Data Sharing in the GREGoR Consortium to Support Rare Genetic Disease Research

Background: The GREGoR Consortium (Genomics Research to Elucidate the Genetics of Rare diseases) seeks to develop and apply approaches to discover the cause of currently unexplained Mendelian genetic disorders.

GREGoR Consortium Data is available on NHGRI's AnVIL platform.

Researchers can access GREGoR data via dbGaP study phs003047.

The GREGoR Consortium data model available at https://github.com/UW-GAC/gregor_data_models

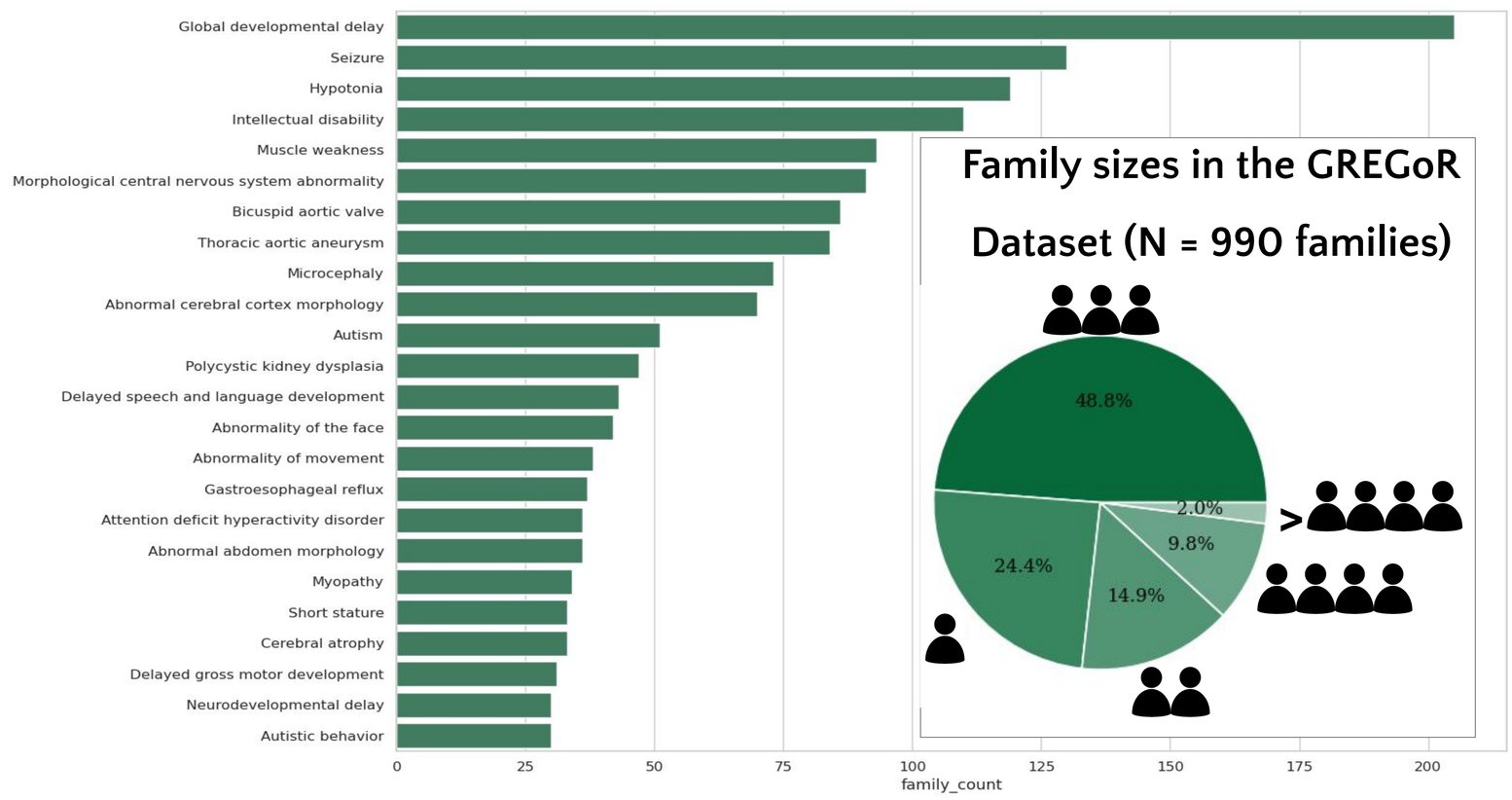
WHO SHOULD STOP HERE?

