Effects of physical activity habits in cardiometabolic health in adults with Achondroplasia

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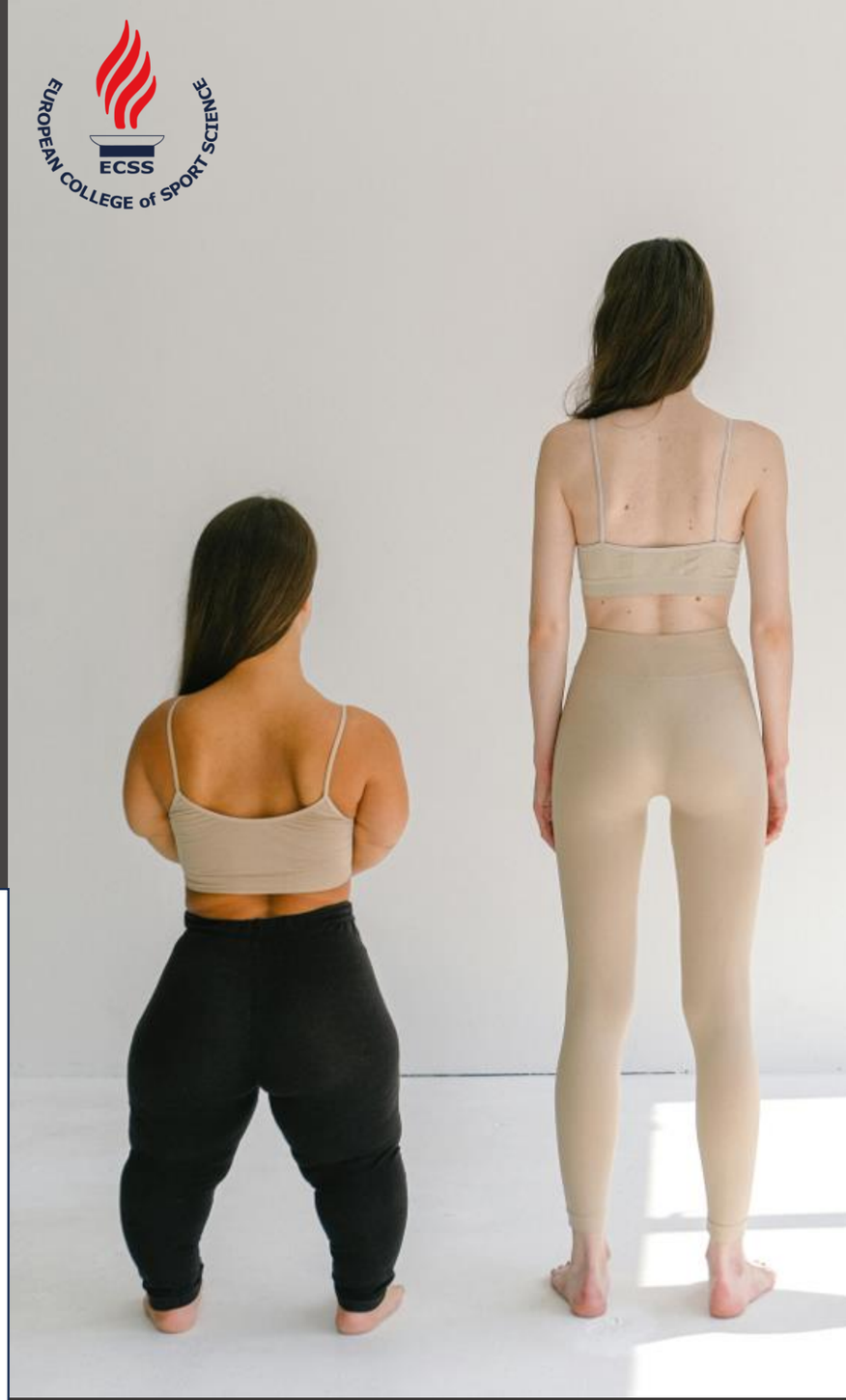
ESTÁDIO CLÍNICA



