Relationships between body composition and exercise types in adults with Achondroplasia

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# Achondroplasia



#### **Genetic Condition**

Rare bone condition caused by a single point mutation at the fibroblast growth factor receptor 3 gene (FGFR3).



#### Skeletal Impact

Disproportionate short stature.

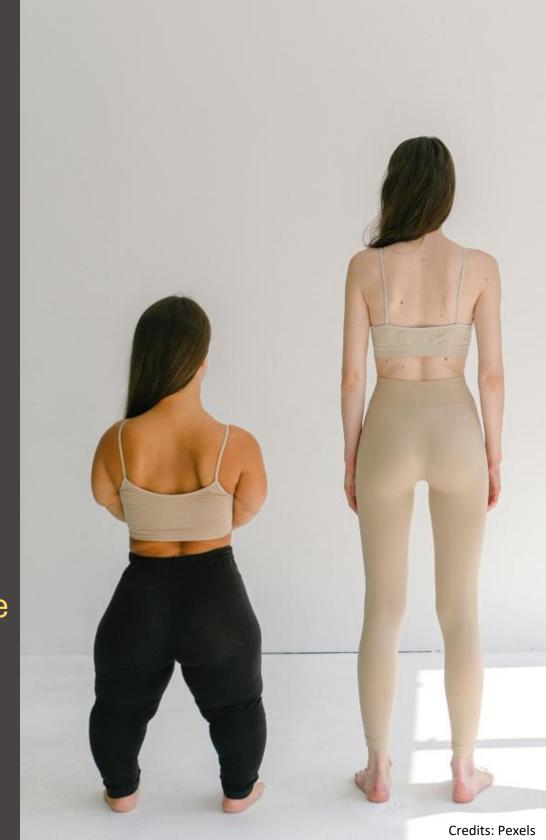
Adult height: 110-130 cm.

Skeletal deformities, spinal stenosis, pain.



#### Prevalence

1 in 25 000 births



# Physical impact



#### Genu varus

Knees bend inward, resulting in a bowlegged appearance.

#### Joints hyperlaxity

Increased joint flexibility and laxity, leading to instability and increased risk of injury.

#### Hyper lordosis

Exaggerated curvature at the lower back, which causes pain and mobility challenges.

#### Small chest

Can lead to respiratory difficulties and increased susceptibility to respiratory infections.

#### Facial hypoplasia

Midface and jaw are underdeveloped, leading to a distinctive facial appearance.

#### Macrocephaly

Larger head size which can contribute to the characteristic appearance of the condition.

# Functional diversity / disability



#### **ACHONDROPLASIA** FGFR3 mutation: Abnormalities of chondrogenesis and endochondral bone formation **BODY FUNCTIONS AND STRUCTURES ACTIVITY AND PARTICIPATION** Mental functions Learning and applying knowledge - Cognitive performance · Structures of the nervous system, resulting in Communication - Ventriculomegaly - Cervicomedullary compression - Development of communication milestones - Social cognition Mobility · The eye, ear, and related structures, resulting in - Development of gross motor activities - Development of fine motor activities - Altered hearing - Otitis media · Structures of the cardiovascular, immunological, and respiratory - Development of independent self-care skills systems, resulting in - Development of eating and drinking skills - Altered functions of the respiratory systems Major life areas - Restrictive and obstructive lung disease - Education - Reduced exercise tolerance - Employment · Structures related to movement, resulting in Community, social, and civic life - Altered functions of the joints and bones - Recreation and leisure - Bony deformity - Soft tissue contracture - Spinal deformity - Macrocephaly - Short limbed short stature - Midface hypoplasia - Kyphosis - Joint hypermobility and hypomobility - Reduced muscle strength - Secondary obesity **ENVIRONMENTAL FACTORS** PERSONAL FACTORS · Products and technology - Lack of equipment, furniture, toys matching anthropometric limitations Natural environment and human-made changes to environment - Interaction between body size and environment · Support and relationships - Family, teachers, community groups · Attitudes - Societal attitudes · Services, systems, and policy - Attitudes and beliefs of family and community

# Aims







Do different types of exercise influence physical well-being?

