

The NIH NeuroBioBank Whole-Genome Catalog: Sequencing from human brain donors with CNS disorders

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10,000 Whole Genomes Paired with Bio-Banked Tissue and DXNs

ABSTRACT

- We generated genome-wide genetic data from 9,663 brain tissue donors with in-depth clinical and neuropathological phenotyping
- The clinical diagnoses of these donors include 148 CNS diseases clustered in 15 ICD-10 categories
- 171,121,209 unique mutations and paired phenotype data is available in the NIMH Data Archive and NeuroBioBank Portal
- To illustrate an application of this resource, we replicated the association between the HTT gene and Huntington's Disease, and searched for broad gene-based associations to categories of neurological and psychiatric disease




NIH NeuroBioBank
Facilitating Research and Creating Awareness



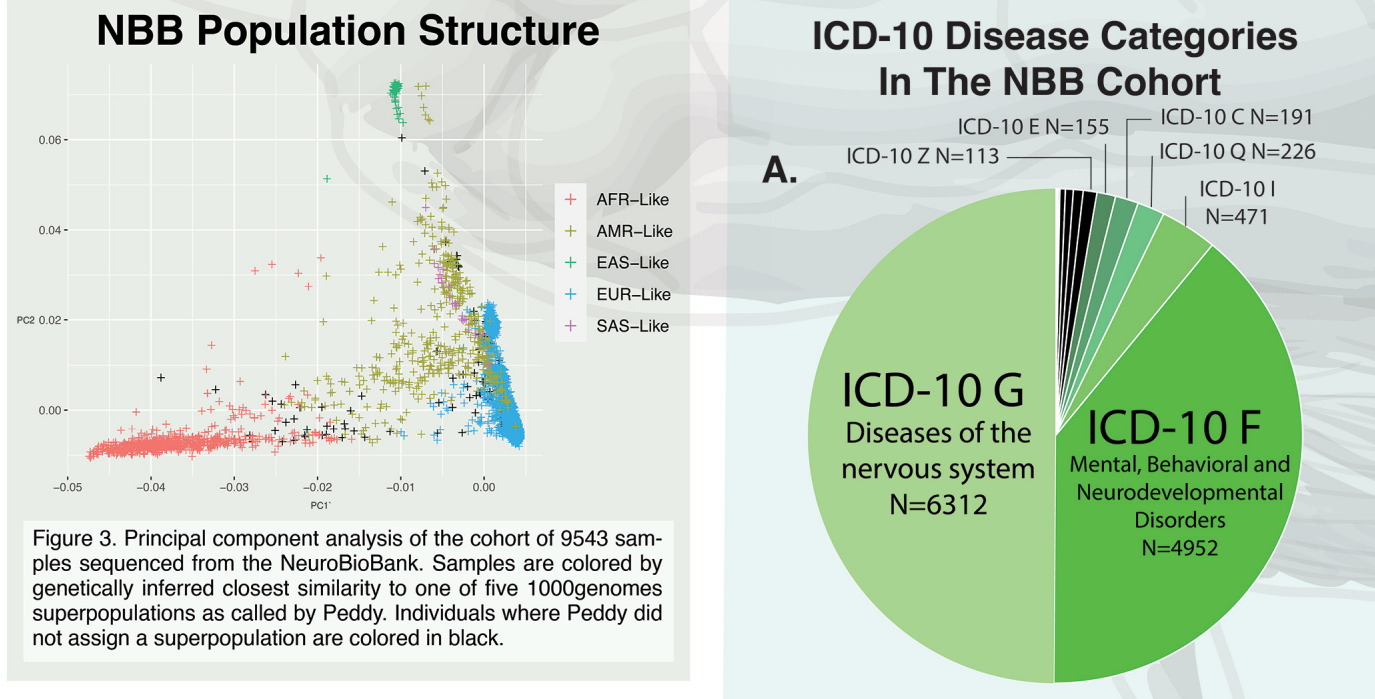
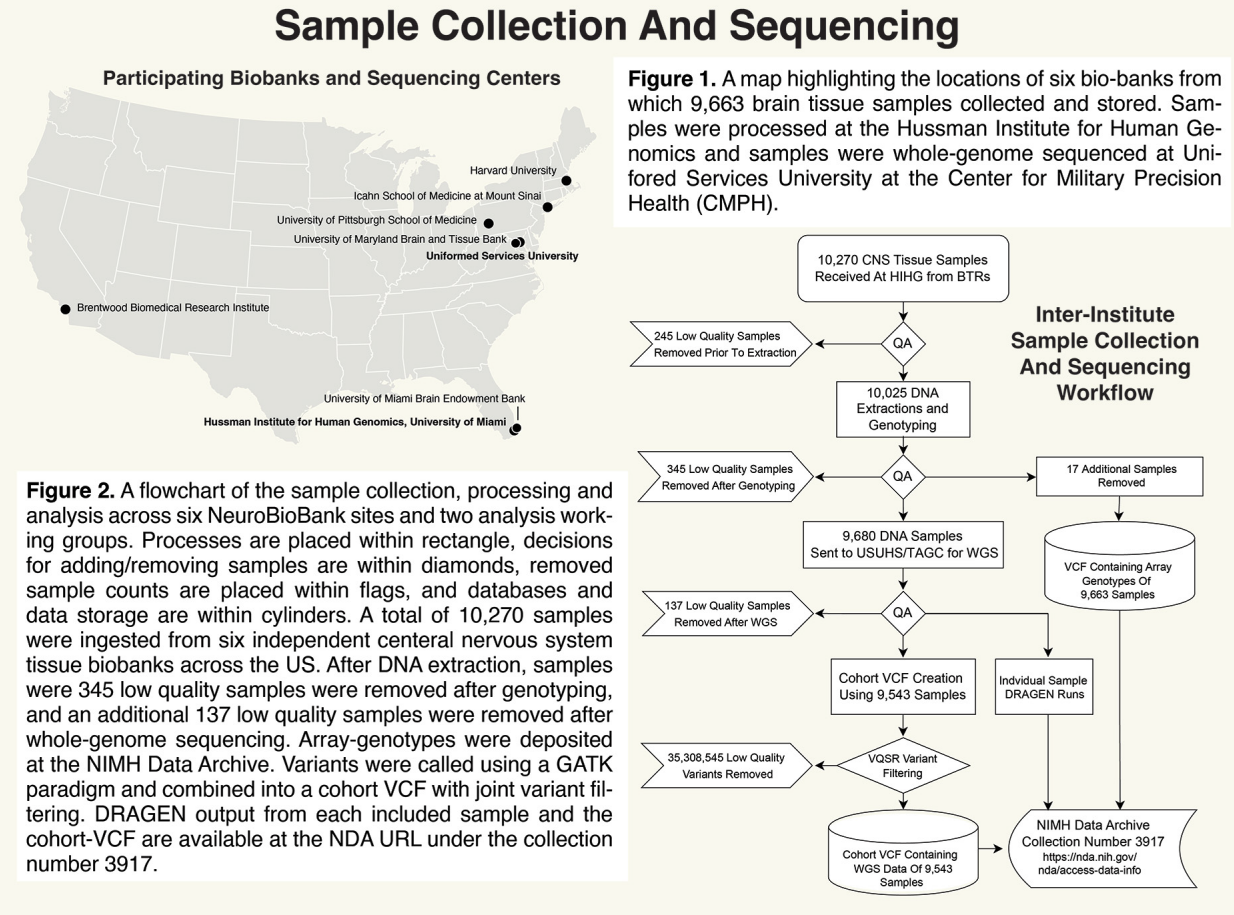
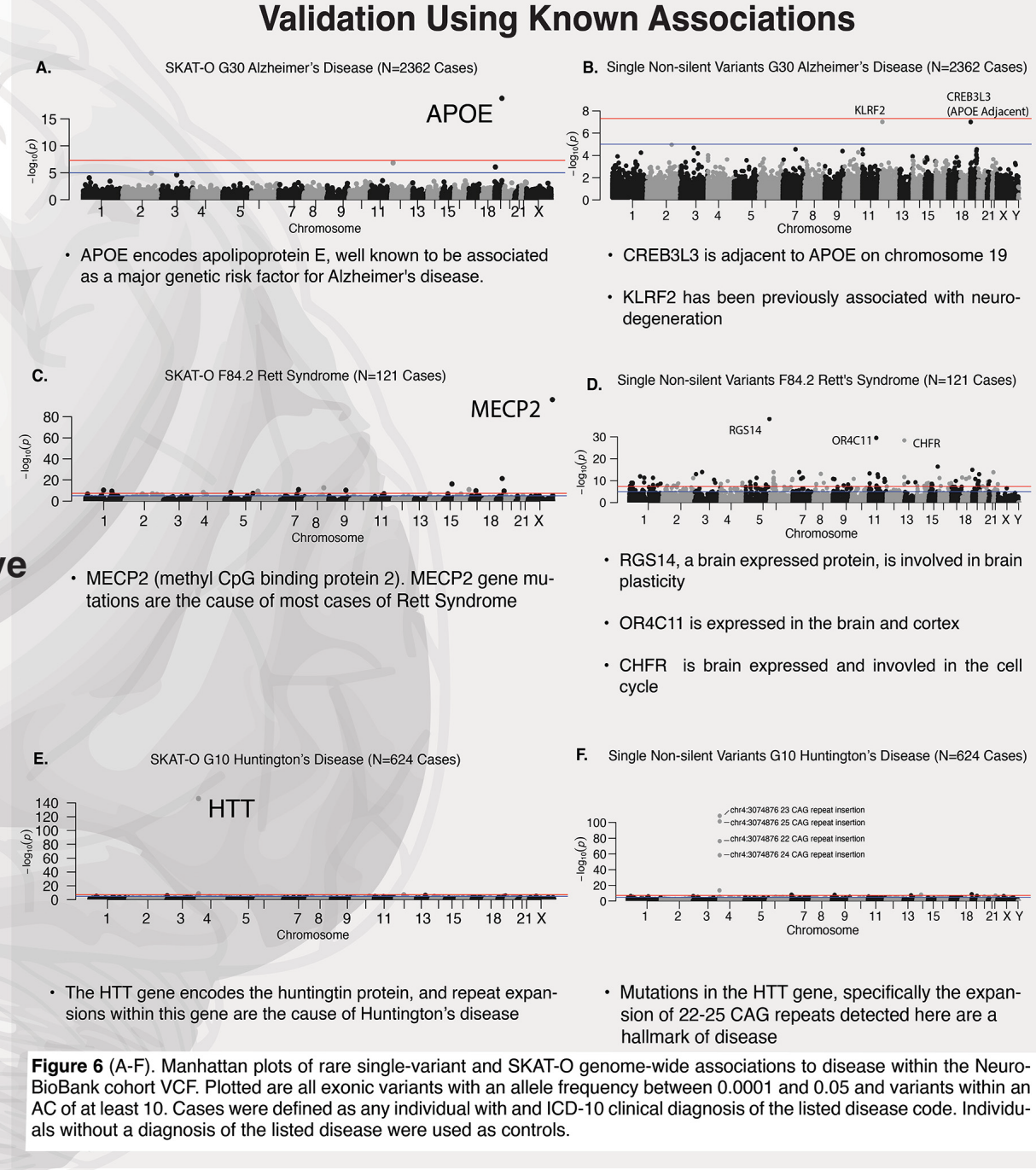
Browse Phenotypes and Request Tissue on the NeuroBioBank Website