Achondroplasia

Rare skeletal dysplasia

Caused by a mutation in FGFR3

Prevalence

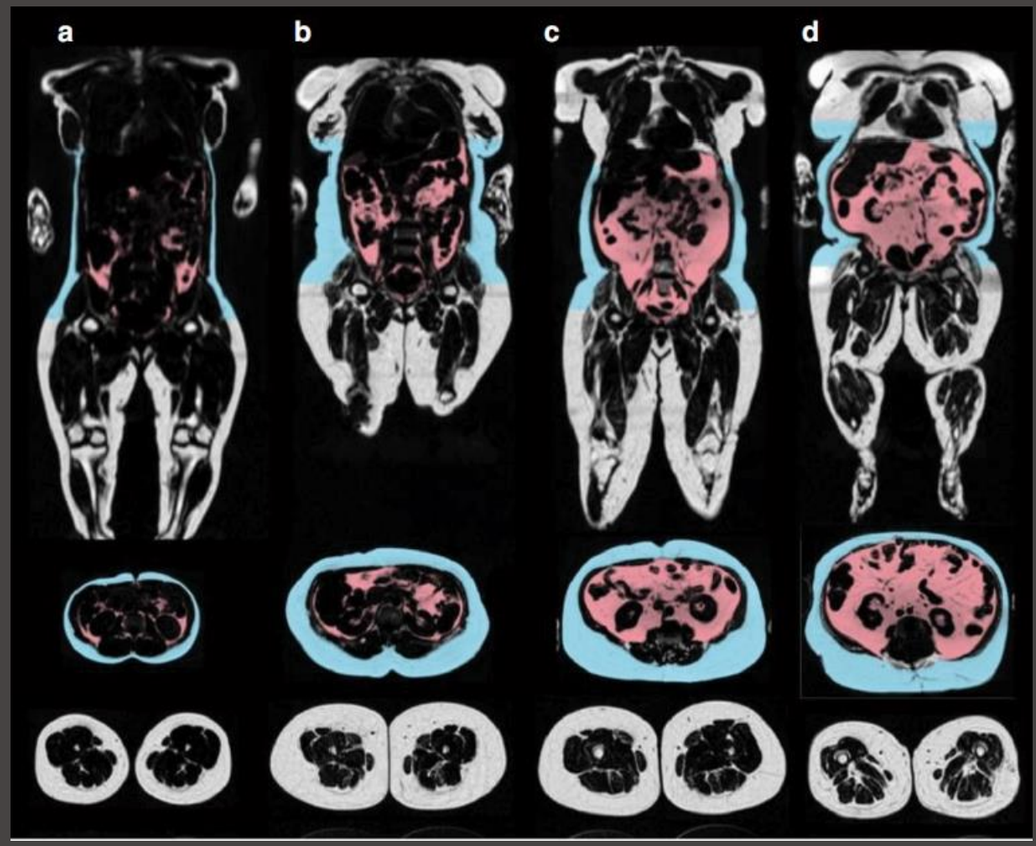
1 in 25 000 births

Skeletal Impact

Disproportionate short stature.
Skeletal deformities, spinal stenosis, joint laxity

Adult height
110 cm - 135 cm

Cardiovascular disease
Obesity [1]

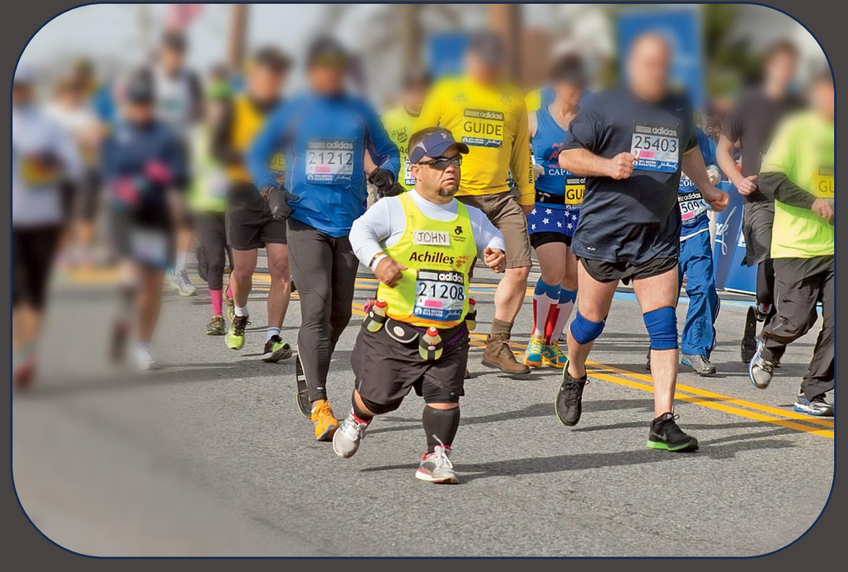


Research Hypothesis

Higher levels of regular physical activity are associated with:

Better physical fitness [2]

