

Different diagnoses – different challenges in work

Two rare diagnoses with mainly physical limitations

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Challenges of having rare disorders

Rare diagnoses – less than 500 people in Norway

- Difficulties identify people with a rare diagnosis
- Small population – small samples
- Little attention – little interest
- Lack of knowledge – barrierer for receiving health- and social services

Congenital limb deficiency and Marfan syndrome are rare disorders.

Background

- Different diagnosis may cause different challenges in work participation
- Two studies with similar design and questionnaires were conducted at TRS
- Different challenges?
- Need of different rehabilitation programs?

Design and methods for both studies

- Cross- sectional
- Inter-disiplinary project groups
- Similar questionnaire developed in cooperation with the user associations
 - Specific questions related to the diagnoses
 - Standardized instruments
- TRS electronic database

Congenital Limb deficiency (CLD)

- Missing/insufficiently developed skeletal in arms and/or legs
- Severity varies
- Deficiency of one arm is most frequent
- Often isolated and without known causes.
- Blood vessels, nerves may be affected
- Prosthesis and grip-improving devices
- Visible, easy to explain and understand
- Practical/physical challenges



Marfan Syndrome

A genetic connective tissue disorder

Common symptoms:

- Aorta system - Aorta enlargement/
aorta dissection
 - Ocular system (Eyes) – visual impairment
 - Skeletal system – Long arm, legs, feet,
fingers/ scoliosis
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- Life-threatening, affecting many different part of the body.
 - Not visible, the severity varies in periods.



The main findings from study 1 and study 2

- High educational level
- Many had disability pension
- Few worked part-time, received vocational guidance, adaptations before retiring
- Many reported chronic pain and severe fatigue
- Factors significantly associated with reduced work participation
 - Both groups: increased age, lower education level
 - Limb deficiency- chronic pain
 - Marfan syndrome- severe fatigue

Issues related to work participation	Limb deficiency	Marfan syndrome
Disability pension	18%	34%
SWLS mean(SD)	25.7(6.6)	20.4 (7.7)
Severe Fatigue (≥ 5)	33%	41%
Reduced work capacity	22%	50%
Diagnose influence financial situation	36%	52%
Met with understanding at work	62%	77%
Diagnose, main cause for leaving work	68%	95%

Different quotes

- Women with upper limb deficiency
 - *“And as the physiotherapist said:*
 - *I was lucky to have a missing arm to show off, because they then had to realize that the remaining arm was used twice as much, - and it had to last a lifetime”*
- Man with Marfan syndrome
 - *“It is easier to tell about my heart problems, but it is also easy to forget when I look so healthy. It is more difficult to talk about my pain and exhaustion, so I try to appear as healthy as possible”*

Different Diagnoses – Different Challenges

Upper limb deficiency	Marfan syndrome
Visible	Invisible
Not Life-threatening	Life-threatening
The major symptom is constant	The major symptoms vary
Biological physical restrictions	Advised physical restrictions
The healthy arm and vulnerability	The risky body and risky life

Implication for rehabilitation

Upper limb deficiencies

- Young people
- People with increasing chronic pain and fatigue

Rehabilitation programs

- *Assistive devices/ grip-improving devices/prosthesis/ergonomic adoptions*
- Vocational guidance
- Adaptations and time flexibility in work
- Psychosocial support
- Strategies of dealing with overuse and pain

Marfan syndrome

- Young people
- People with increasing chronic pain and fatigue
- *People with aortic dissection/surgery – readjustment of work task or occupation*

Rehabilitation programs

- *Restriction of strain. Intensive training and psysocosial support*
- *Visual aids*, ergonomic adoptions
- Vocational guidance
- Adaptations and time flexibility in work
- Psychosocial support
- Strategies of dealing with fatigue and pain

Conclusions and implications

- The studies indicate that the perceived symptoms of the Limb deficiency and Marfan syndrome give similarities and differences in challenges and adaptation requirements in work participation.
- Visible physical limitations may easily be remedied with physical adaptation and assistive devices
- Adaptions in relation to invisible and fluctuating issues as fatigue and pain, are difficult.
- The particular challenges of different diagnoses are important to take into account when work-rehabilitation programs are developed.
- Is paid work all ?

Limitations of the studies

Several limitations

- Cross sectional design –
- Questionnaire
- Small samle groups - low response rate (52/63%)-
- Only quantitative data
- Contrasting and comparing ?

Implications for further research

- More research of work participation for both groups
- Mixed methods
- Verified diagnoses
- International collaborations



Thank you for
your attention!

Keep on flying.....

Questions and
comments ?

Publication list

COGENITAL UPPER LIMB DEFICIENCY

1. Johansen H, Ostlie K, Andersen LO, Rand-Hendriksen S. Adults with congenital limb deficiency in Norway: demographic and clinical features, pain and the use of health care and welfare services. A cross-sectional study. *Disabil Rehabil.* 2015:1-7.
2. Johansen H, Ostlie K, Andersen LO, Rand-Hendriksen S. Health-related quality of life in adults with congenital unilateral upper limb deficiency in Norway. A cross-sectional study. *Disabil Rehabil.* 2016:1-10.
3. Johansen H, Bathen T, Andersen LO, Rand-Hendriksen S, Ostlie K. Chronic Pain and fatigue in adults with congenital unilateral upper limb deficiency in Norway. A cross-sectional study. Submitted Plos one

MARFAN SYNDROME STUDY

1. Velvin G, Bathen T, Rand-Hendriksen S, Østertun Geirdal A. Systematic review of the psychosocial aspects of living with Marfan syndrome. *Journal of Clinical Genetics* 2015; 87 (2): 109–116
2. Velvin G, Bathen T, Rand-Hendriksen S, Østertun Geirdal A. Systematic review of chronic pain in persons with Marfan syndrome. *Journal of Clinical Genetics* 2016; 89 (6): 647-659
3. Velvin G, Bathen T, Rand-Hendriksen S, Østertun Geirdal A. Work Participation in Adults with Marfan syndrome: Demographic Characteristics, MFS Related Health Symptoms, Chronic Pain, and Fatigue. *American Journal of Medical Genetic* 2015; 167A (12):3082-3090
4. Velvin G, Bathen T, Rand-Hendriksen S, Østertun Geirdal A. Satisfaction with life in adults with Marfan syndrome (MFS): associations with health-related consequences of MFS, pain, fatigue, and demographic factors. *Journal of Quality of Life Research* 2016; 25(7):1779-1790
5. Bathen T, Velvin G, Rand-Hendriksen S, Robinson HS. Fatigue in Adults with Marfan syndrome, Occurrence and Associations to Pain and Other Factors. *American Journal of Medical Genetic* 2014; 164A (8):1931-9.
6. Velvin G, Bathen T, Rand-Hendriksen S, Østertun Geirdal A. Chronic pain in Adults with Marfan syndrome (MFS): Occurrence and associations to demographic aspects, MFS related health symptoms and fatigue. *Submitted to Journal of Rare Disorders*