Results of the Research Agenda Survey – Spierziekten Nederland

1. Introduction

Spierziekten Nederland is the Dutch patient advocacy organization for individuals with neuromuscular diseases in the Netherlands. One of the core objectives of Spierziekten Nederland is to promote high-quality scientific research. Meaningful research cannot be conducted without the input of people living with a neuromuscular disease and their relatives (e.g. parents or partners).

To identify which areas of neuromuscular research are considered most important by people with neuromuscular diseases, Spierziekten Nederland conducted a survey among its members in May and June 2023. In total, 1,058 members completed the questionnaire. The results of this survey are presented in this report.

2. Results

Diagnosis

Muscle diseases	Number of responses
ALS or PSMA	19
(AMC)	2
Chronic idiopathic axonal polyneuropathy (CIAP) or MGUS-associated polyneuropathy	132
Chronic inflammatory demyelinating polyneuropathy (CIDP)	33
Congenital muscle diseases	24
Duchenne or Becker	28
Small fiber neuropathy	91
Friedreich's ataxia	6
FSHD	47
Guillain–Barré syndrome	18
Hereditary motor and sensory neuropathies (HMSN), Charcot–Marie–Tooth disease (CMT), or other hereditary polyneuropathies	120
Hereditary spastic paraplegia (HSP)	54
Inclusion body myositis (IBM)	47
Limb-girdle muscular dystrophy	36
Multiple neuromuscular disorders	35
Metabolic muscle diseases	2
Mitochondrial muscle diseases	16
Multifocal motor neuropathy (MMN)	10
Myasthenic disorders (such as myasthenia gravis [MG] and Lambert–Eaton myasthenic syndrome [LEMS])	75
Myositis	16
Myotonic dystrophy / myotonias	74
Neuralgic amyotrophy (NA)	19
Primary lateral sclerosis (PLS)	12
Post-polio syndrome (PPS)	33

SMA	23
Pompe disease	15
Other	58

The age of the participants

Age	Number of responses
0 - 12 years	30
13 - 18 years	21
19 - 30 years	45
31 - 50 years	198
51 - 65 years	422
Older than 65	466
Other	3

2.1 Diagnosis

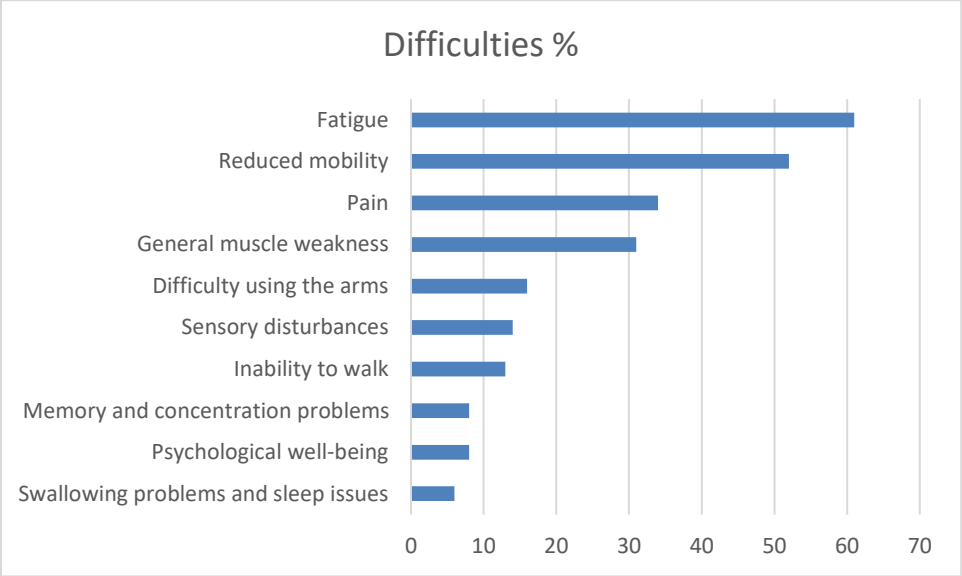
To establish a diagnosis, clinical examination and additional diagnostic tests are required. However, these tests are not always sufficiently sensitive to confirm the diagnosis with certainty. This applies both to the timely and accurate diagnosis of the neuromuscular disease itself and to the timely identification of disease-related symptoms or complications. Participants were therefore asked to indicate which diagnostic research topics they considered most important.

The responses are presented in order of priority, with faster diagnosis of the neuromuscular disease ranked as the highest priority. The priorities were as follows:

1. Faster diagnosis of the neuromuscular disease.
2. Improved assessment and visualization of pain (e.g. through imaging techniques such as MRI) that could be used as outcome measures in future clinical trials.
3. Research into less invasive diagnostic methods (i.e. less burdensome procedures), such as alternatives to muscle biopsy.
4. Other.

2.2 Living with a muscle disease

These symptoms cause the greatest difficulties in daily life for our members with a neuromuscular disease. Participants were allowed to select a maximum of three items.



2.3 Research agenda quality of life

Which quality-of-life research topics do our members consider the most important?

Top 10

1. **Physical activity (research into the effects of exercise and training).**
2. **Fatigue.**
3. **Pain.**
4. **Mobility and personal care.**
5. **Participation in society (education, sports, hobbies, work).**
6. **Maintaining employment**
7. **Alternative therapies (for example, acupuncture, supplements, and osteopathy).**
8. **Psychological impact of the neuromuscular disease / mental well-being.**
9. **Invisibility of neuromuscular diseases.**
10. **Arm and hand function (research aimed at improving and supporting upper limb function).**