Associations between Physical Activity, Strength, Functionality and Quality of life for adults with Achondroplasia

Inês Alves
Maria António Castro
Sofia Tavares
Orlando Fernandes
• Rare bone condition
• Single point mutation at the fibroblast growth factor receptor 3 gene (FGFR3)
• FGFR3 downregulates the proliferation and differentiation of chondrocytes and longitudinal growth of long bones
• Mutation increases FGFR3 signalling, altering proliferation and differentiation of chondrocytes leading to disproportionate short stature

Prevalence 1:25 000 births
Physical impact

- Genu varus
- Joints hyperlaxity
- Hyper lordosis
- Small chest
- Facial hypoplasia
- Macrocephaly
- Small trident hands
QoL in adults with achondroplasia

Comparing to the general population:

- Lower scores in physical measurements
- Psychiatric illness (56% and 3x higher)
Methods

16 adults with achondroplasia (10 women and 6 men)

<table>
<thead>
<tr>
<th>Gender</th>
<th>Age (years)</th>
<th>Height (cm)</th>
<th>Weight (kg)</th>
<th>Fat mass (%)</th>
<th>Waist circumference</th>
<th>Hip circumference</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women (n=10)</td>
<td>34.9 ± 14.6</td>
<td>123.0 ± 11.6</td>
<td>49.3 ± 10.0</td>
<td>27.8 ± 7.2</td>
<td>80.3 ± 9.2</td>
<td>102.0 ± 8.5</td>
</tr>
<tr>
<td>Men (=6)</td>
<td>41.0 ± 11.8</td>
<td>130.0 ± 14.5</td>
<td>61.1 ± 18.5</td>
<td>26.5 ± 15.2</td>
<td>91.7 ± 18.0</td>
<td>100.0 ± 16.2</td>
</tr>
</tbody>
</table>
Results

<table>
<thead>
<tr>
<th>Gender</th>
<th>Handgrip (kg)</th>
<th>6MWT (m)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women (n=10)</td>
<td>9.1 ± 3.5</td>
<td>408.0 ± 57.2</td>
</tr>
<tr>
<td>Men (=6)</td>
<td>15.0 ± 6.4</td>
<td>377.0 ± 123.0</td>
</tr>
</tbody>
</table>

Handgrip (kg) and 6MWT (m) are significantly different between genders (p < 0.001).

Strong correlations:
- Handgrip (kg) with Handgrip (kg): 0.904 (p < 0.001)
- Handgrip (kg) with Waist Circumference: 0.965
- Handgrip (kg) with Fat Mass: 0.907
- Weight with Waist Circumference: 0.904
- Physical activity (MET-min TOTAL/week) with Fat Mass: -0.716 (p 0.002)
Moderate physical activity (MET-min/week) correlates with QoL scoring. MEN scoring higher in all domains with correlation p<0.05.
Predictors

**Fat mass – Physical activity**

- **R: 0.71** (p 0.004)
- **R: 0.750** (p 0.001)
Predictors

QoL pain domain - Physical activity

R: 0.528 (p=0.036)
Conclusion

Physical activity

FAT MASS

QoL
Thank you
orlandoj@uevora.pt