NIMH Data Archive



Request Access To WGS From The NIMH Data Archive



Biobank	Miami	Pittsburgh	Mt.Sinai	Sepulveda	Maryland	Harvard	Total
Passing Samples (Male)	820 (469)	572 (399)	1575 (760)	2419 (1245)	1432 (824)	2725 (1480)	9543 (5177)
Mean Age (SD)	65.6 (19.4)	46.9 (13.7)	76.2 (13.6)	67.6 (17.3)	41.2 (30.7)	67.4 (17.4)	60.8
EUR-like	651	470	1185	2216	1136	2627	8285
AFR-like	87	84	202	45	170	21	609
SAS-like	3	2	7	4	8	2	26
EAS-like	4	1	9	22	9	5	50
AMR-like	55	9	140	100	85	40	429
Admixed	20	6	32	32	24	30	144
Total	820	572	1575	2419	1432	2725	9543
Avg Coverage Dragen	37 (3.4)	37.5 (4.0)	37.3 (3.4)	38.2 (4)	37.4 (3.5)	37.6 (4.2)	37.5

Table 1. Cohort statistics for samples within the NeuroBioBank whole-genome sequencing cohort. Sample statistics are displayed for samples that passed QA thresholds and are binned according to site. Demographic super-populations are abbreviated for those most similar to ancestry from Europe (EUR-Like), Africa (AFR-Like), South Asia (SAS-Like), East Asian (EAS-Like), and the Americas (AMR-Like). Individuals who displayed admixed ancestry were not categorized by most-similar ancestry.

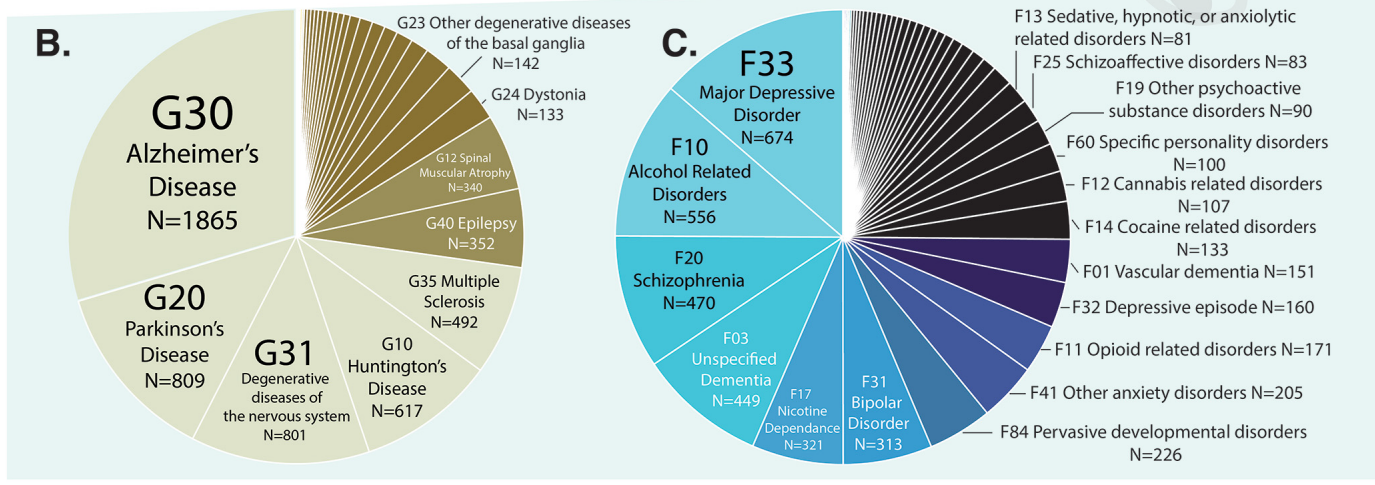



Figure 4. Disease distribution in the NeuroBioBank cohort for clinical diagnoses. **A.** Plot showing the most prevalent ICD-10 codes among clinical diagnoses, highlighting codes G and F as the primary components of the NeuroBioBank cohort. **B.** Prevalence of codes listing individual diseases and disease categories from ICD-10 G, diseases of the nervous system. **C.** Prevalence of codes listing individual diseases and disease categories from ICD-10 F, mental, behavioral, and neurodevelopmental disorders. Total number of diagnoses for each ICD-10 code are listed for each bin for the major components of each category. Diseases with fewer cases than visible are not plotted.

Acknowledgements / Disclaimer

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 hjf Henry M. Jackson Foundation for the Advancement of Military Medicine
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