Supplemental Material

Supplemental Table 1. CMG discovery case summary sheet

See supplemental excel

Supplemental Table 2. CMG data sharing details

Center	ClinVar Name	ClinVar URL	dbGaP/AnVIL Study accession ID
Baylor-Hopkins	Lupski Lab, Baylor-Hopkins CMG, BHCMG	https://www.ncbi.nlm.nih.gov/clinvar/submitters/505572/	phs000711
Baylor-Hopkins	Baylor-Hopkins Center for Mendelian Genomics (Johns Hopkins University)	https://www.ncbi.nlm.nih.gov/clinvar/submitters/505755/	phs000711
Broad Institute	Broad Institute Rare Disease Group (Broad Institute)	https://www.ncbi.nlm.nih.gov/clinvar/submitters/506627/	phs001272
University of Washington	University of Washington Center for Mendelian Genomics (University of Washington), UW-CMG	https://www.ncbi.nlm.nih.gov/clinvar/submitters/505516/	phs000693
Yale	Yale Center for Mendelian Genomics (Yale University)	https://www.ncbi.nlm.nih.gov/clinvar/submitters/506150/	phs000744

Supplemental Table 3. CMG AnVIL metadata dictionary and example files

See supplemental excel

Supplemental Table 4. Access Levels in AnVIL

Access type	Description	Example Dataset	Additional documentation
Open Access	These data sets and tools are accessible to anyone with an AnVIL, Terra or Gen3 account.	1000 Genomes	https://anvilproject.org/learn
Controlled-Access	Access to these data sets is managed by a data access committee designated by dbGaP and is based on intended use of the requester and allowed use of the data submitter as defined by consent codes.	CMG cohorts, GTEx	https://anvilproject.org/learn/accessing-data/requesting-data-access#accessing-controlled-access-data
Consortium Access	These data sets are accessible to consortia members and controlled under the consortium's data sharing agreement.	Centers for Common Disease Genomics (CCDG), Cohorts that are not yet released from their embargo period	https://anvilproject.org/learn/for-consortia/consortium-data-access-guidelines