Identifying missing links within rehabilitation services for DM1-patients with adult onset in a Danish population

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BACKGROUND

RCFM has records of 375 persons with genetically verified DM1 (2-81 years),

66 percent have received their diagnosis during adulthood.

We know from experience that many of these patients have been through a variety of complicated and resource-intensive interventions.

RESULTS

Age distribution for 34 patients (13 male/21 female) at time of diagnosis is 21 to 53 years (median 35 years).

24/34 have a spouse and 4/34 have a parent as closest relative. One patient doesn't have any close relative.

11 of 34 (3 M/8 F) have children with CDM1 or juvenile onset DM1.

23 were diagnosed upon genetic workup of a family member, while 32 percent were newly diagnosed.

All were told they had a genetically verified adult onset type of DM1, and would only experience mild symptoms.

We saw big differences in MIRS scores corresponding to 1-4 at first examination. Median for 28 patients was 2.

Median for possible follow-up period (2007-12) was 8 years (5-10 y).

Interventions in functional rehabilitation

59% had a functional analysis and rehabilitation plan at time of referral

30% had ad functional analysis and rehabilitation plan at follow-up:

56% had participated in workshop upon referral

12% had attended follow-up workshops

11% had been consulting the physician

32% had never participated in workshop

AIM

- To characterize a typical course of disease for DM1 patients with adult onset in order to describe their need for functional rehabilitation and interventions.
- To identify challenges and missing links in functional rehabilitation, especially with a view to labor market attachment.

CASE: SEVEN YEARS WITH "MILD" DM1

Male NN, diagnosed at the age of 38. Reading and learning difficulties during childhood, no physical symptoms. Trained as a carpenter. No known family predisposition at time of diagnosis. Married, two children. Works full-time as courier. Symptoms: fatigue and a great need for sleep. CTG-repeats (PCR): approx.68

REFERRED SEP 2009

COURSE OF DISEASE

INTERVENTION

OCTOBER 2009: Referred to RCFM + cardiac and pulmonary evaluation. — Bi-PAP for sleep apnoea

NOVEMBER 2009: Difficulties keeping a full-time job — NN and wife referred to consultation with RCFM's phycisian

APRIL 2010: Wife wants two children examined — Physical examination and rehabilitation plan

2011: Son diagnosed with DM1 — Application for job on special terms rejected, work hours reduced to 29 h

MAY 2012: Family troubles — Wife referred to RCFM's psychologist; Two consultations with physician + RCFM workshop about DM1

2012-2013: Back pain + son has failure to thrive in school — Part time sick leave. Son in respite care

JULY 2013: Discharged from municipal PT — Resumes exercise at local place with help from RCFM

APRIL 2014: Divorces wife — Moves out of family home; shared custody

MAY 2014: Work capacity assessment at job centre — Physical examination and report from RCFM

OCTOBER 2014: NNs brother and sister test positive with DM1

AUGUST 2015: Lack of initiative and fatigue increases

OCTOBER 2015: Receives supplementary disability pension

JULY 2017: Still receives supplementary disability pension. Wish to work 10 hours/week

Sees his children. Troubled relationship with ex-wife.

Tends to cardiac evaluations but never uses Bi-PAP. Does not exercise.."I am OK"!

