The Newcastle upon Tyne Hospitals NHS
NHS Foundation Trust

THE John Walton MUSCULAR DYSTROPHY RESEARCH CENTRE

Newcastle University

A worldwide study into the prevalence and incidence of spinal muscular atrophy

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TREAT-NMD Neuromuscular Network

Spinal muscular atrophy

- Degeneration of spinal cord motor neurons
- Caused by mutations in *SMN1* gene

SMA type	Age at onset	Highest achieved function	Natural age of death
0	Prenatal/Foetal	Nil	<6 months
I	**<6 months**	**Never sit**	**<2 years**
II	**6-18 months**	**Sit**	**>2 years**
III	**>18 months**	**Stand and walk**	**Adult**
IV	Adult (2nd or 3rd decade)	Walk during adulthood	Adult

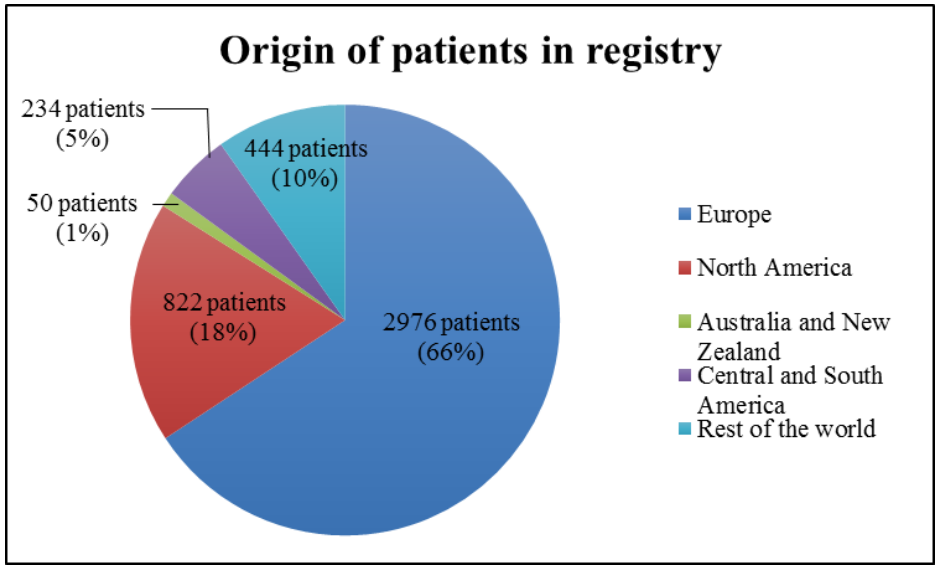
- Prevalence ~1-2 per 100 000
- Incidence 1 in 10 000

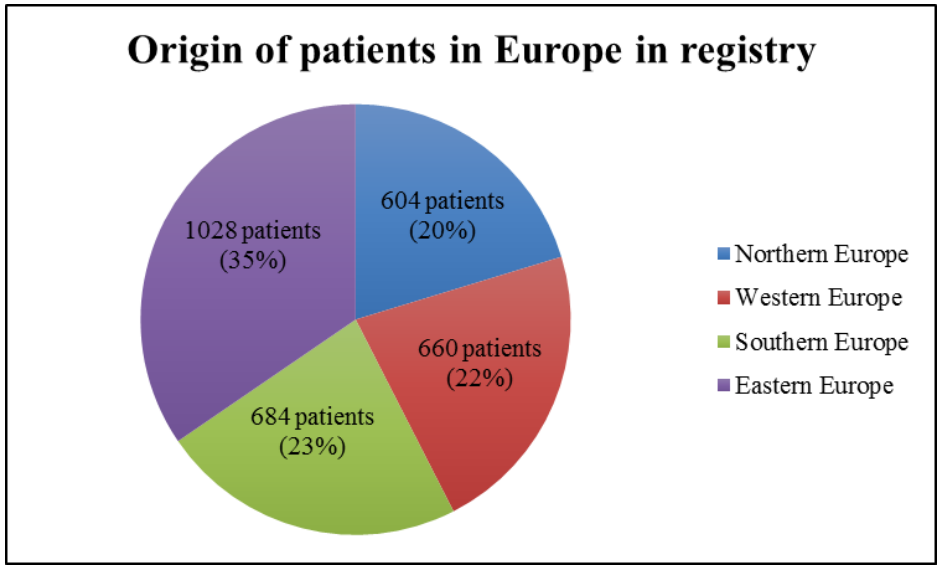
Methods

- Literature review
- **TREAT-NMD Global SMA Registry enquiry**
- **TREAT-NMD Care and Trial Sites Registry enquiry**
- Survey among genetic laboratories

