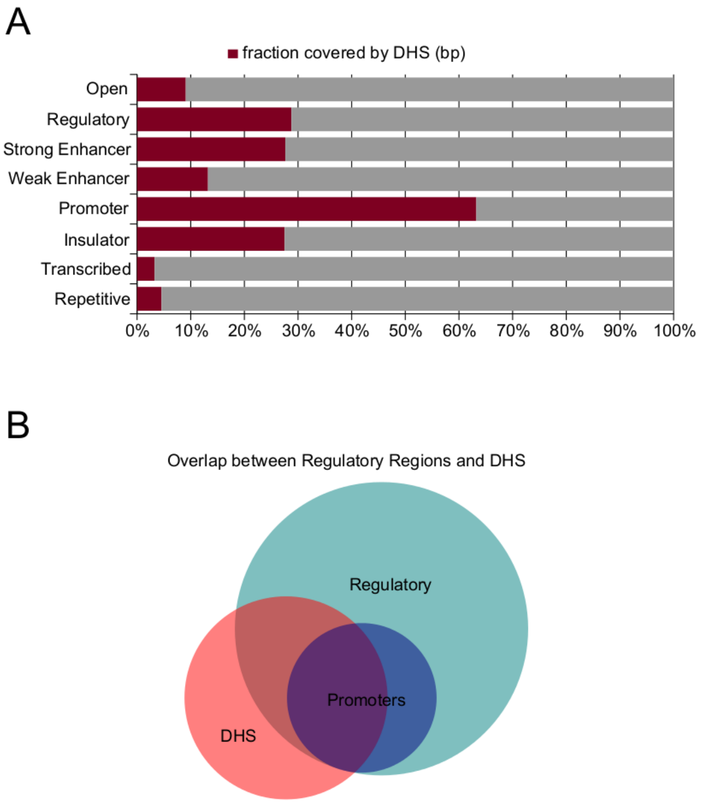

Figure S3: Overlap between DHS and chromatin-based regions in H1hesc cells. A: for each class of regions determined from chromatin data in the Ernst et. al. data[14], the fraction (in terms of base pairs) that is found in open regions according to DHS data; “regulatory” is the union of promoters, strong and weak enhancers, and insulators; “open” is the union of regulatory and transcribed. B: Venn diagram representing the overlap of DHS regions (red), promoters (blue) and regulatory genome (green), showing that promoters are overrepresented in DHS regions compared to other classes of regulatory DNA.