Physical Capacity in Achondroplasia – a study of Young, Mature, and Middle-Aged Adults

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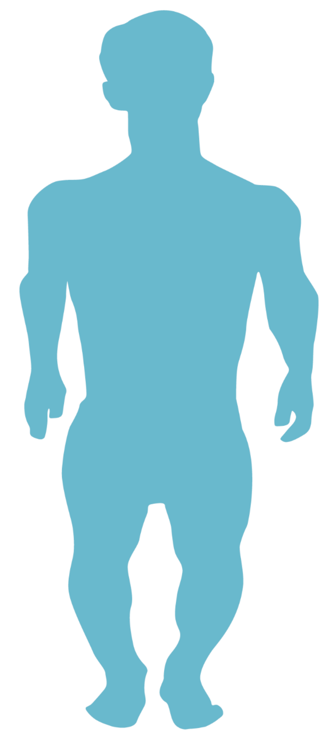

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INTRODUCTION

Skeletal dysplasias are rare bone conditions of genetic origin, being a heterogenous group of 771 forms, with short stature as a common feature.

Achondroplasia is one of the more frequent skeletal dysplasias with a prevalence of 1:25.000 [1]. It is characterized by disproportionate short stature with shortening of the lower as well as the upper limbs with an average trunk length [2]. Adult standing height is -6.0 standard deviation score, which translates in average height of 135 cm for men and 127 cm for women [3]. The skeletal features affect physical functioning and tend to originate multiple medical complications including neurologic compression as in the spinal canal, hyper lordosis, joints hyperlaxity, genu varus [4] and obstructive breathing [2]. These complications are aggravated by obesity, highly prevalent in achondroplasia. Physical limitations that highly impact health and quality of life and as this population ages, understanding factors influencing physical capacity is key.

AIM

Characterization of functional exercise capacity of young adults, mature and middle-aged adults with achondroplasia using the 6-min walking test (6MWT).

METHODS

16 adults with achondroplasia were divided into 3 age groups: young (18-25y), mature (26-44y) and middle-aged (45-59y). Functional exercise capacity was assessed using the 6-minute walk test (6MWT) and hand grip strength. Multiple regression analyzed the effect of age, height and weight on 6MWT and descriptive analyses for the hand grip strength and physical activity habits, evaluated by the IPAQ questionnaire, measured in metabolic equivalent (MET)-minutes per week (MET-min Total/week).

16 participants

G1 - Young adults
(18-25 years)
5 participants

G2 - Mature adults
(26-44 years)
6 participants

G3 - Middle-aged adults
(45-59 years)
5 participants

Results

Table 1 – Sample results (mean ± standard deviation)

Groups	weight (kg)	height (cm)	6MWT (m)	MET-min total/week	Hand grip (kg)
1 (n=5)	49.1 ± 12.4	126 ± 14.3	418 ± 78.0	1090 ± 966.0	8.36 ± 4.12
2 (n=6)	54.4 ± 17.7	129 ± 14.9	388 ± 115.0	980 ± 935.0	12.1 ± 5.81
3 (n=5)	57.6 ± 13.9	120 ± 8.9	384 ± 59.5	373 ± 328.0	13.3 ± 5.88

Figure1- Group 1 participant walking

Table 2 – Model coefficients for the 6MWT. "a" as reference level.

Model Fit Measures

Model	R	R^2	F (Overall Model Test)	df1	df2	p
1	0.807	0.652	5.15	4	11	0.014

Predictor	Estimate	SE	t	p	Stand. Estimate
Intercept [a]	-164.99	159.81	-1.032	0.324	
Weight	-3.54	1.22	-2.911	0.014	-0.602
height	5.99	1.39	4.300	0.001	0.903
Age:					
2 – 1	-26.96	35.96	-0.750	0.469	-0.317
3 – 1	32.80	40.45	0.811	0.435	0.385