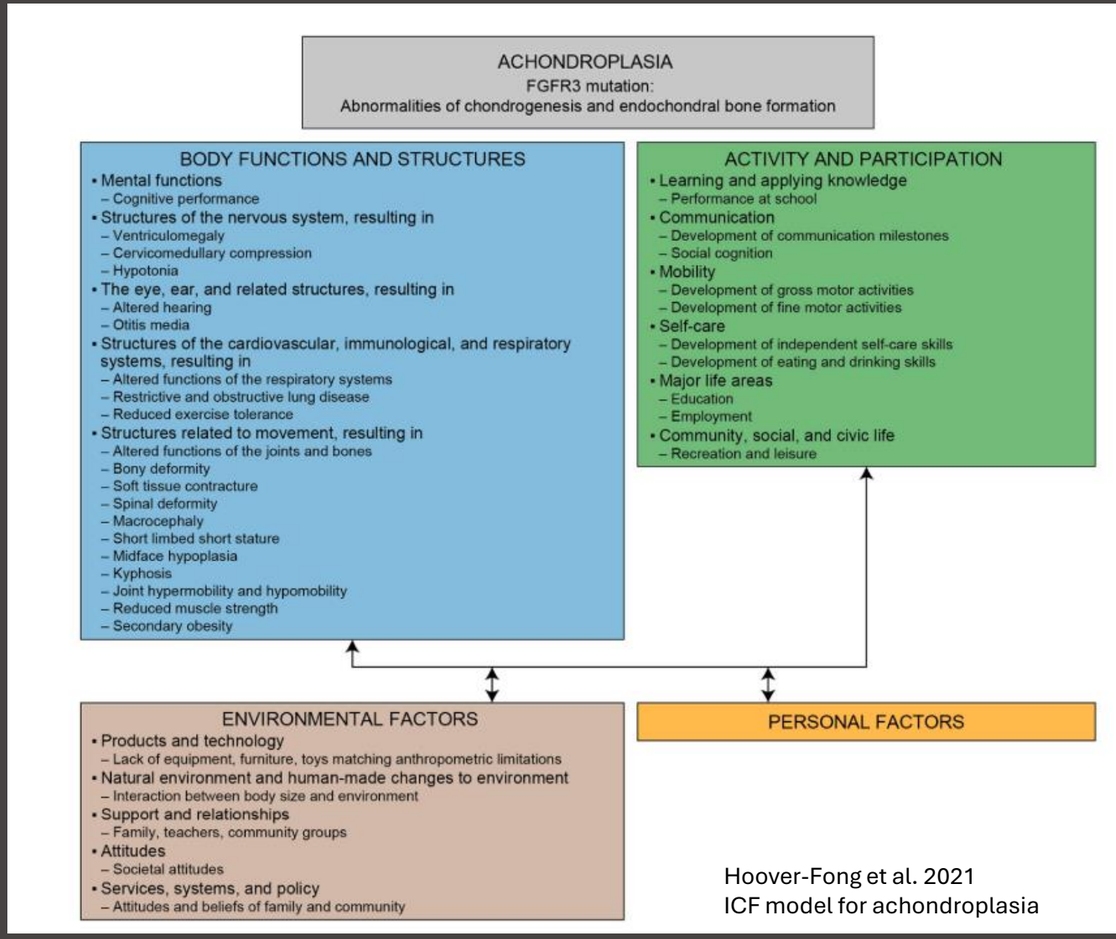
Lower adiposity



Improved cardiometabolic health [3]

Functional diversity / disability

Credits: Tom Shakespeare



Hoover-Fong et al. 2021
ICF model for achondroplasia

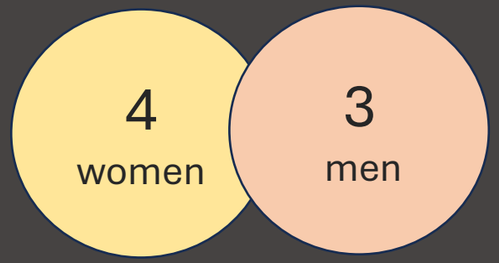
Methods

ASSESSMENTS

- Anthropometrics and body composition
- Physical fitness test (6MWT, handgrip)
- IPAQ questionnaire (PA score and level)
- **Maximal treadmill exercise test**

- VO2max
- peak VO2 (pVO2)
- peak ventilation (pV)
- maximum heart rate (maxHR)
- anaerobic threshold (AT)
- Functional Capacity (FC)

Participants characteristics



	Age (years)	Weight (kg)	Height (cm)	Waist circunf (cm)	Hip circunf (cm)	W/R	Fat mass (%)	Lean mass (%)
Mean ± SD	37.3 ± 11.9	53.7 ± 15.2	129 ± 13.9	83.5 ± 15.6	99.3 ± 11.7	0.839 ± 0.096	24.2 ± 11.7	70.3 ± 14.8

	6MWT (m)	Handgrip strength (kg)	IPAQ - PA score (METs)
Mean ± SD	395 ± 86.4	13 ± 6.41	723±768

Physical activity assessment

METs – multiple of resting metabolic rate

Activity Type	MET Value
Walking	3.3 METs
Moderate Physical Activity	4.0 METs
Vigorous Physical Activity	8.0 METs

Results

Physical activity levels

PAL 1 - Inactive

No exercise, walking only

N = 5

PAL 2 – Minimally active

Lower MET exercises (leisure swimming or slow cycling)

