

5th Congress of the European Academy of Neurology

Oslo, Norway, June 29 - July 2, 2019

Teaching Course 17

Congenital myasthenic syndromes and the myotonic dystrophies - diagnostics and possible treatment (Level 3)

Interview with a patient and a caregiver on the impacts of Myotonic dystrophy 1

Kristin Ørstavik
Oslo, Norway

Email: krorstav@live.no

Interview with a patient and a caregiver on the impacts of Myotonic Dystrophy 1

Kristin Ørstavik

Section for Rare Neuromuscular Disorders, Department of
Neurology,
Oslo University Hospital

- Nothing to disclose

The Literature...

American Journal of Medical Genetics 136A:17–24 (2005)

Journal of Neurology (2019) 266:998–1006
https://doi.org/10.1007/s00415-019-09228-w

ORIGINAL COMMUNICATION

Disease burden of myotonic dystrophy type 1

Erik Landfeldt^{1,2} · Nikoletta Nikolenko^{3,4} · Cecilia Jimenez-Moreno³ · Sarah Cumming³ · Darren G. Monckton³ · Grainne Gorman⁶ · Chris Turner⁵ · Hanns Lochmüller^{2,5}

The Patient - Patient-Centered Outcomes Research
https://doi.org/10.1007/s40271-019-00237-y

SYSTEMATIC REVIEW

Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review

Erik Landfeldt^{1,2} · Josefin Edström³ · Cecilia Jimenez-Moreno³ · Baziel G. M. van Engelen¹ · Janbernd Kirschner⁵ · Hanns Lochmüller^{2,5}

J Rehabil Med 2010; 42: 823–830

ORIGINAL REPORT

MYOTONIC DYSTROPHY: THE BURDEN FOR PATIENTS AND THEIR PARTNERS

Reinier Timman, PhD^{1,2}, Aad Tibben, PhD² and Axel R. Wintzen, MD, PhD³

¹From the ¹Department of Medical Psychology and Psychotherapy, Erasmus University Medical Centre Rotterdam, ²Department of Human and Clinical Genetics, Leiden University Medical Centre and ³Department of Neurology, Leiden University Medical Centre, Leiden, The Netherlands

Living With a Hereditary Disease: Persons With Muscular Dystrophy and Their Next of Kin

Josefin Edström^{1,2} and Gerd Ahlström³

¹Sh Institute for Disability Research, Örebro and Linköping universities, Örebro, Sweden
²Rehabilitation Research, Örebro County Council, Örebro, Sweden
³Unit of Caring Sciences, Örebro University, Örebro, Sweden

J Genet Counsel (2015) 24:169–178

DOI: 10.1007/s10887-014-9752-1

ORIGINAL RESEARCH

Understanding the Experience of Myotonic Dystrophy. Mixed Method Study

Amy Osterinn Geirdal · Inger Lund-Petersen · Arvid Heiberg



Archives of Physical Medicine and Rehabilitation
Journal homepage: www.ajpmr.com

Archives of Physical Medicine and Rehabilitation 2019

ORIGINAL RESEARCH

Progressive Decline in Daily and Social Activities: A 9-year Longitudinal Study of Participation in Myotonic Dystrophy Type 1

Kateri Raymond, MOT^{1,2,3,4} · Melanie Levasseur, PhD^{5,6} · Jean Mathieu, FRCP(C)^{1,2,3,4} · Cynthia Gagnon, PhD^{1,2,3,4}

From the ¹School of Rehabilitation, Faculty of Medicine and Health Sciences, Université de Sherbrooke, Sherbrooke, Québec; ²Unité de recherche interdisciplinaire sur les maladies neuromusculaires (UMIRN), Neurologique Clinique, Centre intégré universitaire de santé et de services sociaux de Saguenay-Lac-Saint-Jean, Université, Saguenay, Québec; ³Neurologie Clinique et Réhabilitation, Centre intégré universitaire de santé et de services sociaux de Saguenay-Lac-Saint-Jean, Université de Sherbrooke, Sherbrooke, Québec; ⁴Centre de recherche en Physio-Occupation et Santé, Centre de recherche en santé (CRCS), Centre intégré universitaire de santé et de services sociaux de Saguenay-Lac-Saint-Jean, Saguenay, Québec, Canada.

«Nothing about us without us»



Spouse, mum, widow, medical secretary, author, speaker and part time employee

Torill Knutsen-øy

HAPPILY MARRIED IN 1981



Torill Knutsen-øy

JAN`S DM1 HAND



Torill Knutsen-Øy

HELENE ONLY A FEW YEARS AFTER GETTING THE DIAGNOSIS



Torill Knutsen-Øy

JAN'S OUTGOING SIDE



Torill Knutsen-Øy

TOTALLY EXHAUSTED



Torill Knutsen-Øy

FRAMBU



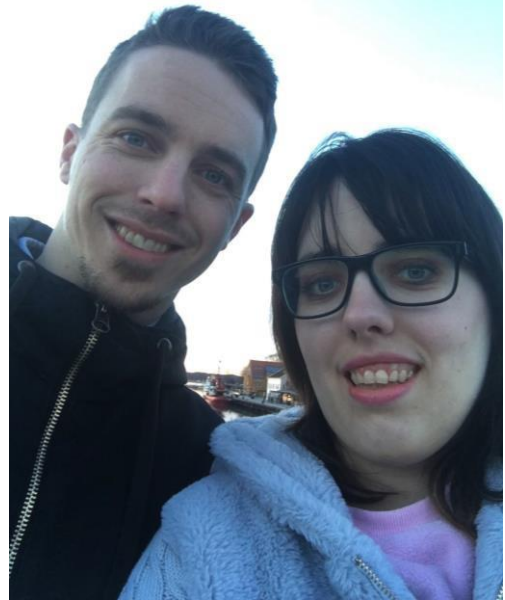
Torill Knutsen-øy

JAN IS DYING



Torill Knutsen-øy

MY 2 KIDS, DAN AND HELENE



Torill Knutsen-Øy

DAN AND HIS PERSONALITY



Torill Knutsen-Øy

MY ADVICE FOR A BETTER LIFE

COMMUNICATE EARLY
ENOUGH

REMEMBER THE WHOLE
FAMILY

USE US NEXT OF KIN

OUR OXYGEN MASK

SEE THE WHOLE PERSON

WHAT ABOUT COMPETENT
TO CONSENT?



Torill Knutsen-Øy

MY BOOK, NAMED "ALONE TOGETHER"



Torill Knutsen-Øy