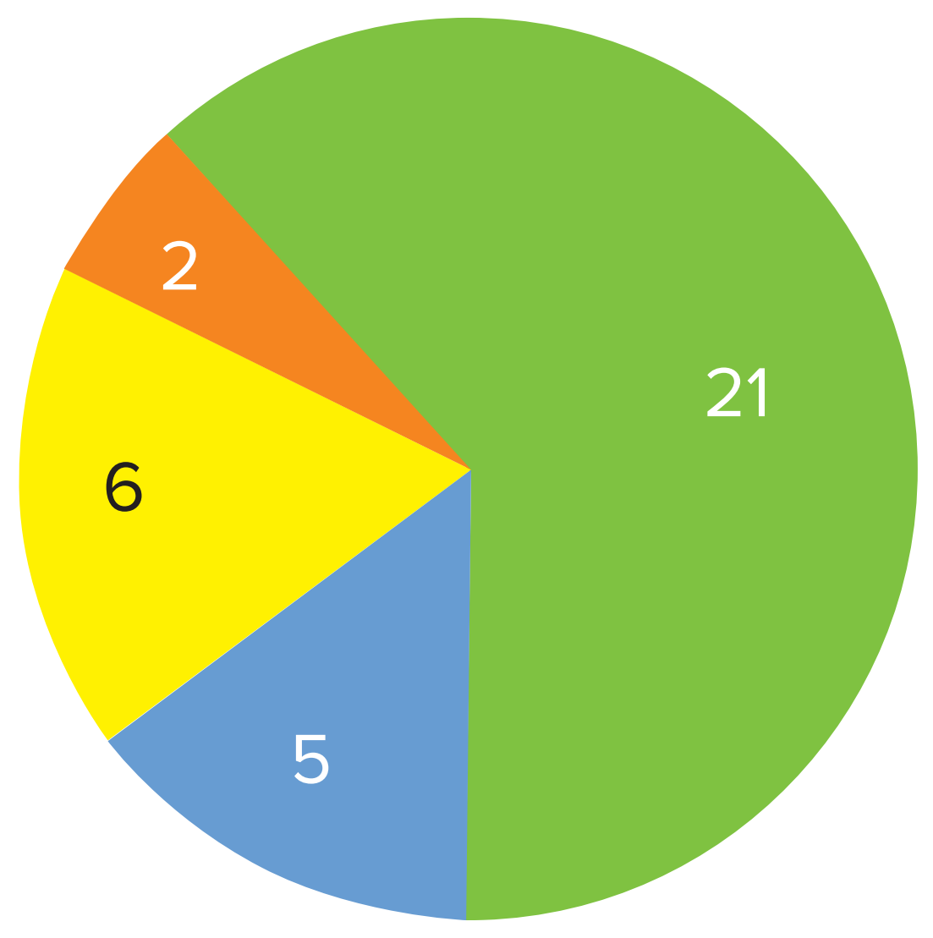
METHOD

We identified 78 patients with adult onset DM1 between 18-65 years (Danish working age) referred to RCFM from January 1st 2007 to December 31st 2012 (with possibility for follow-up after 5-10 years),

