Physical parameters associated with the ability to exercise in women with achondroplasia - A case control pilot study

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Achondroplasia

- Skeletal dysplasia (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
# Achondroplasia Challenges

<table>
<thead>
<tr>
<th>Infants</th>
<th>Children</th>
<th>Teenagers</th>
<th>Adulthood</th>
</tr>
</thead>
<tbody>
<tr>
<td>Delayed Milestones</td>
<td>Back, Joints and Leg Pain</td>
<td>Dental Problems</td>
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<tr>
<td>Lower Muscle Tone</td>
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<td>Compression of the Spinal Cord</td>
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<tr>
<td>Sleep Apnoea</td>
<td>Kyphosis (Outward Curving of the Spine)</td>
<td>Lordosis (Inward Curving of the Spine)</td>
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<td>Increased Fluid Pressure in the Brain</td>
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<td>Elbow Stiffness</td>
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<td>Otitis Media</td>
<td></td>
<td>Bowed Legs</td>
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<td>Obesity</td>
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<td>Psychological Challenges</td>
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</tbody>
</table>
Benefits of physical activity in general population

**Promote**
- physical fitness
- mental health

**Improve**
- quality of life
- resistance
- physiological and biomechanical factors

**Reduce risk**
- metabolic disease
- cardiovascular diseases

QoL in adults with achondroplasia

Comparing to the general population:
- Lower scores in physical measurements
- Psychiatric illness (56% and 3x higher)
Identify physical evaluations that best adapt to adults with achondroplasia.
1. Anthropometric measurements

- Weight
- Standing height
- Sitting height
- Waist perimeter
- Length
  - Arm
  - Forearm
  - Thigh
  - Leg
  - Hand
  - Foot
- Foot width
- Arm span

2. Body composition analysis

- Body fat mass
- Fat free mass
- Lean mass
- Total body water

Tanita MC780-PMA
3. Strength

❖ **Lower limbs**
30 second Sit to Stand Test (30CST)

❖ **Upper limbs**
Hand strength
30 seconds biceps curl (2kg)
30 seconds push-up
1kg chest throw
### Results

<table>
<thead>
<tr>
<th></th>
<th>Active AG (2)</th>
<th>Non-active NAG (2)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Weight (kg)</td>
<td>48.2</td>
<td>55.4</td>
</tr>
<tr>
<td>BMI (Kg/m²)</td>
<td>32.8</td>
<td>44.4</td>
</tr>
<tr>
<td>Waist circumference (cm)</td>
<td>77.8</td>
<td>92.5</td>
</tr>
<tr>
<td>Fat mass (%)</td>
<td>26.9</td>
<td>36.7</td>
</tr>
<tr>
<td>Lean mass (%)</td>
<td>69.3</td>
<td>60</td>
</tr>
<tr>
<td>Biceps curl</td>
<td>22.5</td>
<td>16.5</td>
</tr>
<tr>
<td>Push-ups</td>
<td>15</td>
<td>13</td>
</tr>
<tr>
<td>Chest throw</td>
<td>250</td>
<td>236</td>
</tr>
<tr>
<td>30CST</td>
<td>16.5</td>
<td>11</td>
</tr>
</tbody>
</table>

Mean age 42.3 [22-51]
Correlations

95% CI $p<0.05$
BMI $\leftrightarrow$ waist circumference
BMI $\leftrightarrow$ Fat mass %
BMI $\leftrightarrow$ lean mass %
waist circumference $\leftrightarrow$ hand strength
weight $\leftrightarrow$ push-ups

95% CI $p<0.001$
BMI $\leftrightarrow$ biceps curl

Tendency of association between
physically active
strength
body composition
Thank you
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