ALS/MND Genetically-Targeted Therapy Acceleration Timeline

DISCOVERY + IDENTIFICATION

1993 — Mutations in the ***SOD1* gene** are discovered as the first known cause of ALS/MND.

2008 — Mutations in the ***TDP-43* gene** are identified as causing familial ALS. In addition, TDP-43 pathology is observed in ~97% of ALS/MND cases.

2009 — Mutations in the ***FUS* gene** identified as causing familial ALS/MND.

2010 — Intermediate repeats in the ***ATXN2* gene** identified as increasing risk of developing ALS/MND.

2011 — Hexanucleotide repeats in the ***C9orf72* gene** identified as causing both ALS/MND and frontotemporal dementia (FTD).

C9orf72 is the most common genetic cause of ALS, in both familial and sporadic patients, and its discovery cemented the concept of genetic pleiotropy playing a role in ALS/MND genetics.

2011-2018 — **Many ALS/MND genes identified.** Importance, biological mechanisms and therapeutic relevance still being evaluated.

17 YEARS

11 YEARS

10 YEARS

7 YEARS

CLINICAL TRIALS

2010 — Multiple clinical trials targeting ***SOD1* mutations** initiated.

2018 — Clinical trial targeting ***C9orf72* repeat expansions** begins.

2020 — Clinical trial targeting ***ATXN2* repeat expansions**, in both patients with the expansion and sporadic ALS/MND, begins.

2021 — Clinical trial targeting ***FUS* mutations** begins. Clinical trial for presymptomatic individuals carrying ***SOD1* mutations** begins.

INTERNATIONAL ALLIANCE OF ALS/MND ASSOCIATIONS