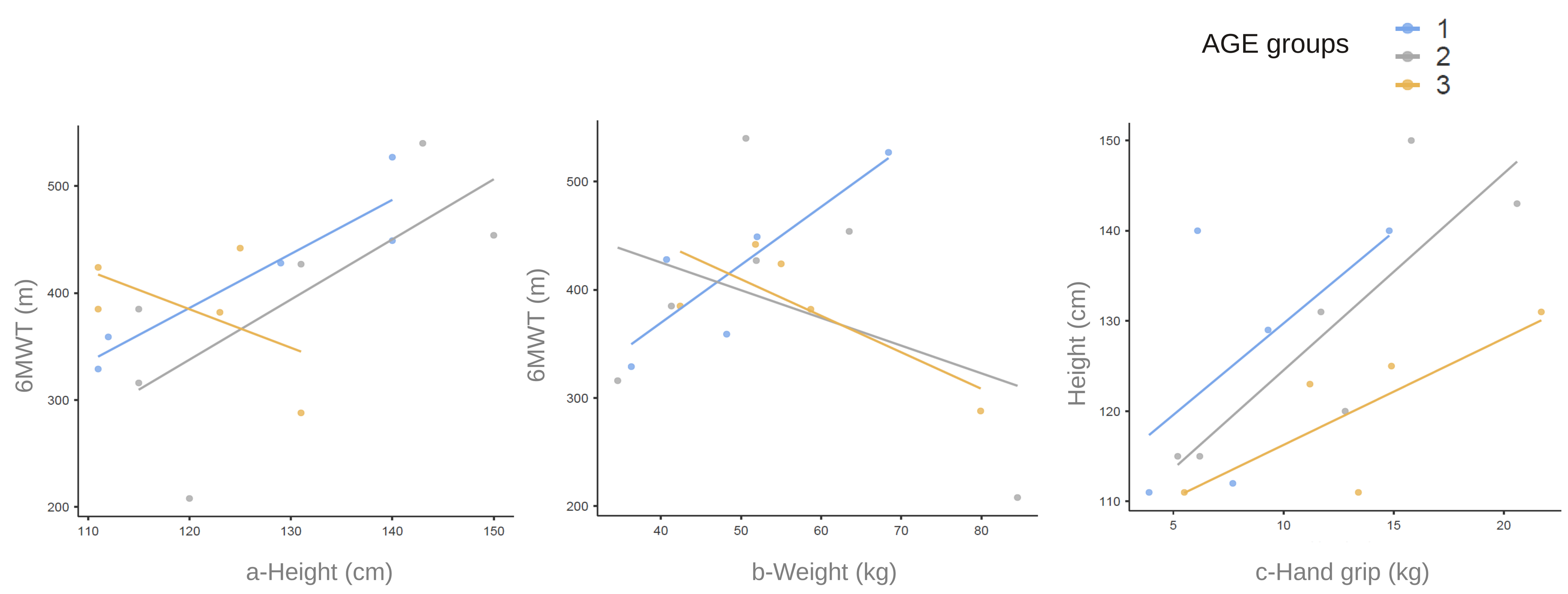

Figure 2 – Relationship between the 6MWT and height (a) and weight (b), and between height and handgrip strength (c), for the 3 age groups.

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Weight and height explained 65% of 6MWT variance. Weight was a negative predictor (-0.62, p<0.014) while height was a positive predictor (0.903, p<0.001). Age group difference was not significant.