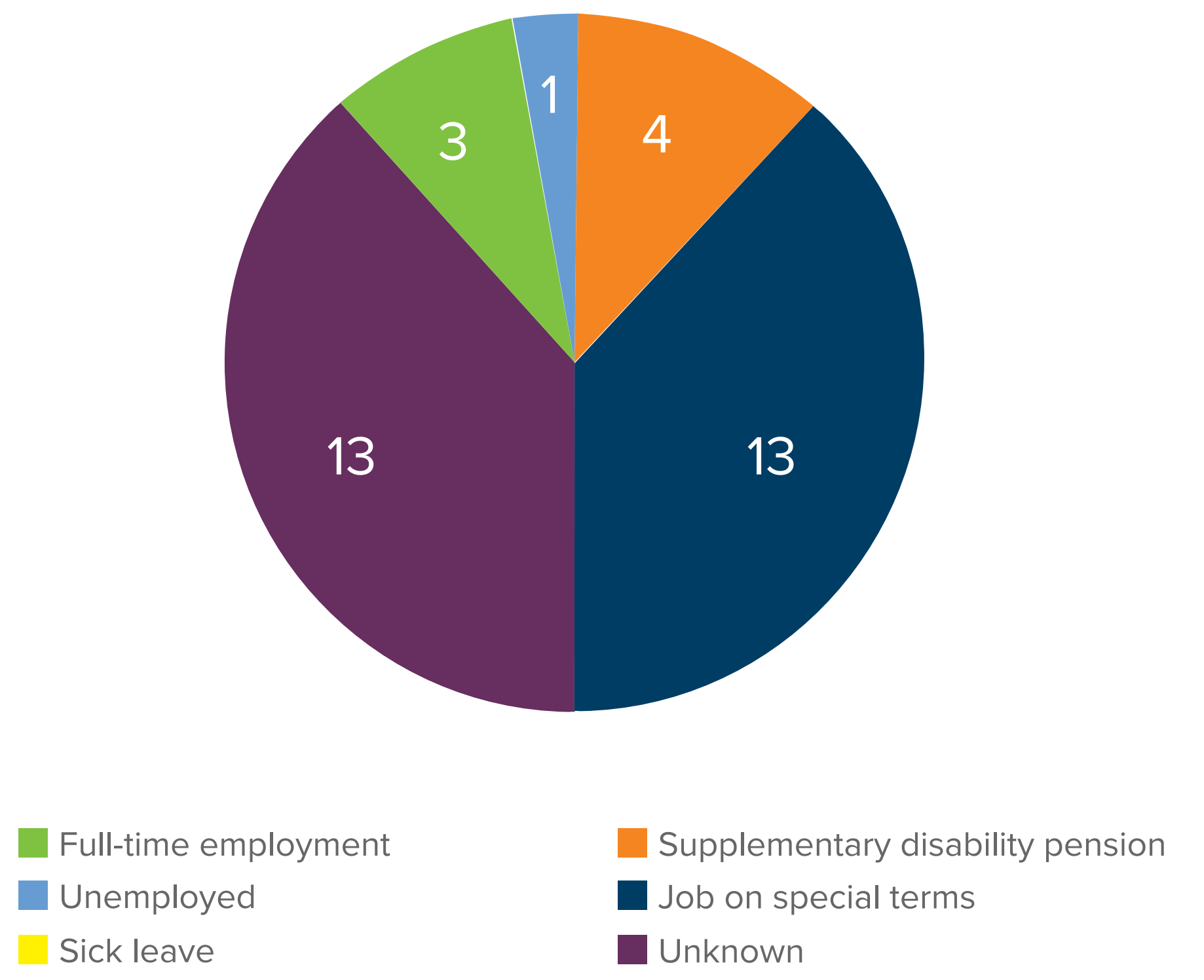
44 were excluded due to age more than 60 years (n=13) and/or referral to RCFM *more than* 1 year after diagnosis (n=31)).

This left us with 34 patients whose courses of disease were identified retrospectively based on their medical records.

