# THE JOURNEY TO DIAGNOSIS

## FOR PEOPLE WITH MITOCHONDRIAL DISEASE

A recent study published in *Neurology: Genetics* documented the complex journey to diagnosis for people with rare mitochondrial diseases. Follow the patient journey and learn about some of the common challenges they face as brought to light by the study.

### MOTIVATING SYMPTOM
TO SEEK A DIAGNOSIS

1. **WEAKNESS**
2. **FATIGUE**
3. **DIFFICULTY WALKING**
4. **DROOPY EYELIDS**
5. **IMPAIRED COORDINATION**

### NUMBER OF CONSULTATIONS
WITH DOCTORS

**PARTICIPANTS CONSULTED AN AVERAGE OF 8 DOCTORS ON THEIR JOURNEY TO A DIAGNOSIS**

### DIAGNOSIS
OF MITOCHONDRIAL DISEASE

**MORE THAN HALF** OF RESPONDENTS REPORTED RECEIVING AT LEAST ONE ALTERNATIVE DIAGNOSIS BEFORE THEIR FINAL DIAGNOSIS

**55%** OF PARTICIPANTS RECEIVED THEIR MITOCHONDRIAL DIAGNOSIS FROM A NEUROLOGIST

**AT 22%, MYOPATHY** WAS THE MOST COMMON MITOCHONDRIAL DISEASE DIAGNOSIS REPORTED BY PATIENTS

### ROLE OF PATIENT ADVOCACY
FOLLOWING DIAGNOSIS

**72%** JOINED A PATIENT ADVOCACY GROUP AS A RESULT OF THEIR DIAGNOSIS

**85%** FOUND PARTICIPATION IN AN ADVOCACY GROUP WAS BENEFICIAL

To learn more, read the full article at Neurology: Genetics.

Stealth BIOTHERAPEUTICS



Source: Diagnostic odyssey of patients with mitochondrial disease. J Grier, M Hirano, A Karaa, E Shepard, JLP Thompson. Neurol Genet. 2018:4(2) e230.