# MET values for Physical Activity



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

MET-minutes are calculated by multiplying the MET score by the minutes performed. This provides a measure of the volume of activity, which is equivalent to kilocalories for a 60 kg person.

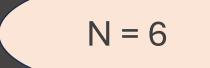
# Grouping types of exercises

#### Applying a semi-structured interview



#### Group 0 (G0)

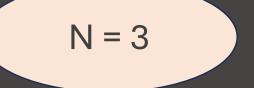
No exercise (Includes walking)





#### Group1 (G1)

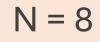
Lower MET exercises (leisure swimming or slow cycling)



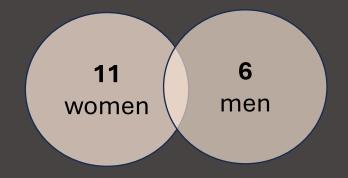


**Group 2 (G2)** 

Higher MET exercises (gym workout, martial arts)

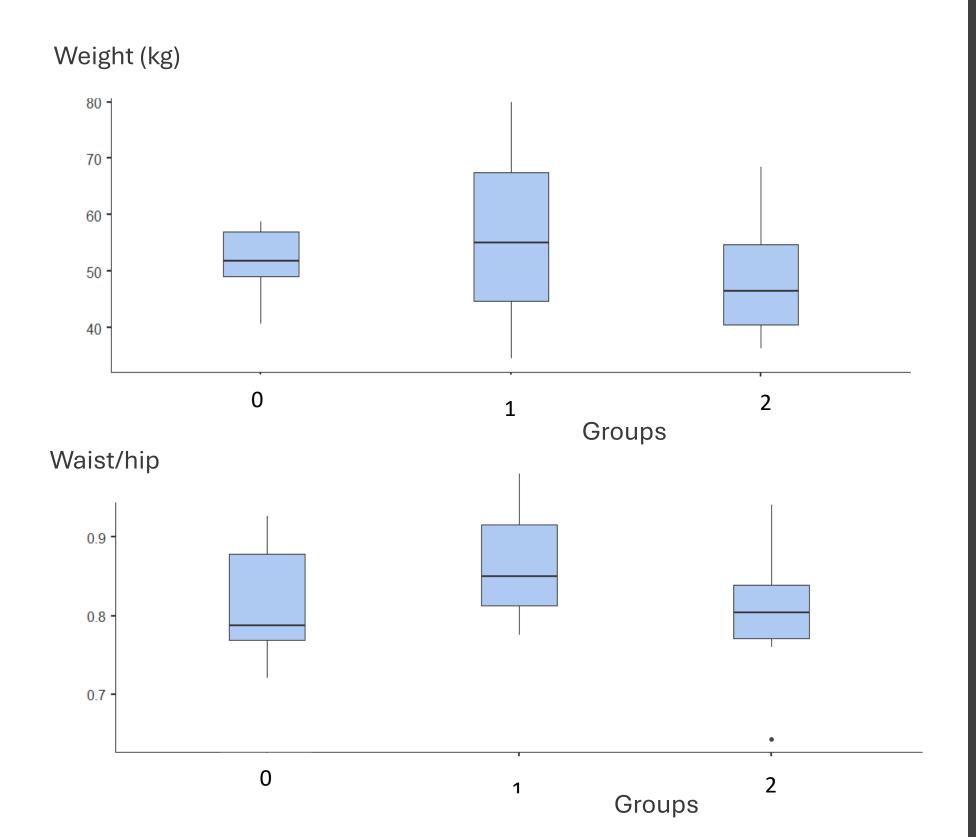


# Anthropometric measures



Exercise groups (G)	Age (years)	Weight (kg)	Height (cm)
G0 (n=6)	35.7±16.7	56±15.2	125±9.4
G1 (n=3)	48±6.2	56.5±22.7	119±10.6
G2 (n=8)	35.8±12.2	49±11.9	128±15.5
	Waist (cm)	Hip (cm)	Waist/Hip
G0 (n=6)	86.6±15.1	106±11.6	0.82±0.08
G1 (n=3)	91.7±20.8	105±13.6	0.87±0.11
G2 (n=8)	77.8±10.4	96.9±9.5	0.80±0.09

