Amyotrophic Lateral Sclerosis (ALS)
Patient Journey

European Reference Network for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)
Patient Advisory Board

MUTATION CARRIERS:
Pre-symptomatic

FOR pALS (people living with ALS)

Early genetic testing, while daunting, can open up access to treatments sooner and provide clearer guidance on future steps. If you or your children wish to have children, in vitro fertilization (IVF) can prevent transmission of the genetic mutation, helping to prevent the disease in future generations. Stay proactive by attending regular checkups, and if you notice any changes, consult a neurologist promptly.

FOR PROFESSIONALS

Emphasize the importance of genetic counseling and early testing. With emerging therapies targeting specific genetic mutations (Tofersen/Qalsody for SOD1 e.g.), testing can be crucial for timely intervention.

ALS IN GENERAL:
Pre-diagnosis

FOR pALS

ALS symptoms vary widely. You might notice foot drop, balance issues, difficulty opening bottles or buttoning clothes, or a muscle twitches in your limbs. Speech may become slurred. Often, loved ones might observe changes before you do. Look out for painless, progressive weakness, and consult a physician promptly. Avoid self-diagnosing; always see a medical professional.

FOR PROFESSIONALS

Onset typically begins between 40 and 70 but varies widely. Quick referral to neurology is essential. ALS symptoms present in different ways, such as:

- Bulbar Onset: Slurred speech, swallowing difficulties, tongue twitches, shortness of breath.
- Limb Onset: Foot drop, Hand weakness, Muscle cramps.
- Cognitive Symptoms (15% of cases): behavioral changes and FrontoTemporal Dementia (FTD).

STEP 1
DIAGNOSIS

FOR pALS

Reaching a diagnosis can be lengthy and may involve initial misdiagnoses. Bring a supportive person to your consultation to help process information. A second opinion and further follow-up from a neuromuscular reference center is advisable. These centers offer a MultiDisciplinary Team (MDT) for a comprehensive approach.

FOR PROFESSIONALS

The ALS diagnosis is a clinical diagnosis that requires extensive exams (EMG, MRI, blood tests) with an average delay of 8-12 months from symptom onset. Awareness of diagnostic challenges is critical.

STEP 2
WHAT NOW?

FOR pALS

Avoid comparing your situation to others' stories online, as worst-case scenarios are often highlighted. Every ALS journey is unique. Allow yourself time to process and consider joining support communities, which can offer connection but should not replace professional medical advice.
Some people may react with uncertainty or distance; lean on friends, family, and support networks for strength.

FOR PROFESSIONALS

Specialised multidisciplinary centres and patient organisations are there to offer their expertise on all facets of ALS.
Information on participation in studies/research (not only medical but also observational) should be provided. Participation can be considered an opportunity to be part of the solution, to make a difference in the ALS community. People might also worry about their family/children getting sick, passing on the disease. Genetic testing guidance is increasingly important, as it can provide access to relevant therapies and trials.

STEP 3
ALS ON THE MIND

FOR pALS

Feelings of denial, anger, and depression are normal. The unpredictability of ALS can be challenging, but some find it brings hope.
Certain symptoms, like uncontrollable laughter or crying, are not reflective of mental abilities—seek medical support if needed.
Family members and caregivers also need support. Look for resources like support groups, counseling, and educational materials.

FOR PROFESSIONALS

It is important to know that the body is not a reflexion of the mind. Engage directly with the patient, recognizing individual coping timelines. Be mindful of patients' needs and offer tailored psychological support when needed. Some may experience pseudobulbar symptoms, such as uncontrolled laughing, crying, yawning,... Sometimes medication can help.

STEP 4
HOW TO LIVE WITH ALS

FOR pALS

Identify what you can still do, however small. Hope and humor may not seem obvious but can lighten this path. Consider options like voice preservation, eye tracking systems to control a computer, adaptable housing, and organizinglegal affairs, based on your wishes.
You may find support in contact with peers or to become a member of a patient organisation. Take these steps at your own pace.
Your MDT can help you Live your life. Be aware of alternative treatments or therapies outside the regular channels, always discuss them with your team.

FOR PROFESSIONALS

Even though the evolution of ALS is often rather fast, some people live with ALS for a very long time. Try to look at the possibilities and not at the limitations of the patient. It is important to give the right information so people can make their own informed decisions. On average, people only live up to 33 months after diagnosis.

STEP 5
WHAT ABOUT LATER

FOR pALS

Even as physical challenges increase, regular checkups are essential. Your MDT is here not only for medical support but for guidance on practical concerns, too. Increased susceptibility to infections like pneumonia means taking precautions, but social connection is still valuable. Surround yourself with people who give you confidence and who support you.

FOR PROFESSIONALS

Provide early, detailed information about potential interventions (ventilation, tube feeding) to give patients and families time to consider options. A robust professional network ensures consistent support. ALS is a varied and complex pathology, especially in a more advanced stage, therefor always be open to further educate yourself on the subject.

STEP 6
END OF LIFE

FOR pALS

Dignity in Life and Death: open dialogue about end-of-life preferences lightens the burden for all involved. Patient organisations or MDT members can assist with practical, social, and financial guidance. Cultural and religious beliefs may guide these decisions, so communicate them clearly. Decisions can also be made about donations to science (ex. organs, tissue,...). Think about where you want to spend your last moments, at home or in a medical residence, who has to be there,... Some countries have palliative and hospice care and even offer euthanasia.

FOR PROFESSIONALS

Initiate end-of-life discussions timely but sensitively, ensuring patients feel in control of decisions within their legal rights. In this whole process it is important to also have space for the next of kin to be heard and voice their thoughts and concerns. The final decision, however, should always lie with the patient.

STEP 7
AFTERCARE/BEREAVEMENT CARE

BEREAVED

After a period of intense caregiving, the adjustment can feel empty.
Consider these steps:

- Allow yourself time; there is no "right" length for grief.
- Connect with peers or support groups for shared understanding.
- Fill time with activities that bring you peace.
- Seek professional support if needed.

PROFESSIONALS

Offer continued psychological support and follow-up conversations. Discuss genetic testing with family members if appropriate.

To know what support and legal systems apply in your country, contact your specialised medical centre and/or patient organisation:

https://als.eu/
https://www.encals.eu/
https://ern-euro-nmd.eu/group/als/

For more information:

The ERN EURO-NMD is funded by the European Commission under the EU4Health programme (EURO-NMD 23-27 — 101156434 — EU4H-2023-ERN2-IBA)