Labor market attachment at time of diagnosis



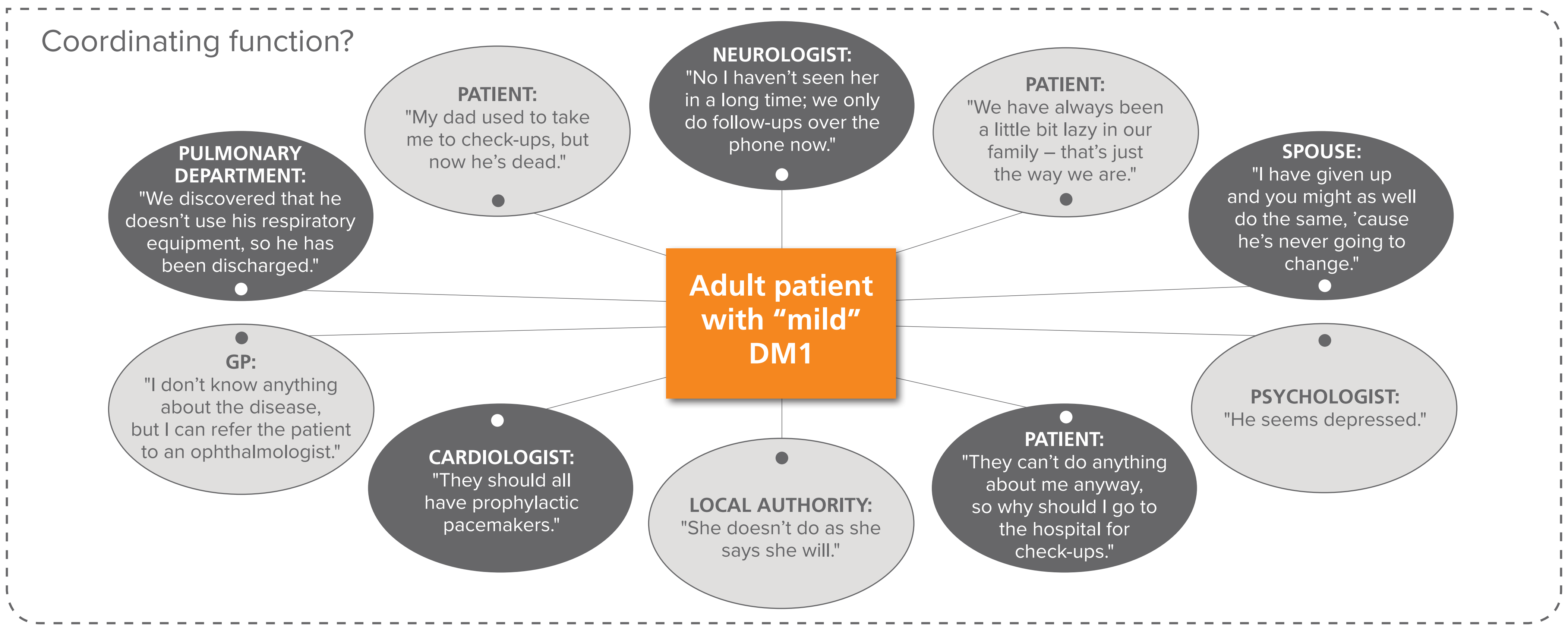
Labor market attachment 5-10 years after time of diagnosis



CONCLUSION

- Our findings indicate that labor market attachment has declined radically 5-10 years after time of diagnosis
- 1/3 of the group was lost to follow-up 5-10 years after initial contact.

POSSIBLE CAUSES FOR DISCONTINUATION OF INTERVENTIONS, - IDENTIFYING THE "MISSING LINKS"



Coordinating function?

Adult patient with "mild" DM1

PULMONARY DEPARTMENT: "We discovered that he doesn't use his respiratory equipment, so he has been discharged."

PATIENT: "My dad used to take me to check-ups, but now he's dead."

NEUROLOGIST: "No I haven't seen her in a long time; we only do follow-ups over the phone now."

PATIENT: "We have always been a little bit lazy in our family – that's just the way we are."

SPOUSE: "I have given up and you might as well do the same, 'cause he's never going to change."

GP: "I don't know anything about the disease, but I can refer the patient to an ophthalmologist."

CARDIOLOGIST: "They should all have prophylactic pacemakers."

LOCAL AUTHORITY: "She doesn't do as she says she will."

PATIENT: "They can't do anything about me anyway, so why should I go to the hospital for check-ups."

PSYCHOLOGIST: "He seems depressed."

TO CONSIDERATION:

- Is the "mild adult type" really that mild?
- How to talk about cognitive deficits?
- No need for follow-up?
- Resignation, depression and/or progression?
- Loss of carers and network?

RCFM
www.rcfm.dk

THE NATIONAL REHABILITATION CENTRE FOR NEUROMUSCULAR DISEASES (RCMF):

RCFM is a national tertiary center of Rehabilitation of excellence for neuromuscular disorders situated between the central hospital services, the local municipal system and the general practitioner. RCFM defines rehabilitation as a collaborative process, which aims to ensure that the individual person with NMD makes the most of his or her physical, mental and social resources.