TREAT-NMD Global SMA Registry enquiry (1)

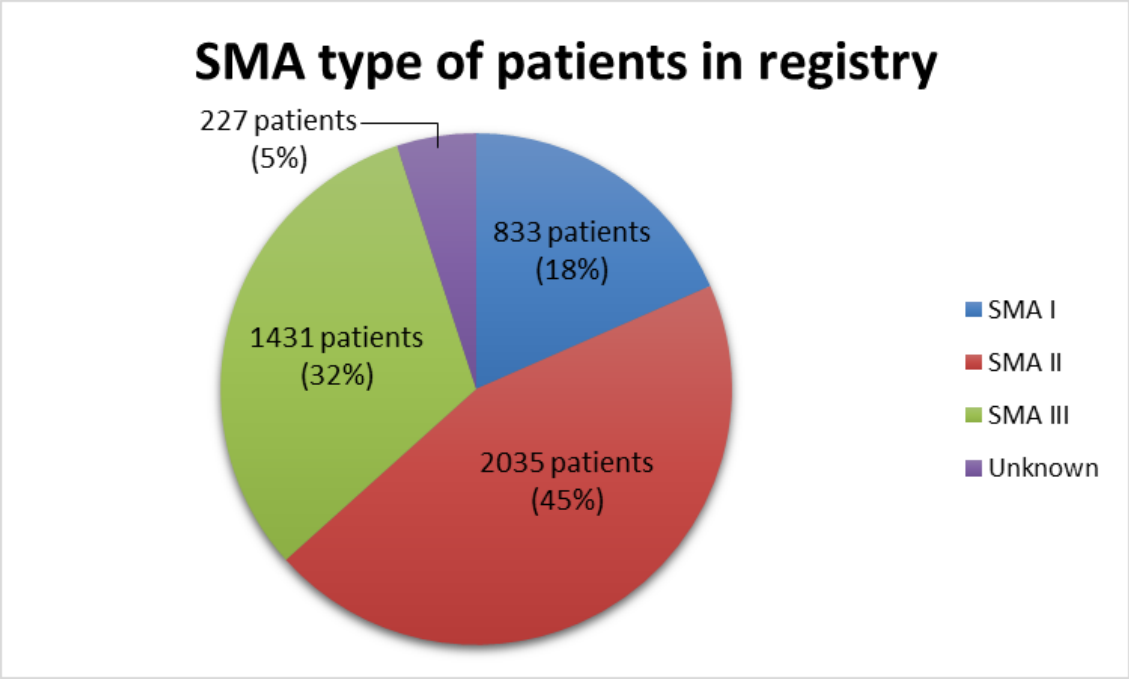
- Response
  - 26 registries (29 countries)
  - Total 4526 patients (1 Sept 2015)





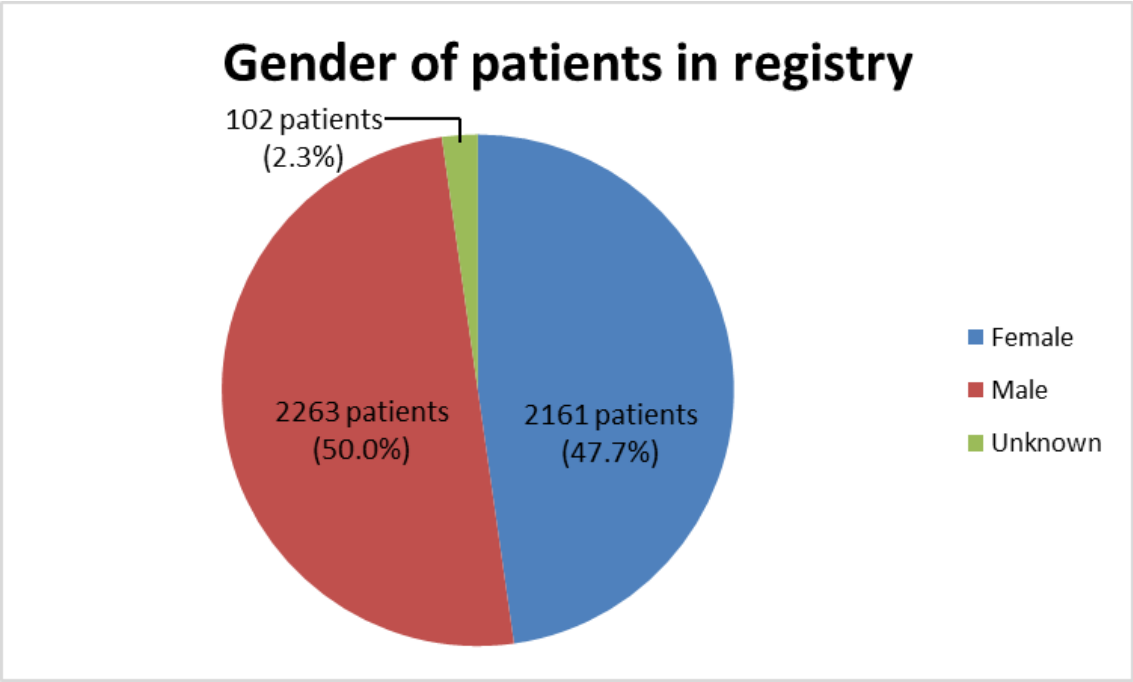
TREAT-NMD Global SMA Registry enquiry (2)