- Rare disease curators/analysts/tool developers
- Clinicians who want to query patients' variants in a rare disease cohort
- Bioinformaticians interested in data processing and modeling
- Anyone interested in accessing a rare disease dataset

Consortium Data Summary

	Participants	+ WES/WGS +phenotype	+WES/WGS +RNAseq +phenotype
Probands	951	854	75
Other Affected	182	157	7
Unaffected	1239	86	59
Possibly Affected or Unknown Status	140	10	0
Total	2512	1107	141

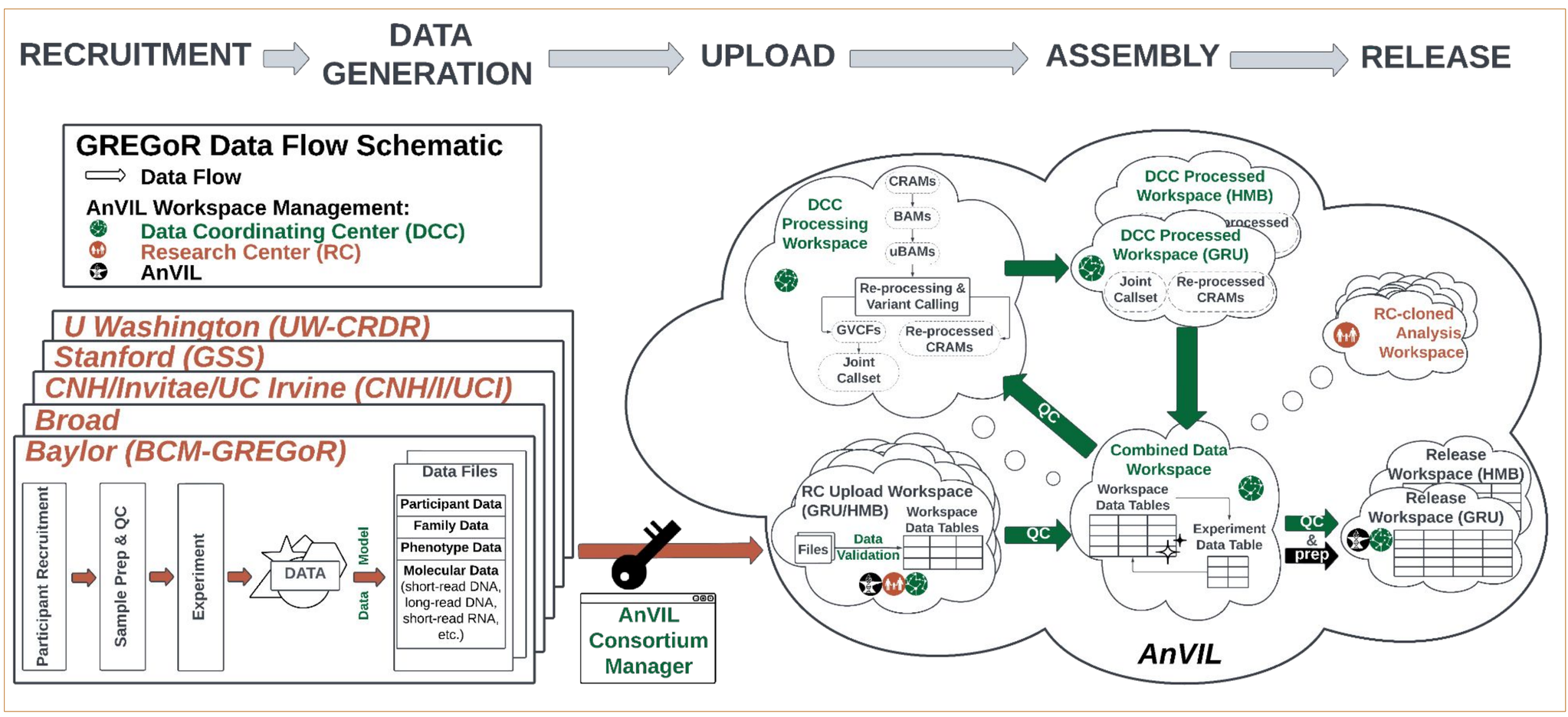
Most Frequent GREGoR Phenotypes (HPO Term Counts)



Supported Data Types

Current	Coming Soon
Family structure	Optical Mapping
Phenotypes (HPO)	Metabolomics
Short Read ES/WGS	Functional Data
Short Read RNAseq	+ more!
Candidate Variants	
Long Read WGS (ONT)	
Long Read WGS (Pa Bio)	
ATAC-Seq	
Joint Callset	

Using AnVIL workspaces for GREGoR Data Sharing and Analysis



http://gregorconsortium.org/

Heavner BD, Berger SI, Chong JX, Jhangiani SN, O'Donnell-Luria A, Austin-Tse C, Baxter S, Conomos MP, Coban-Akdemir Z, Délot E, Goddard PC, Gogarten SM, Jolly A, Mahmoud M, Marwaha S, Miller DE, Posey JE, Rehm H, Sanchis-Juan A, Stilp AM, Tong CC, Ungar RA, Wheeler M, Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR)

GREGoR consortium

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