ENIGMA-Vis: A Portal to View Genetic Effects on the Human Brain Based on Large-Scale GWAS

#1546

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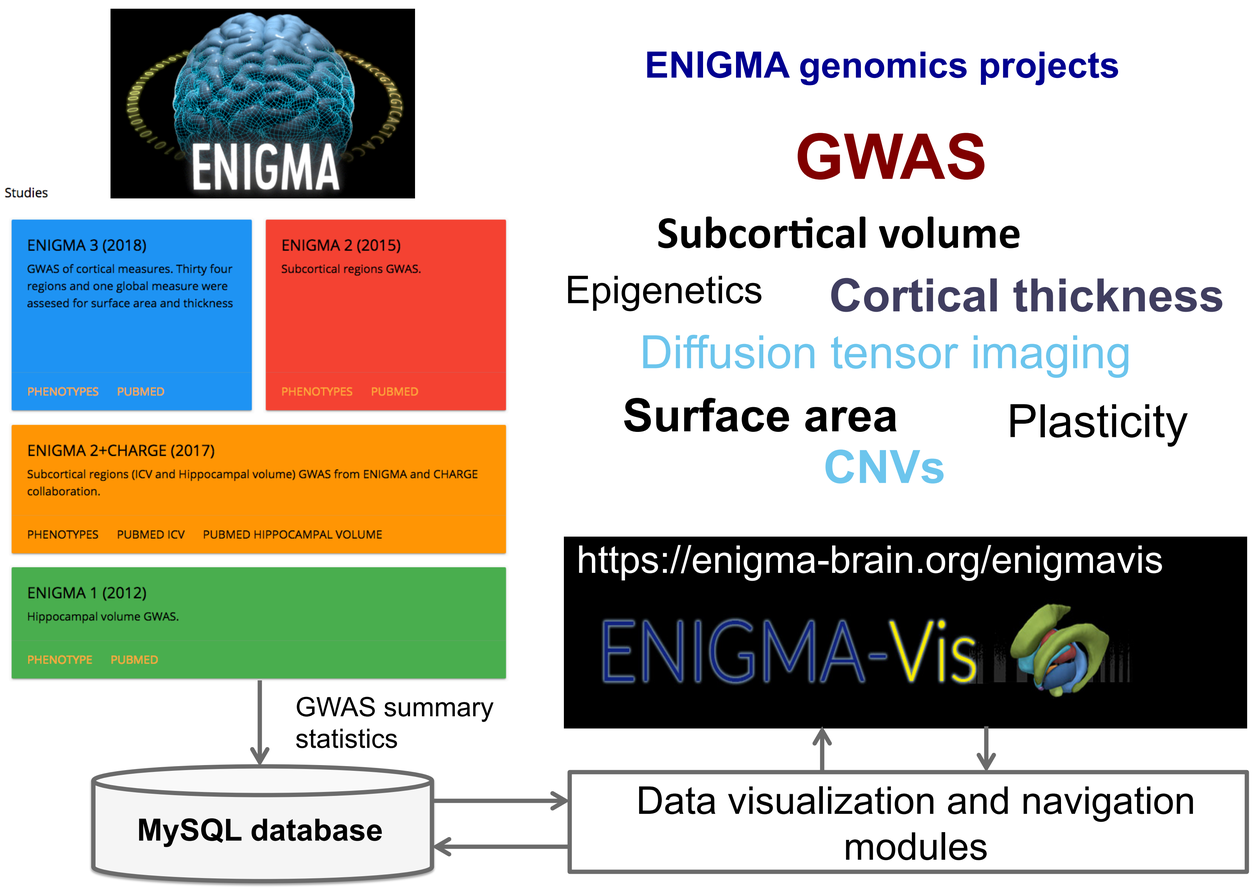
Introduction

