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

Gry Velvin- SW/MSc/PhD-Candidate
Heidi Johansen-OT/MSc
TRS National Resource Centre for Rare Disorders.
Sunnaas Rehabilitation Hospital, Norway
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, 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, 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
<table>
<thead>
<tr>
<th>Issues related to work participation</th>
<th>Limb deficiency</th>
<th>Marfan syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disability pension</td>
<td>18%</td>
<td>34%</td>
</tr>
<tr>
<td>SWLS mean(SD)</td>
<td>25.7(6.6)</td>
<td>20.4 (7.7)</td>
</tr>
<tr>
<td>Severe Fatigue (&gt;5)</td>
<td>33%</td>
<td>41%</td>
</tr>
<tr>
<td>Reduced work capacity</td>
<td>22%</td>
<td>50%</td>
</tr>
<tr>
<td>Diagnose influence financial situation</td>
<td>36%</td>
<td>52%</td>
</tr>
<tr>
<td>Met with understanding at work</td>
<td>62%</td>
<td>77%</td>
</tr>
<tr>
<td>Diagnose, main cause for leaving work</td>
<td>68%</td>
<td>95%</td>
</tr>
</tbody>
</table>
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

<table>
<thead>
<tr>
<th>Upper limb deficiency</th>
<th>Marfan syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Visible</td>
<td>Invisible</td>
</tr>
<tr>
<td>Not Life-threatening</td>
<td>Life-threatening</td>
</tr>
<tr>
<td>The major symptom is constant</td>
<td>The major symptoms vary</td>
</tr>
<tr>
<td>Biological physical restrictions</td>
<td>Advised physical restrictions</td>
</tr>
<tr>
<td>The healthy arm and vulnerability</td>
<td>The risky body and risky life</td>
</tr>
</tbody>
</table>
Implication for rehabilitation

Upper limb deficiencies
- Young people
- People with increasing chronic pain and fatigue

Rehabilitation 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

Rehabilitation programs
- Restriction of strain. Intensive training and psyscososial 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.
- Adoptions 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 sample 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 ?
COGENITAL UPPER LIMB DEFICIENCY

MARFAN SYNDROME STUDY