- Majority of patients SMA type II



TREAT-NMD Global SMA Registry enquiry (3)

- Nearly even split male and female

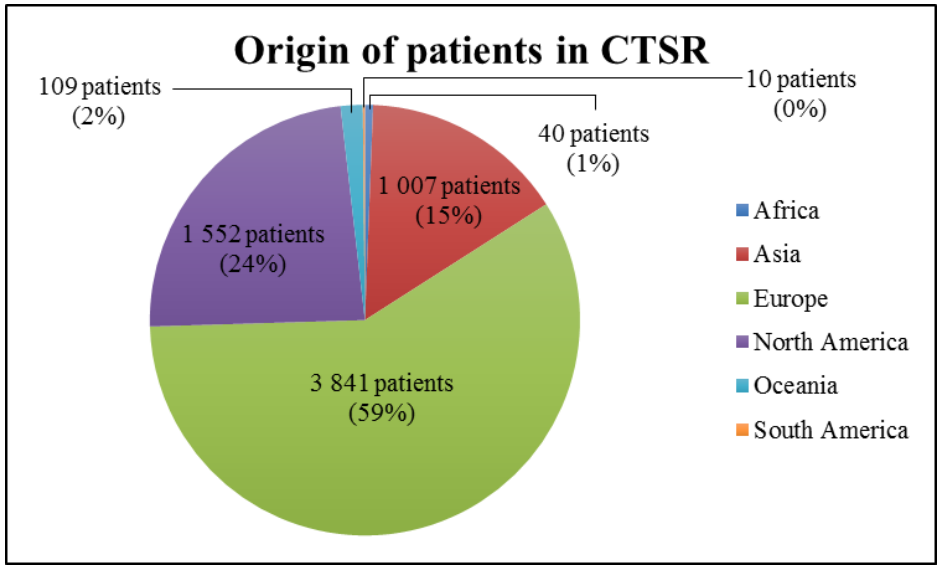


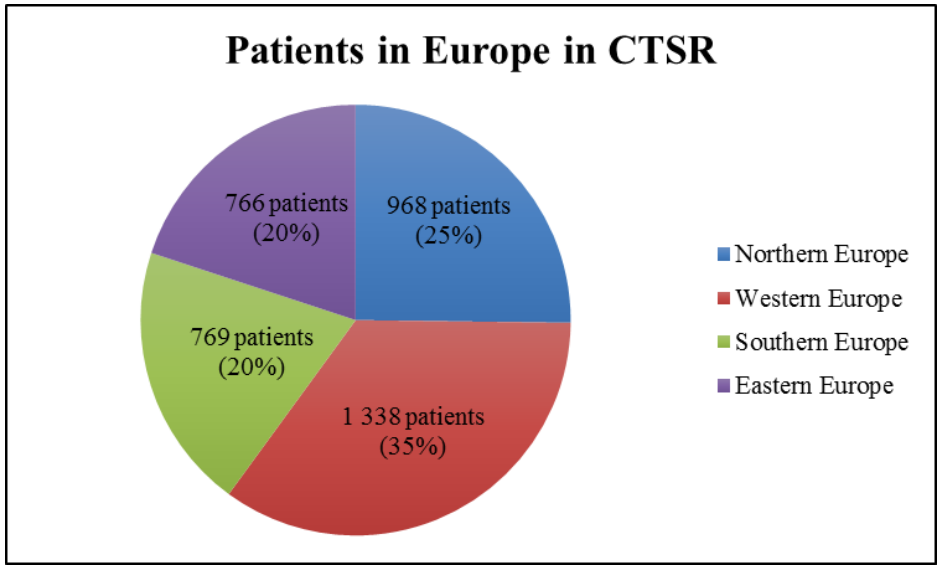
TREAT-NMD Global SMA Registry enquiry (4)