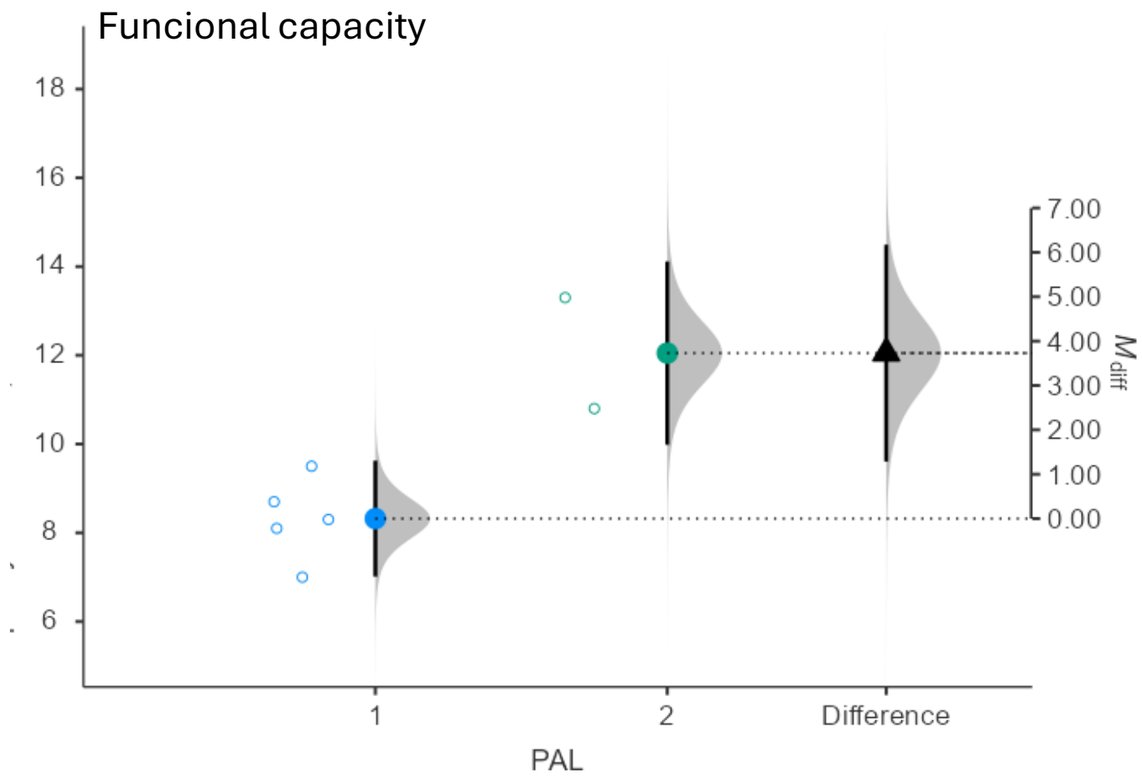
N = 2

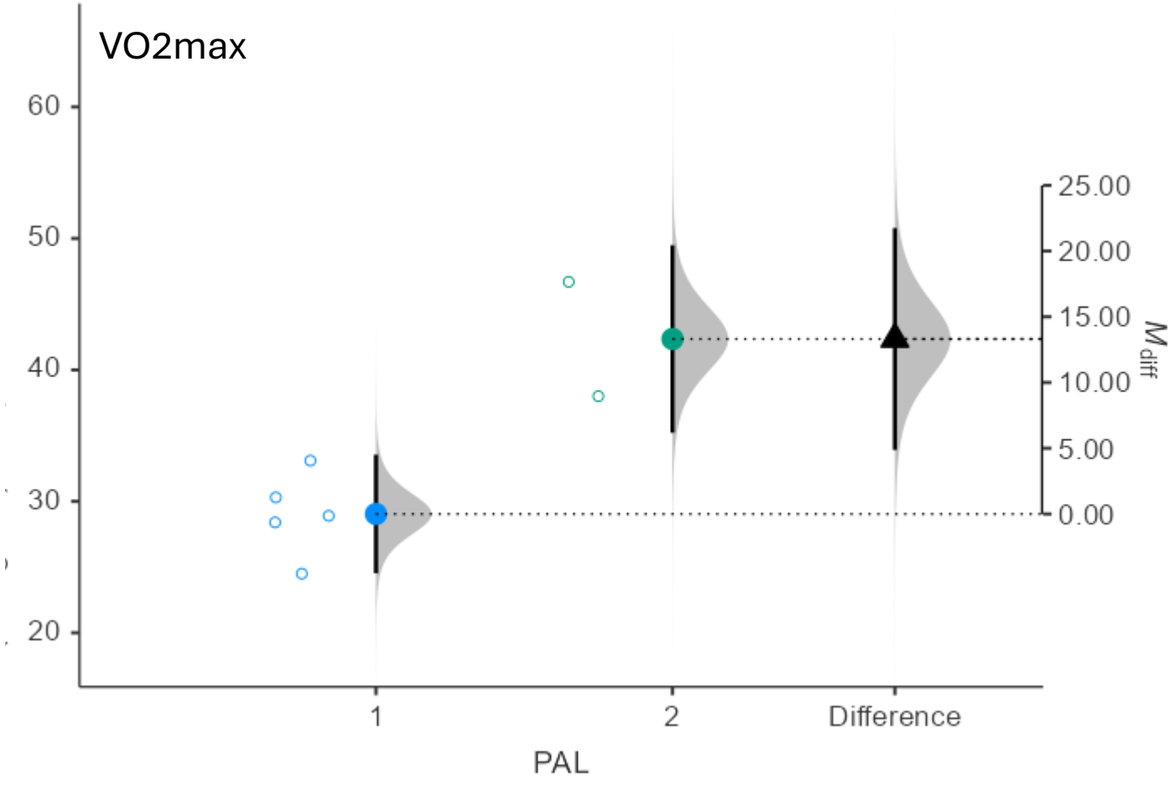
Results

Comparison between PAL1 and PAL2

Significant differences
(p<0.05, effect size >2.1)
in favor of **PAL2**

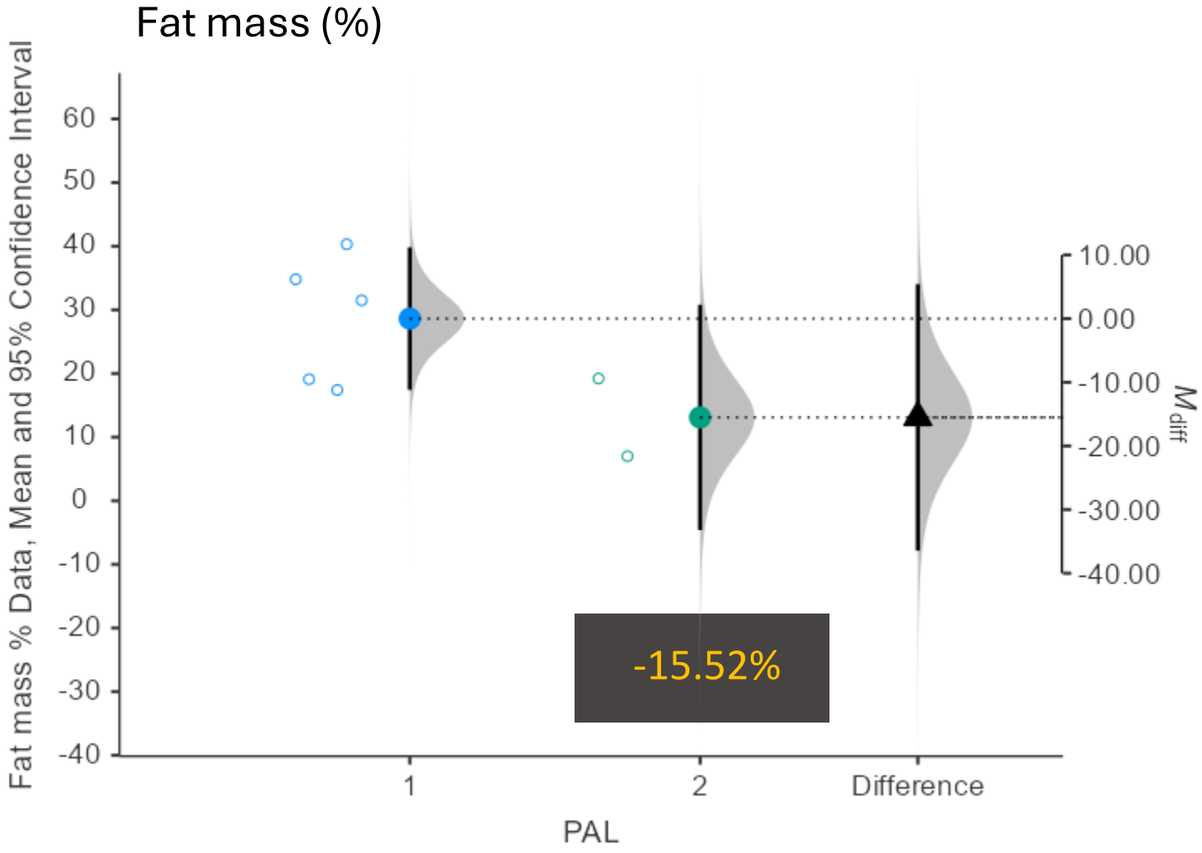
	Functional capacity METS	Peak VO2 mL/min	Exercise capacity mL/kg/min	Aerobic treshold mL/kg/min	6MWT mts	PAS
PAL 1	8.32 ± 0.91	1492 ± 484	29 ± 3.13	20.5 ± 4.26	355 ± 55	355 ± 212
PAL 2	12.1 ± 1.77	2550 ± 223	42.4 ± 6.15	28.7 ± 0.85	497 ±60.8	1642 ± 996

