

# Patient organisations/foundations and research

Barbro Westerholm

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# Federation Rare Diagnoses

About 50 groups of rare diagnoses

Established in 1998

11 000 members

# The Foundation for Research about Muscular Dystrophies

Founded 1986 by  
Christina and Elwyn Mandley

# Duchennes Muscular Dystrophy

- About ten boys diagnosed per year in Sweden
- About 200 boys in Sweden
- Inherited via X-chromosome
- Characterized by lack of dystrophin which results in inability to build muscles
- Symptoms: when the boys are about three years old waddling gait, difficulties to run, jump, rise from the floor.

# Board

- **Chair:**
- Barbro Westerholm
- **Scientists:**
- Anna-Karin Kroksmark
- Thomas Sejersen,
- Már Tulinius, scientist
- **Parent representatives:**
- Ricky Andreis
- Sara Härensten, adj. Ulrik Härensten
- Eric Jernryd
- Fredrik Rundqvist
- Ulf Seger

# Stiftelsen for Research about Muscular Dystrophies

- **Purpose:**
  - to support research about Duchenne`s and Becker`s muscular dystrophies
  - to spread information about the two muscular dystrophies

# Information

- Website – [www.smdf.se](http://www.smdf.se)
- Brochures
  - "All are different, no one is perfect"
  - "Hopp för framtiden" med Stefan Holm
- Seminars

# Seminars

Every second year a two-day seminar is arranged about research and advances about treatment of the boys/young men

- The first day for health care staff
- The second day for parents and boys

Invited lecturers – the most prominent scientists in this field



# Research

- Swedish boys participate in international studies depending on which genetic defect they have
- Register about the boys with type of genetic defect is being established to facilitate research on habilitation, delay of impairment of muscular function, treatment of the genetic defect

# Other activities

- Pay attention to problems like
- Difficulties in getting parking permits
- Waiting for walking aids

# Types of funding

- Support for scientists to participate in conferences, international networks and research projects
- Support for parents to participate in seminars and international meetings
- Establishment of the Muscle dystrophy register

# Where does the money come from?

- Testimonies
- Donations
- Participation in sport arrangements

Eg. Stefan Holm

# Administration of the foundation

- Plusgiro 35 34 90-6 , no 90-konto – too expensive
- County administrative board controls
- No employed staff, auditor obtains a small fee

Thus the work is idealistic