In the Big Data era of collaborative neuroimaging, large-scale genome-wide association studies (GWAS) are revealing common genetic variants associated with key brain metrics implicated in neurological and psychiatric diseases (Stein et al., 2012; Hibar et al., 2015). The ENIGMA Consortium, in particular, is leading several efforts that have now identified over 100 loci consistently associated with structural brain variations (Hibar et al., 2015; Adams et al., 2016; Jahanshad et al., 2018; Satizabal et al., 2017), in partnership with the CHARGE consortium. Results from published analyses have been downloaded and used by over 150 labs worldwide, to understand how brain-related genetic loci relate to, for example, disease risk, brain development, cognition, and human brain evolution. ENIGMA's new cortical GWAS adds rich information on patterns of genetic influences on the brain, for large numbers of brain phenotypes and over a million genetic loci. Going from 8 subcortical to 70 cortical phenotypes, means new tools are needed to query, visualize, and navigate the effects, and relate them to other GWAS (e.g., on disease risk).

Inspired by the design and features of the PheWeb tool VandeHaar, 2017; http://pheweb.sph.umich.edu/) which provides useful navigation and visualization functionality for non-neuroimaging datasets, we implemented added functionality into ENIGMA-Vis, the ENIGMA consortium's GWAS visualization tool:

https://enigma-brain.org/enigmavis



ENIGMA genomics projects

GWAS

Subcortical volume

Epigenetics **Cortical thickness**

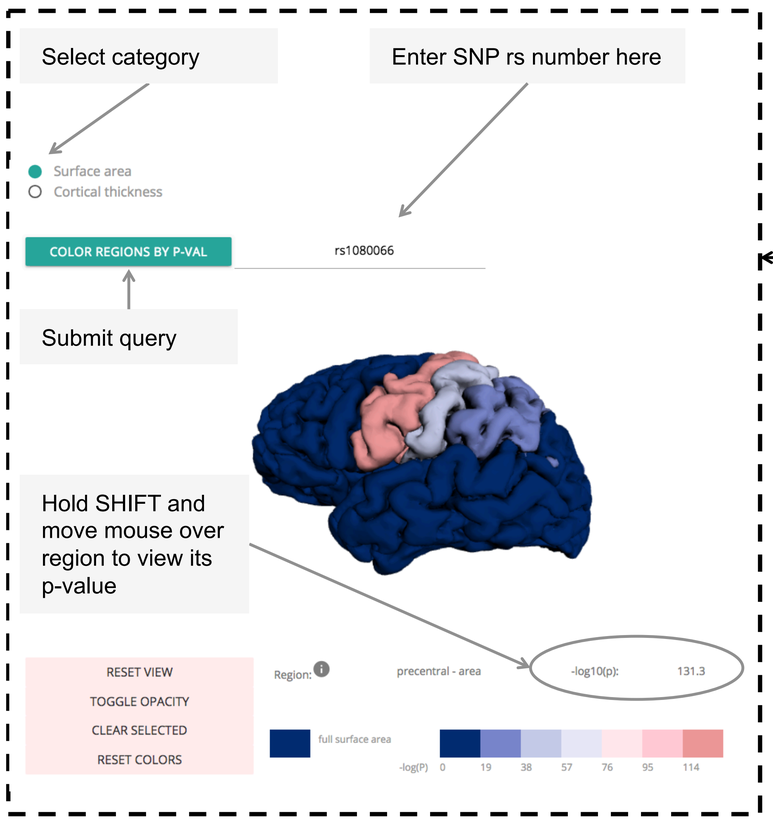
Diffusion tensor imaging

Surface area Plasticity

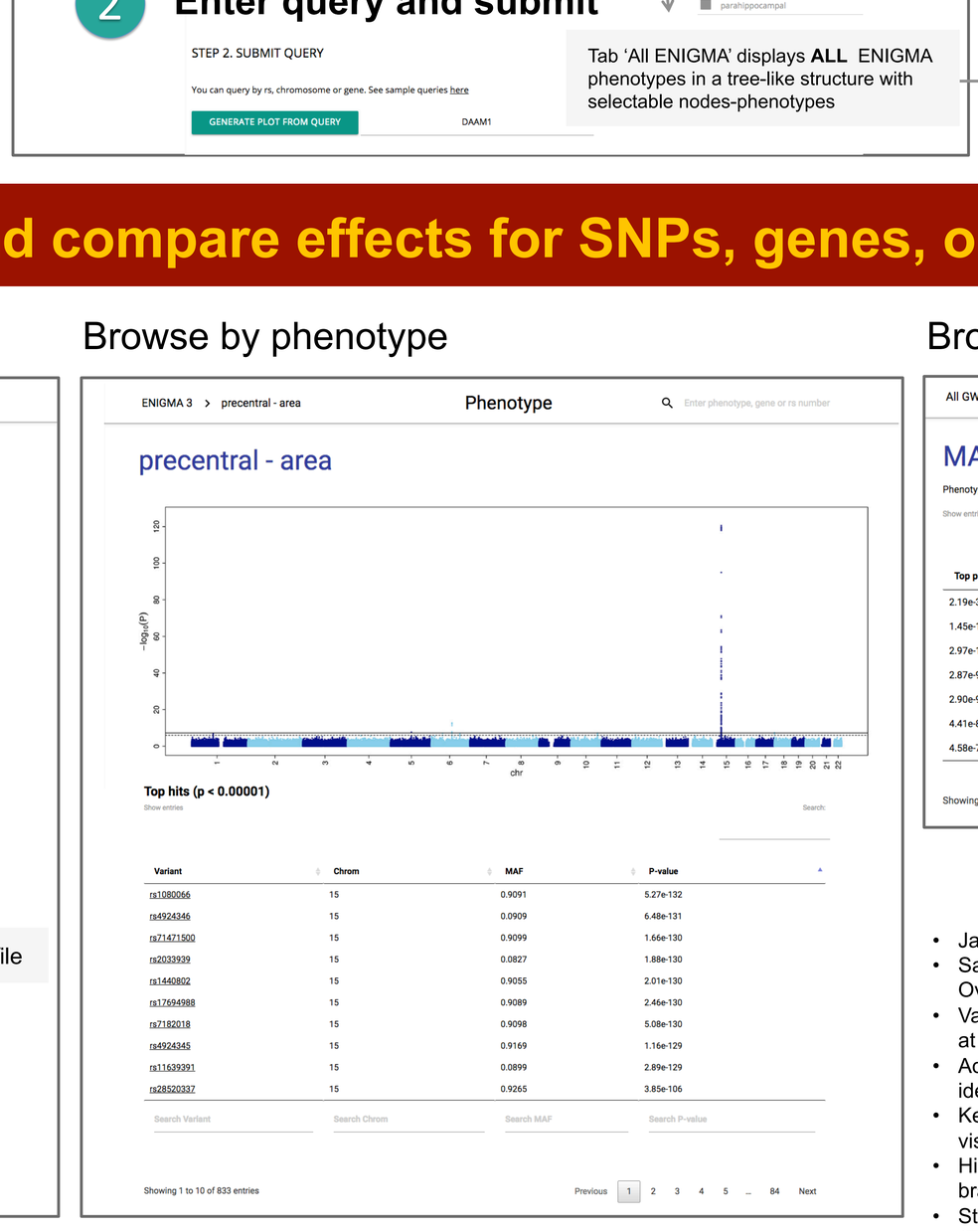
CNVs

Visualizing GWAS results in multiple ways

Tool 1: Color cortical regions by their p-value for cortical thickness or surface area traits (ENIGMA 3 cortical study only)



Tool 2: Generate regional Manhattan plot from query



Browse results and compare effects for SNPs, genes, or brain phenotypes

Browse by SNP and compare effects

Browse by phenotype

Browse by gene

Acknowledgements

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