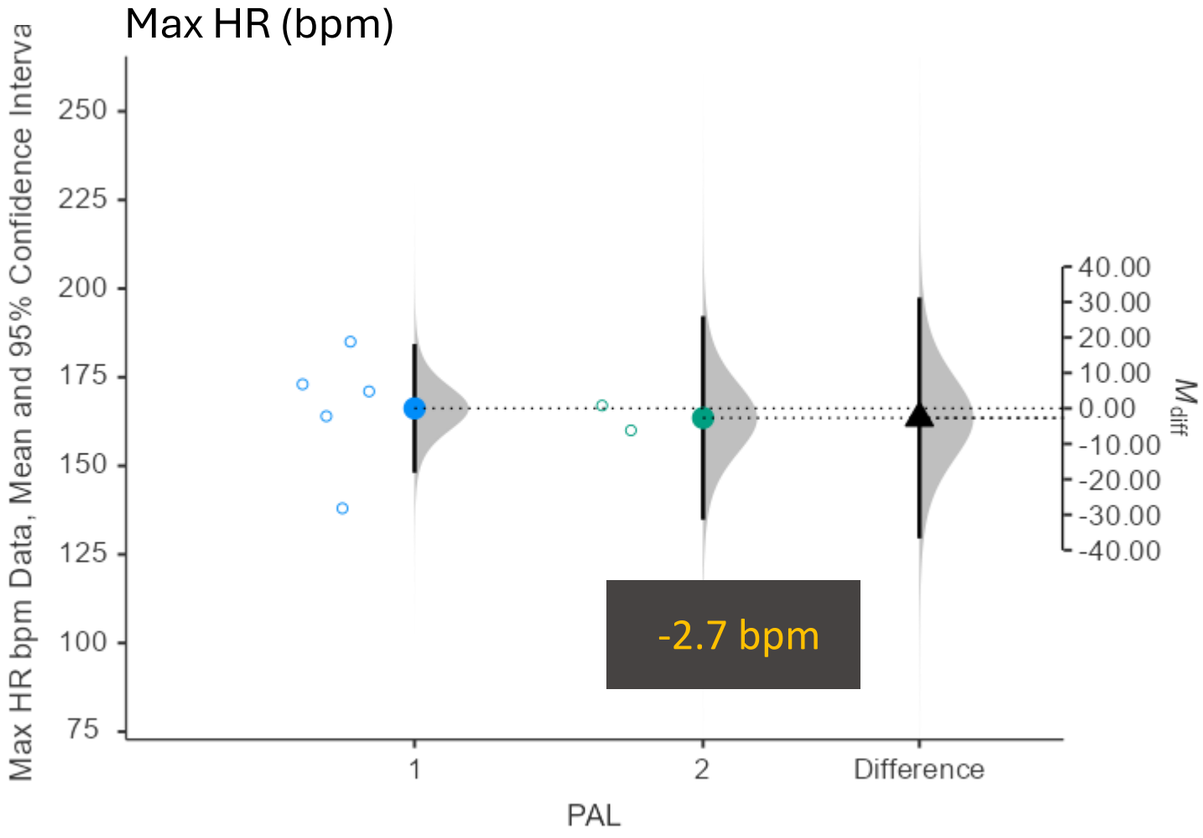




Results

Body Composition and Heart Rate





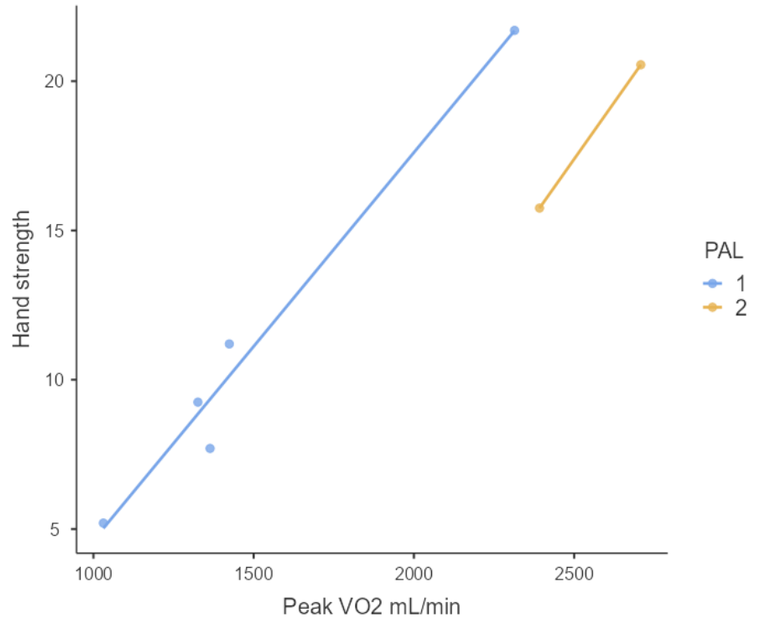
Results

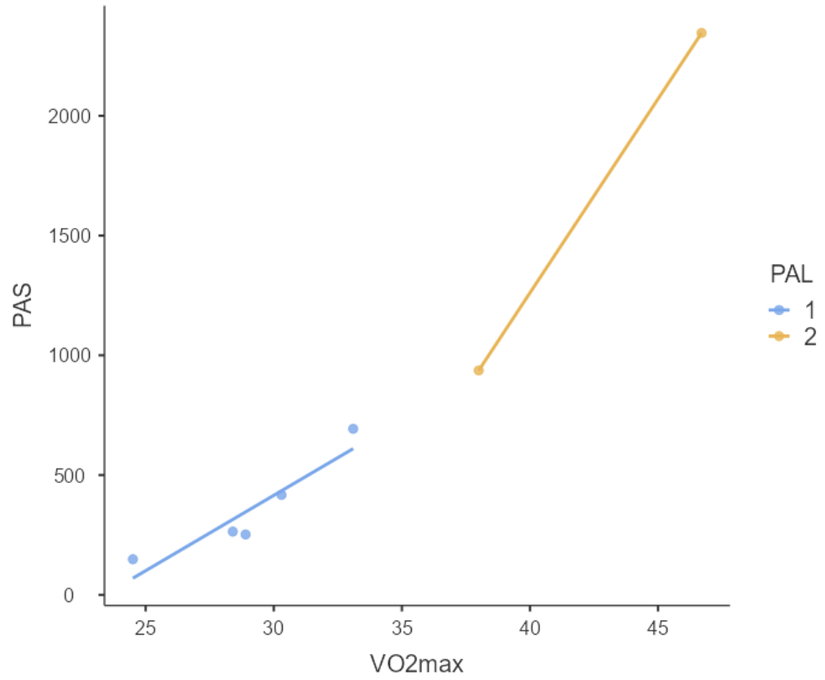
Correlations

- **Strong correlations (p<0.001):**
  - Handgrip strength and peak ventilation (r = 0.955)
  - VO2max and Physical Activity Score (r = 0.967)
  - VO2max and Fat Mas % (r = -0.842)

Other significant correlations (p<0.01):

- Peak VO2 and Handgrip strength (r = 0.939)
- Peak VO2 and Body Weight (r = 0.928)
- Lean Mass and Anaerobic Threshold (r = 0.873)

