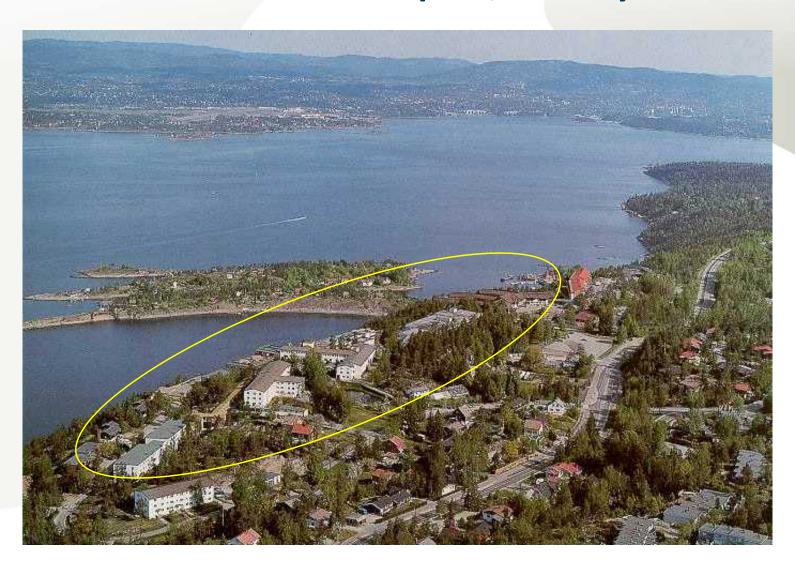


# Different diagnoses – different challenges in work Two rare diagnoses with mainly physical limitations

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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?



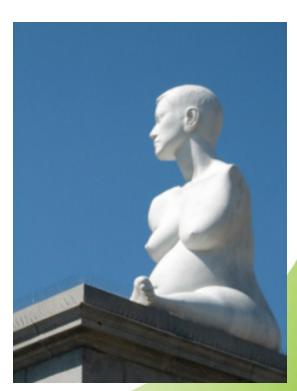
# Design and methods for both studies

- Cross- sectional
- Inter-disiplinary project groups
- Similar questionaire 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, feets, fingers/ scoliosis



- 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, adaptions 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



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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 capasity	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 vulnarbility	The risky body and risky life



# Implication for rehabilition

### **Upper limb deficiencies**

- Young people
- People with increasing chronic pain and fatigue

### **Rehabilition programs**

- Assistive devices/ grip-improving devises/prosthesis/ergonomic adaptions
- Vocational guidance
- Adapations 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

### Rehabilition programs

- Restriction of strain. Intensive training and psysocosial support
- Visual aids, ergonomic adaptions
- Vocational guidance
- Adapations 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

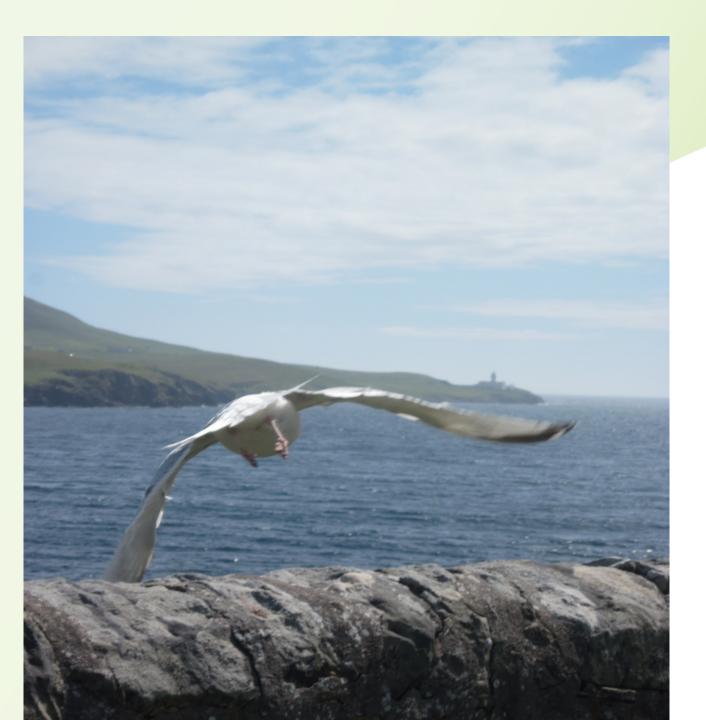
- Cross sectional design –
- Questionnaire
- Small samle groups low response rate (52/63%)-
- Only quantitative data
- Contrasting and comparing?



# Implications for further research

- More reseach 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 DEFIENCY**

- 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 SYNROME 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