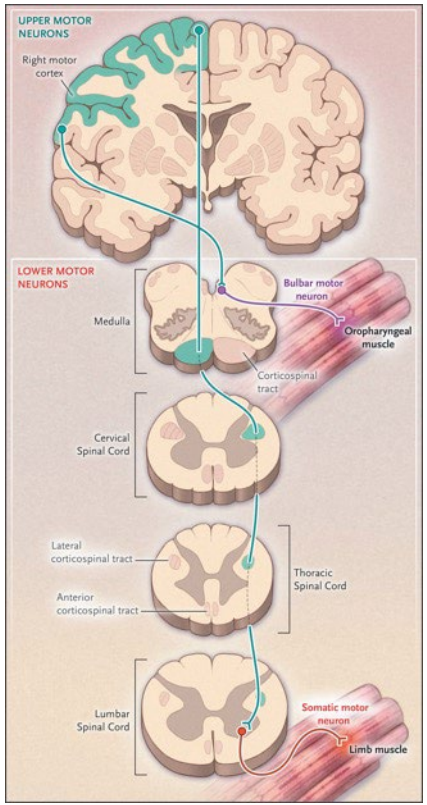
# Excess Sphingolipid Synthesis and Motor Neuron Disease

**Payam Mohassel, MD**
**Associate Professor of Neurology (PAR)**

**Johns Hopkins University School of Medicine,**
**Baltimore, Maryland, USA**

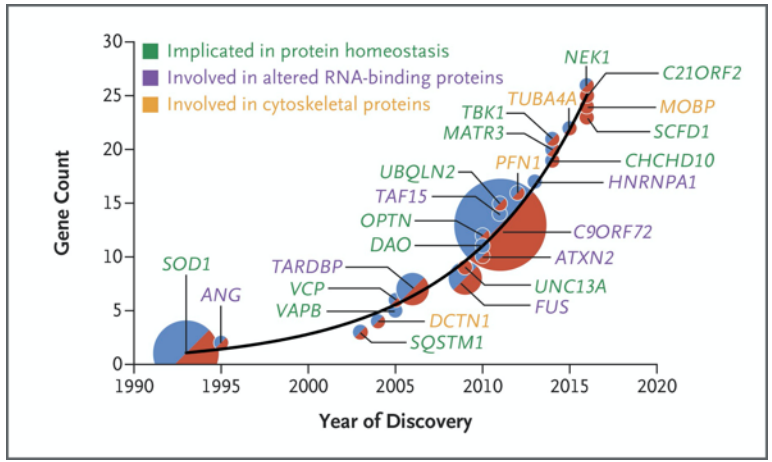
# Juvenile amyotrophic lateral sclerosis (jALS)




**Upper motor neuron:**
Pseudo bulbar affect
Spasticity
Hyperreflexia
Extensor plantar responses
Clonus
Synkinesias
Co-contraction (RAM deficits)

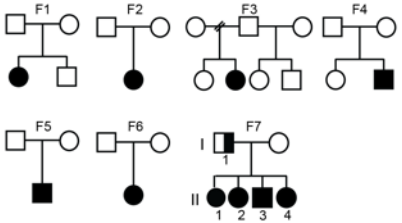
**Lower motor neuron:**
Weakness
Atrophy
Muscle hypotonicity
Fasciculations
Hyporeflexia

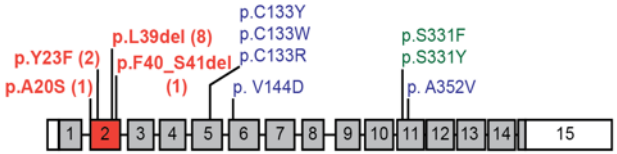
ALS is a **clinical diagnosis**
History, Clinical examination
Supported by electrodiagnostic and other ancillary studies
Sporadic ALS vs familial/genetic ALS
ALS is a neurodegenerative disease

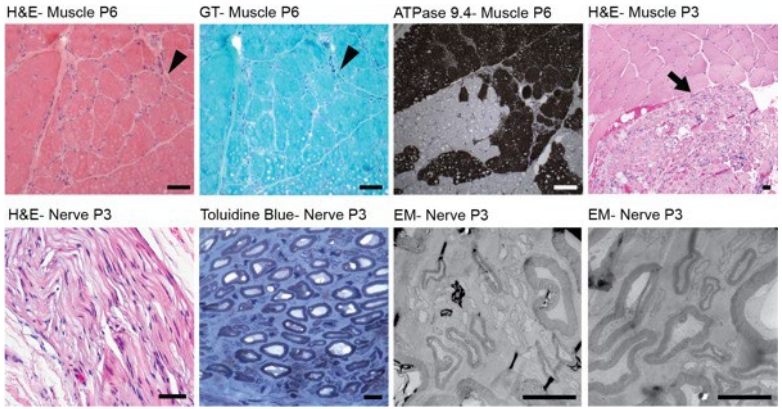


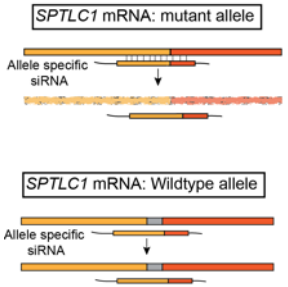

Brown and Al-Chalabi, NEJM, 2017

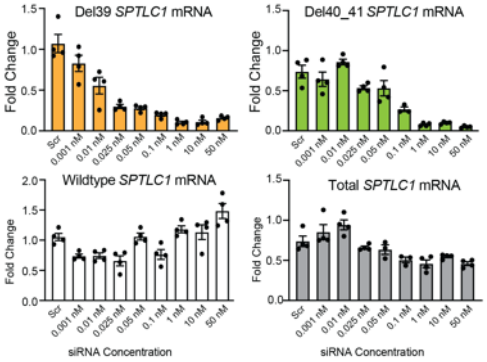
# Gene discovery: a new monogenic form of childhood ALS



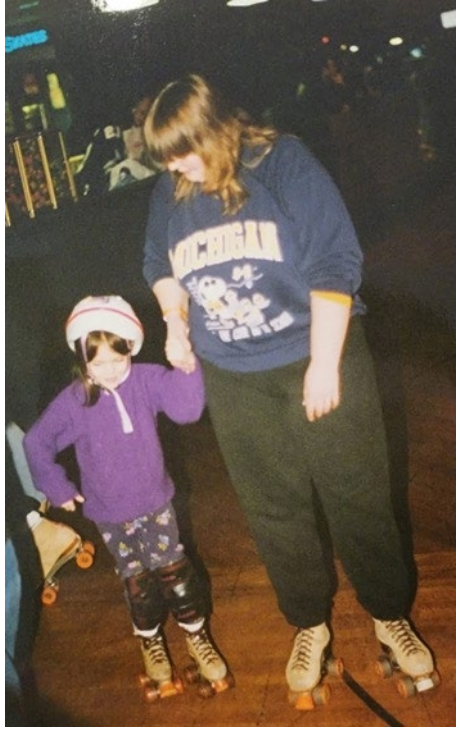




Mohassel, et al Nature Medicine 2021

# Voice of the participant

- Rivka Herzfeld


MICHIGAN

2011

2018


SHARSHERET


GO
EDSO


GOLDEN