

Study ID	Data category	Dataset title
STUDY0127	Genomics	Exome and SNP array reanalysis
STUDY0070	Genomics	Genetic contributions to NMO
STUDY0011	Genomics	Genetic determinants of MS disease course
STUDY0100	Genomics	Genome sequencing and SNP genotyping
STUDY0077	Genomics	MS susceptibility genetic association study
STUDY0065	Genomics	Risk factors for pediatric-onset NMO
STUDY0012	Genomics and serology	Vitamin D-related genes and MS risk

STUDY0123	Immune repertoire	T-cell receptor repertoire in MS
STUDY0060	Proteomics	Mass spectrometry signatures of MS
STUDY0092	Proteomics	Protein biomarkers of MS inflammatory activity
STUDY0144	Serology	AQP4 and MOG antibodies in demyelinating disease
STUDY0095	Transcriptomics	Cell-free RNA signatures of MS disease activity
STUDY0141	Transcriptomics	RNA sequencing and MS disease severity
STUDY0094	Transcriptomics	Serum microRNA biomarkers for MS diagnosis

STUDY0130	Transcriptomics	Sex-based differences in MS gene expression
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Term	Definition
Counts	Counts are reproduced from the supplied catalog. Do not sum across datasets:
Samples (n)	Number reported under “Samples” in the source. The source does not specify \
Longitudinal (n)	Number reported under “Longitudinal” in the source. The unit and relationship t
Disease groups	Counts retain the original disease categories. NMO is the source category and
MS	Multiple sclerosis
TM	Transverse myelitis
NMO	Neuromyelitis optica
ADEM	Acute disseminated encephalomyelitis
CIS	Clinically isolated syndrome
ON	Optic neuritis
Controls	Source control group; health status is not specified consistently.

Dataset description

Secondary analysis of exome sequencing and SNP genotyping data associated with STUDY0100.

Reanalysis of genome-wide association data to investigate genetic contributions to neuromyelitis optica (NMO).

Genotyping data used to validate findings from a genome scan of 1,000 people with MS and to explore genetic variants associated with MS susceptibility and disease course.

Genomic data for studying genetic variation in MS and related disorders, including structural variants. Genome-wide array data were generated using a global screening array.

Genotyping data used to extend and replicate genetic associations with MS susceptibility. Repository samples contributed to a larger study of 18,000 cases and 18,000 controls using an exome array with additional MS-relevant variants.

Genotyping data from a study of pediatric-onset NMO risk factors. The study also examined vitamin D and previous viral infections.

Genetic data for exploring associations between vitamin D metabolism and MS risk. Data also include antibody measurements against Epstein–Barr nuclear antigen 1 (EBNA-1).

T-cell receptor (TCR) sequences for studying immune signatures in MS and related demyelinating disorders. Returned data also include analyses of TCR associations with human leukocyte antigen (HLA) types, including HLA-A*03:01 and glatiramer acetate treatment response.

Mass spectrometry data for exploring protein signatures that distinguish MS from controls and active MS from remission.

Protein measurements for investigating inflammatory activity in MS. Data include relative protein expression and absolute protein concentrations, including serum neurofilament light (sNfL) and glial fibrillary acidic protein (GFAP).

Aquaporin-4 (AQP4) and myelin oligodendrocyte glycoprotein (MOG) antibody measurements in samples originally categorized as NMO. These data support classification of neuromyelitis optica spectrum disorder (NMOSD) and MOG antibody-associated disease (MOGAD).

RNA expression data for exploring transcriptional signatures associated with MS disease activity.

Pilot RNA sequencing dataset from 100 repository samples for exploring associations between gene expression and MS disease severity, measured by the Expanded Disability Status Scale (EDSS). The study aimed to support development of models of disease progression and treatment response.

Serum microRNA measurements for evaluating candidate biomarkers that may aid MS diagnosis. MicroRNAs are short RNA molecules that regulate gene expression.

Gene expression data for investigating biological differences between males and females with MS. Serum neurofilament light (sNfL) measurements are also available.

: samples may overlap.

whether these are unique participants or specimens.

o the sample count require confirmation.

is not automatically relabeled as NMOSD or MOGAD.

Assay / data type	Available files
Exome sequencing and SNP genotyping reanalysis	PLINK files (BED, BIM, FAM)
Genome-wide SNP genotyping and reanalysis	SNP identifier data in encrypted GPG files and decrypted TXT/CSV files, with compressed GZ files
SNP genotyping	Raw genotype data (CSV) and cleaned datasets
Genome sequencing and SNP array genotyping	CRAM files, gVCF/pVCF files, and copy-number/structural variant calls
Custom MS SNP array genotyping	Raw and quality-controlled genotype datasets
SNP array genotyping	PLINK files (BED, BIM, FAM)
SNP genotyping and EBNA-1 IgG measurements	SNP datasets and antibody measurements

T-cell receptor repertoire sequencing	Returned TCR sequence and analysis datasets
Mass spectrometry	Raw spectra in TXT files linked to repository sample barcodes
Protein expression and concentration assays	Protein measurement datasets
AQP4/MOG serological immunoassays and serum neurofilament light (sNfL) assay	CSV files with antibody titers, positivity thresholds, and NfL concentrations (pg/mL)
RNA expression profiling	Processed expression datasets
Barcode-integrated reverse transcription (BIRT) RNA sequencing, including coding and non-coding RNA	Molecular datasets and associated metadata
Reverse transcription quantitative PCR (RT-qPCR) microRNA profiling	CSV files with quantitative microRNA measurements, microRNA identifiers, and sample barcodes

RNA sequencing with differential expression analysis

Normalized counts and differential expression results (CSV)

Samples (n)	Longitudinal (n)	MS (n)	TM (n)	NMO (n)	ADEM (n)
3,148	468	1,745	169	372	32
114	40	0	0	114	0
654	179	527	53	9	3
3,148	468	1,745	169	372	32
1,069	220	1,069	0	0	0
32	2	0	16	16	0
2,217	388	1,397	136	107	22

939	330	657	123	41	19
123	10	83	0	0	0
125	16	125	0	0	0
58	6	0	0	58	0
45	21	31	0	0	0
100	45	80	0	0	0
219	14	112	64	29	14

45

6

33

0

0

0

CIS (n)	ON (n)	Controls (n)
88	16	726
0	0	0
22	3	37
88	16	726
0	0	0
0	0	0
47	11	497

84	14	1
0	0	40
0	0	0
0	0	0
0	0	0
3	0	11
0	0	20
0	0	0

