NIH Intramural

# Whole Genome Sequencing in Multiple System Atrophy

**MODIMSA Meeting**

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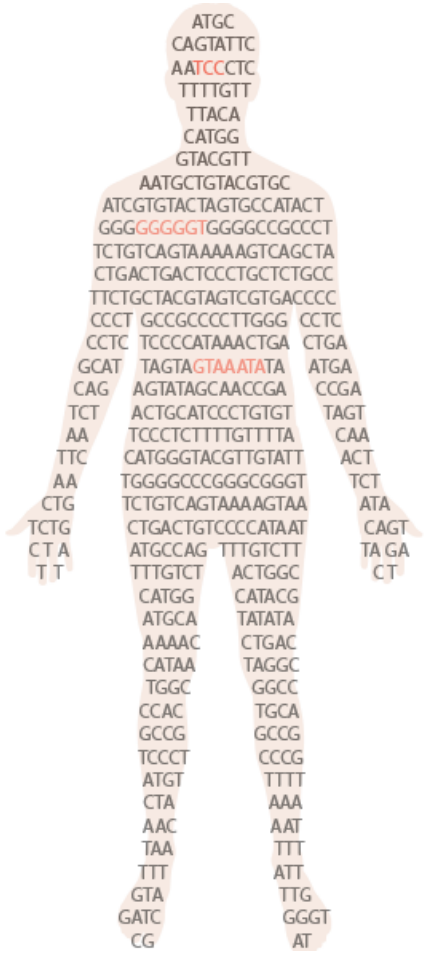
# Overview




## NIH Genome Sequencing Projects:

a) Lewy body dementia (LBD) (n=3,000; ongoing)

b) Neurologically Healthy, Aged Controls (n=2,000; ongoing)

c) Multiple system atrophy (MSA) (n=1,000; start: 2018)

## Objectives:

a) To extend gene discovery efforts to the broader synucleinopathy spectrum

b) To generate a genomic resource that is openly accessible to the research community

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# Genome Sequencing


- **We would like to request sample contributions**

- **Inclusion criteria:**
  - **European-ancestry MSA patients**
  - **<u>Definite or probable</u> MSA per Gilman criteria**

- **Exclusion criteria:**
  - **if de-identified genomes <u>cannot be shared</u> via dbGaP and AMP-PD**

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# Genome Sequencing


## Samples:

- 3 µg of DNA/patient required
- Alternatively, 3 mL of EDTA blood, saliva, or brain tissue are also acceptable

## Shipping:

- paid by NIH

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# Genome Sequencing

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Cohorts | Library Prep. | Sequencing | Google Cloud | Alignm. QC | Analysis

**State-of-the-art sequencing**

**Illumina X10**
(150 bp, paired-end, 30x coverage)
(single library, single lane clustering)

**Timeline:**

- Sample recruitment: started Sept. 2018
- Sequencing: summer 2019
- Analysis/release of data: early 2020

**All genomes will be available at AMP-PD, dbGaP at no cost**

**Contributors will be named authors on resulting publications**

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# Contact

- If you are interested in participating or have any questions, please contact


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