Norwegian National Register of congenital and hereditary Neuromuscular Diseases

10-year anniversary!

Conference meeting in Tromsø 06.09.2018

Presentation by
Synnøve Jensen
Consultant neurologist, registry holder
NMK/Nehr, UNN
10-Year anniversary
1440 registered

• Quality register of Norwegian health-services
• Launched 2008
• 1440 (+60) enrolled out of 4000 available.
• 1. registration is professional reported.
  Formal written consent mandatory.
• Follow-up every 5th yr patient reported
• Independent from industry
• Steering commitee: interregional. Neurologists, pediatrician, geneticist.
Register population
Age and gender

Alders- og kjønnsfordeling

Andel (%)

Aldersgrupper

[0,15) [15,30) [30,45) [45,60) [60,75) [75,120]

Menn
Kvinner
3 diagnostic categories

- Myopathies: 68.4%
- Neuropathies: 25.7%
- SMA: 5.8%
ICD-10 diagnoses

1. Muscular dystrophy 35.7%
2. CMT 19%
3. Myotonias 18.4%

Unspecific diagnosis: 89/1441 patients

Molecular confirmation

Specific myopathies 70% (26% missing data)
LGMD: 80% (10% missing)
CMT: 47% (42% missing)
New era

• Mainly collected patients and data. Recruited patients to research projects and Global FKRP Registry.

• In order to measure quality of health care, higher coverage is required.

• Role of the register evolves as new, causal drugs are developed.
AIMS
1. High Quality Health Care

Targets

1. Early diagnosis

2. Molecular confirmation

3. Compliance with key guidelines
   - Follow-up of heart/respiration
   - Treatment

4) High coverage
2. Uniform standard of care

• Benchmarking of hospitals across the nation? Less relevant to rare diseases (RD).
3. Health planning

• Infrastructure in hospitals
  Effective, qualified and targeted work-up?

• Unmet needs of health services?
  Rehabilitation services
  Support in daily life
  Quality of life
Physiotherapy

No, but needed: 25%
Årsak til manglende fysioterapi

- Ukjent (39,6%)
- Annet (26,6%)
- For kostbart (4,4%)
- For lang reisavdel (1,9%)
- Ingen effekt av eksisterende tilbud (19,2%)
- Ikke fysialt bud i kommunen (2,5%)
- Står på venteliste (5,8%)

Hele landet (N=364)
4. Increase visibility of NMD

Together not that rare.
More power to influence health care
Increase awareness and knowledge among professionals.
5. Collect natural history data

Milestones:
- Age of onset
- Loss of walking function
- Ventilatory support
- Cardiac failure

Contribute to natural history studies. Required for
1) Targeted trials
2) Assessment of long-term effect
6. Facilitate clinical trial recruitment

Easily find participants that fit to inclusion criterias:

- Mutation, age, disease stage (ambulant, ventilatory status), treatment status

-> increases the industry’s interest in investing in RD.
7. Role in extension studies/post-marketing surveillance

Monitor
• objective effects
• safety
• patient’s feedback
8. Reach patients suitable for treatment

9. Prevalence overview

10. Empower collaboration and knowledge sharing between countries.
Next step?
• Diagnosis-specific quality measurement (SMA, DM 1, DMD).

• Keep dynamic and responsive to new treatment and guidelines.

• Stepwise implementation of QoL

• National E-Health strategy
  Electronic interactivity registry-patient
  Connection registry-journal?
Thank you for your contribution!