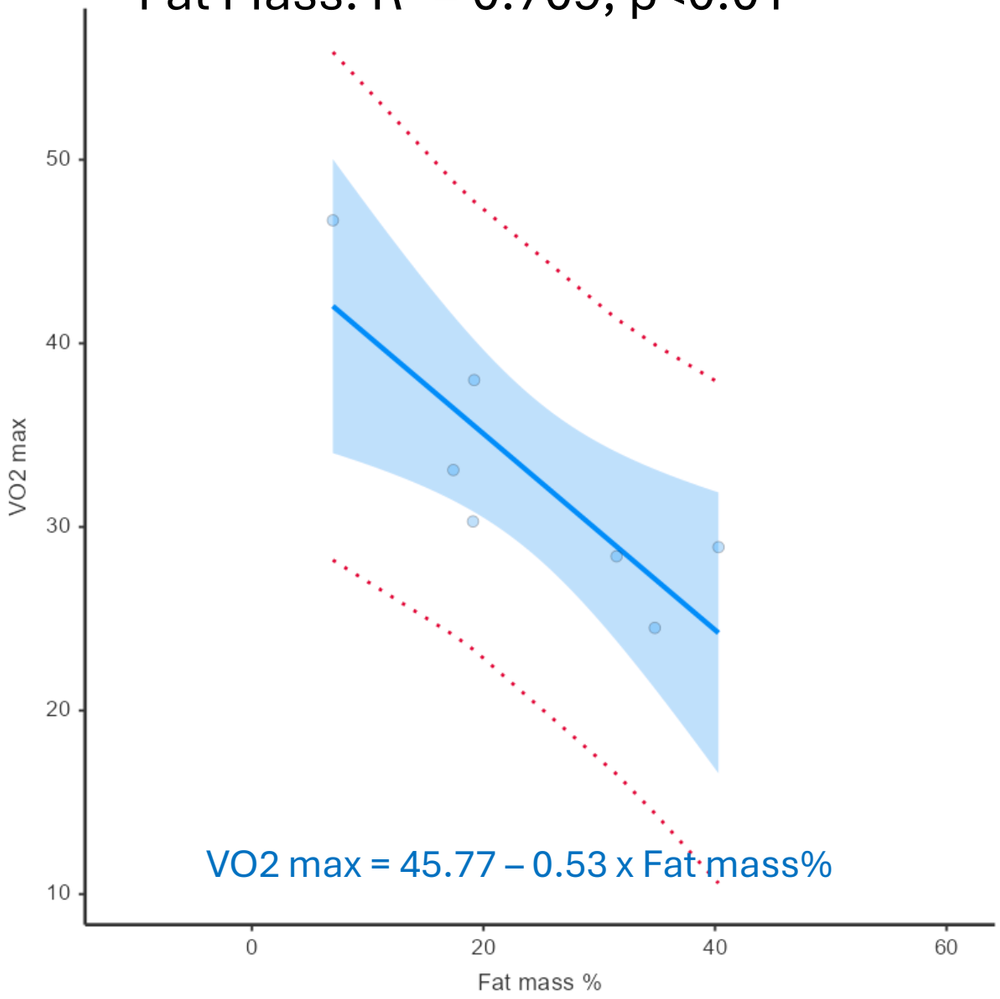




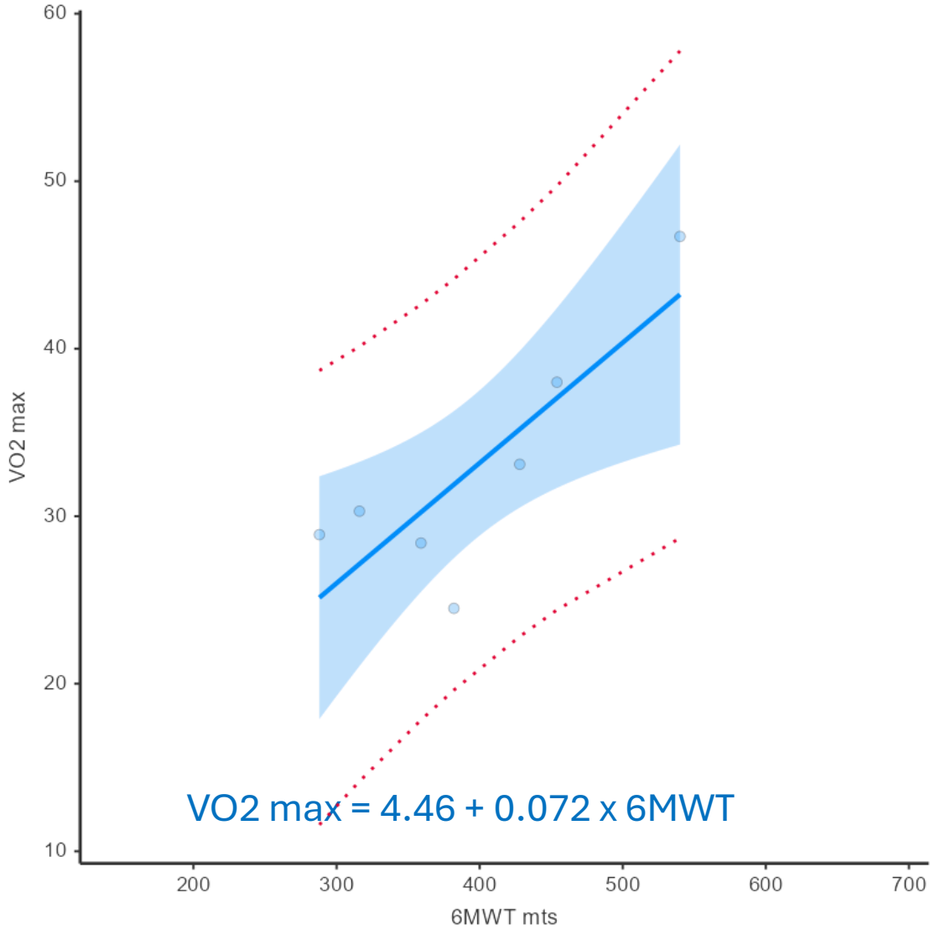
Results

VO2 max predictors

Fat Mass: $R^2 = 0.709$, $p<0.01$



6-Minute walking Test: $R^2 = 0.699$, $p<0.01$



Conclusions

- Increased physical activity associated with improved cardiorespiratory function
- Higher physical activity levels linked to lower fat mass and max heart rate
- 6MWT and handgrip strength as potential clinical proxies for exercise tolerance
- Insights for developing strategies to increase physical activity in adults with achondroplasia

Future Directions

- Larger sample size
- Longitudinal studies
- Interventional studies

References

1.Hoover-Fong J, et al., Lifetime impact of achondroplasia. Bone. 2021 May; 146:115872.

2. de Vries O, et al., Physical fitness and activity level in Norwegian adults with achondroplasia. AmJMedGenet, 2021 Apr;185(4):1023-1032.

3.Mezzani A et al., Standards for the use of cardiopulmonary exercise testing for the functional evaluation. EJCardiov Prev Rehabil. 2009 Jun;16(3):249-67

Credits: IDSF 2013

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Thank you

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