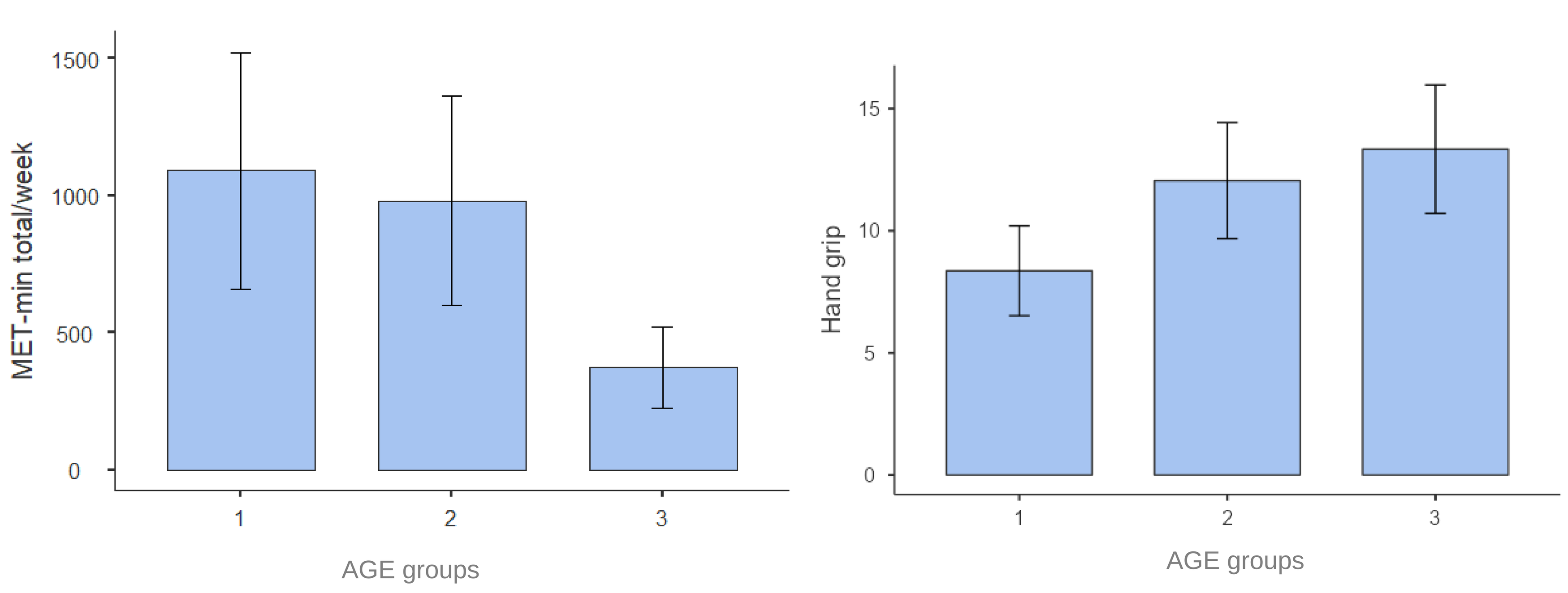

Figure 3 – Frequency of MET-min total/week and hand grip among the 3 age groups

DISCUSSION

A taller height was associated with a greater walking capacity for G1 and G2. The G3 presented lower walking fitness most likely due being the group with higher weight. Also, joint pain and lumbar spinal stenosis are more prevalent among middle-aged adults. This is in line with Fig2b, as young adults with higher weight were still able to walk a longer distance, favoring their younger age. An increased height was also related to a superior upper strength across ages, which translated physical fitness challenged due the shorter stature in individuals with achondroplasia. Overall, a younger age and reduced weight are good physical capacity indicators while increased weight and a shorted stature represented added challenges for walking distance capacity, most likely, higher height is associated with a longer stride, resulting in a longer distance travelled.

CONCLUSION

Weight and height are major factors influencing physical capacity in achondroplasia across adulthood. Sustaining ideal weight and activity levels through aging may optimize function and quality of life. Strategies to stimulate physical activity within with population, may positively reflect in an improved physical capacity and weight reduction.

Figure 4– Older Adult (>65 years) biking and mature adult walking.
Credits: Isla bikes (left) and Shutterstock

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Promoting a heathier aging and sustainability for adults with achondroplasia

Acknowledgments

RoboCorp Laboratório

ANDO