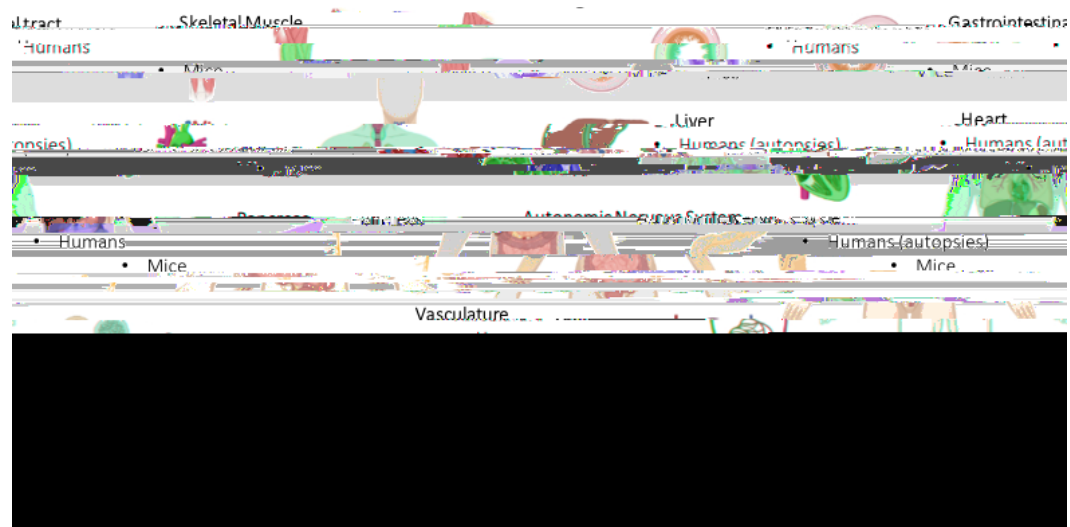


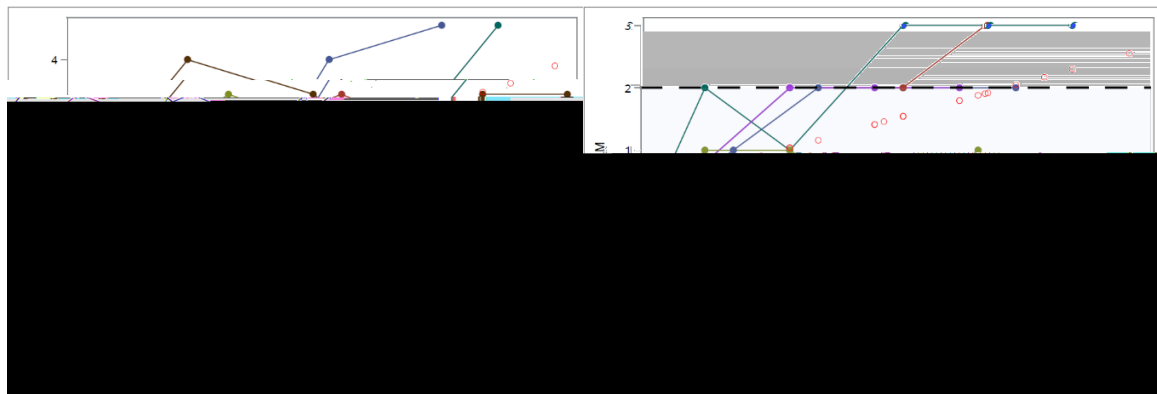


2. Discover important tissue-intrinsic and systems-wide signaling and metabolic pathways which drive disease
3. Discover common signaling and metabolic pathways in more than 1 neurodegenerative disease
4. Investigate non-cell autonomous influences of neuronal degeneration and loss
5. From (1) and (2), investigate novel biomarkers in blood and/or skin to monitor disease progression
6. From (2) and (3), investigate high-yield therapeutic targets for the development of drugs which can be used to treat diseases in more than 1 organ system, and across different neurological diseases.
7. From (4), investigate neurotrophic and tissue growth factors which can act downstream of faulty genes and complement gene therapies.
8. Robust and reliable dissection of peripheral tissue and immune responses in neurodegeneration

We use a range of techniques to investigate the basic molecular biology of disorders of the muscle, neuromuscular junction, nerve fibers, neurons, spinal cord and brain and the clinical implications:

1. Clinical trials and observational studies

Motor Assays in Adult Patients with SMA over Time on Nusinersen Treatment



2. Electrophysiology including nerve conduction studies and electromyography
3. Neuromuscular ultrasound and neurohistopathology
4. High throughput screening using systems wide transcriptome and proteasome analysis
5. Robust dissection of phenotypes using spatial transcriptomics and single cell RNA-Seq
6. Preclinical modeling involving patient iPSC derived cell/tissue cultures and mouse models
7. Functional knockdown and repletion models using antisense oligonucleotides

We provide opportunities for people with neuromuscular diseases to volunteer by providing biosamples or enroll in clinical trials:

1. This is a collaboration which helps the translational medicine team to understand their diseases and reach the goal of finding a cure.
2. These biosample gifts will be shared widely to maximize the impact of these gifts.