- In general ~2-5 times lower prevalence than literature
- Possible reasons
  - Most literature predates genetic testing
  - Date of registry set-up
  - Genetic diagnosis pending for some patients
  - Purpose of the registry

TREAT-NMD Care and Trial Sites Registry Enquiry (1)

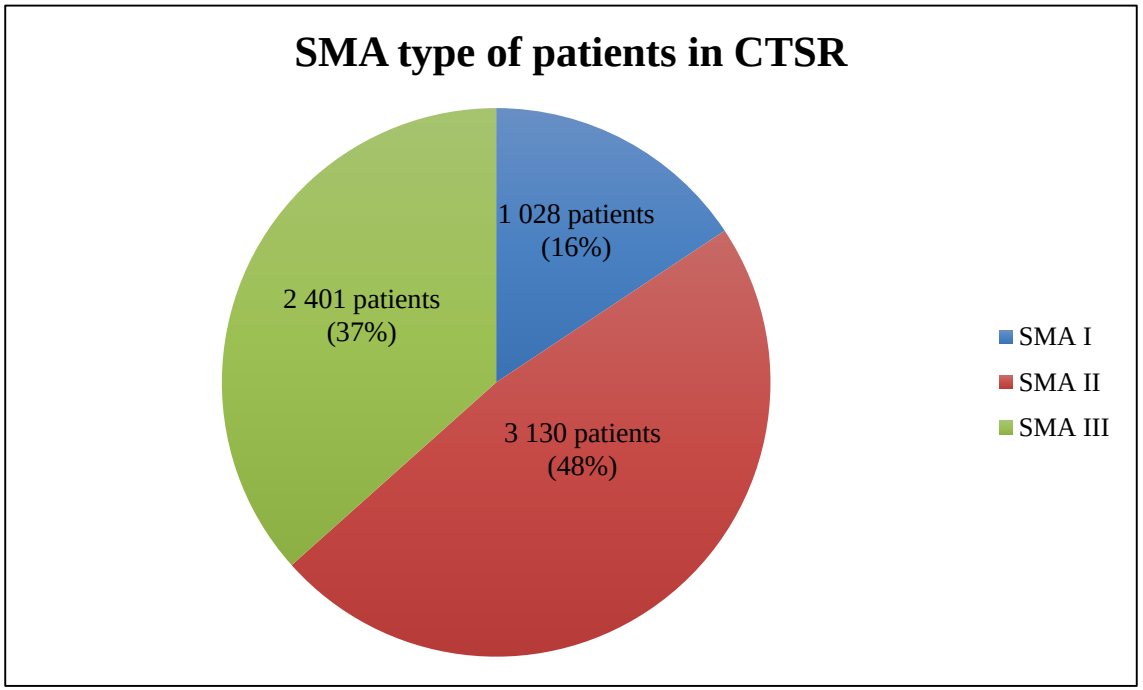
- Response
- Data from 42 countries
  - Total 6559 patients (15 Dec 2015)





TREAT-NMD Care and Trial Sites Registry Enquiry (2)

- Majority of patients SMA type II



TREAT-NMD Care and Trial Sites Registry Enquiry (3)

- Prevalence also lower than literature
  - Differs per country: differences in health system
- Possible reasons
  - Only specialised neuromuscular clinical sites:
    - Not all patients visit/have access to these sites
  - Some patients only clinically diagnosed
  - Some countries patients not identified due to lack genetic testing

Survey among genetic laboratories

- Online questionnaire to 294 laboratories
  (53 countries)
- Response rate variable
  - In total 158 responses (40 countries)
    - 17 countries (fairly) good response rate
      - All in Europe
- Results
  - Incidence around 1 in 5000 to 10 000
  - Generally incidence rates increase when moving from North Europe to the South and the East

Acknowledgements

TREAT-NMD

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Participating genetic laboratories

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