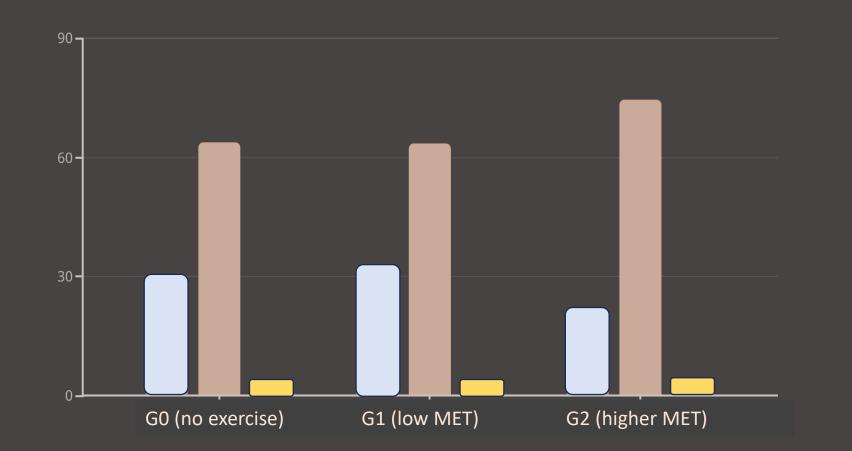




# Anthropometric measures analysis

# Body composition

Exercise groups (G)	Fat mass %	Lean mass %	Bone mineral mass%
G0 (n=6)	30.5±10.3	63.8±11.3	3.58±0.52
G1 (n=3)	33±12.0	63.5±11.3	3.47±0.72
G2 (n=8)	21.9±8.1	74.5±8.74	4.08±0.45

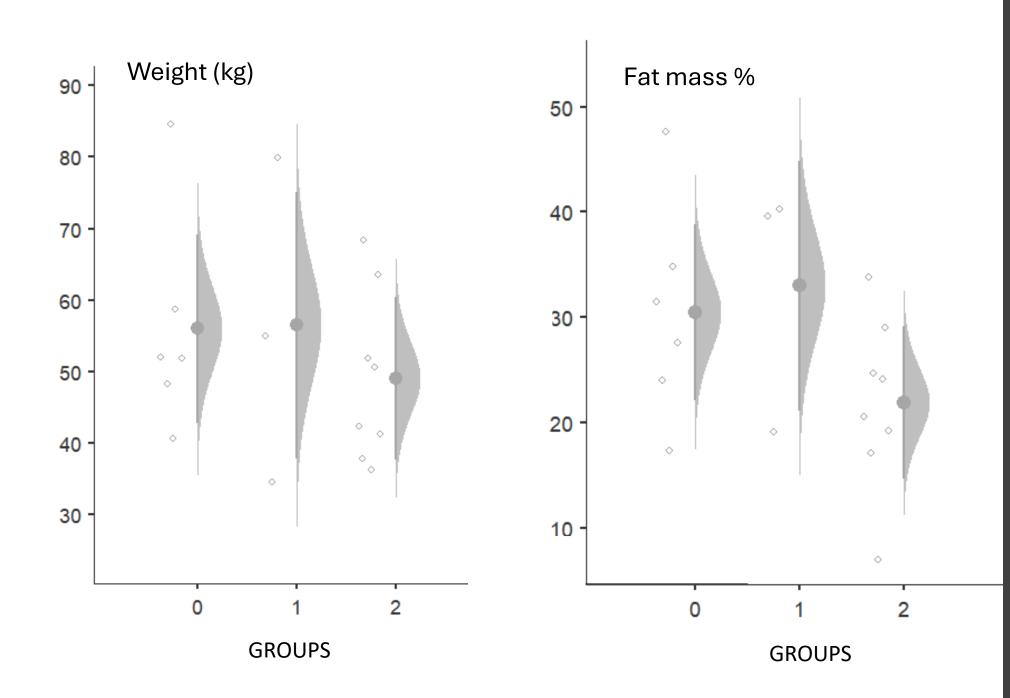


Bioimpedance Tanita MC-780

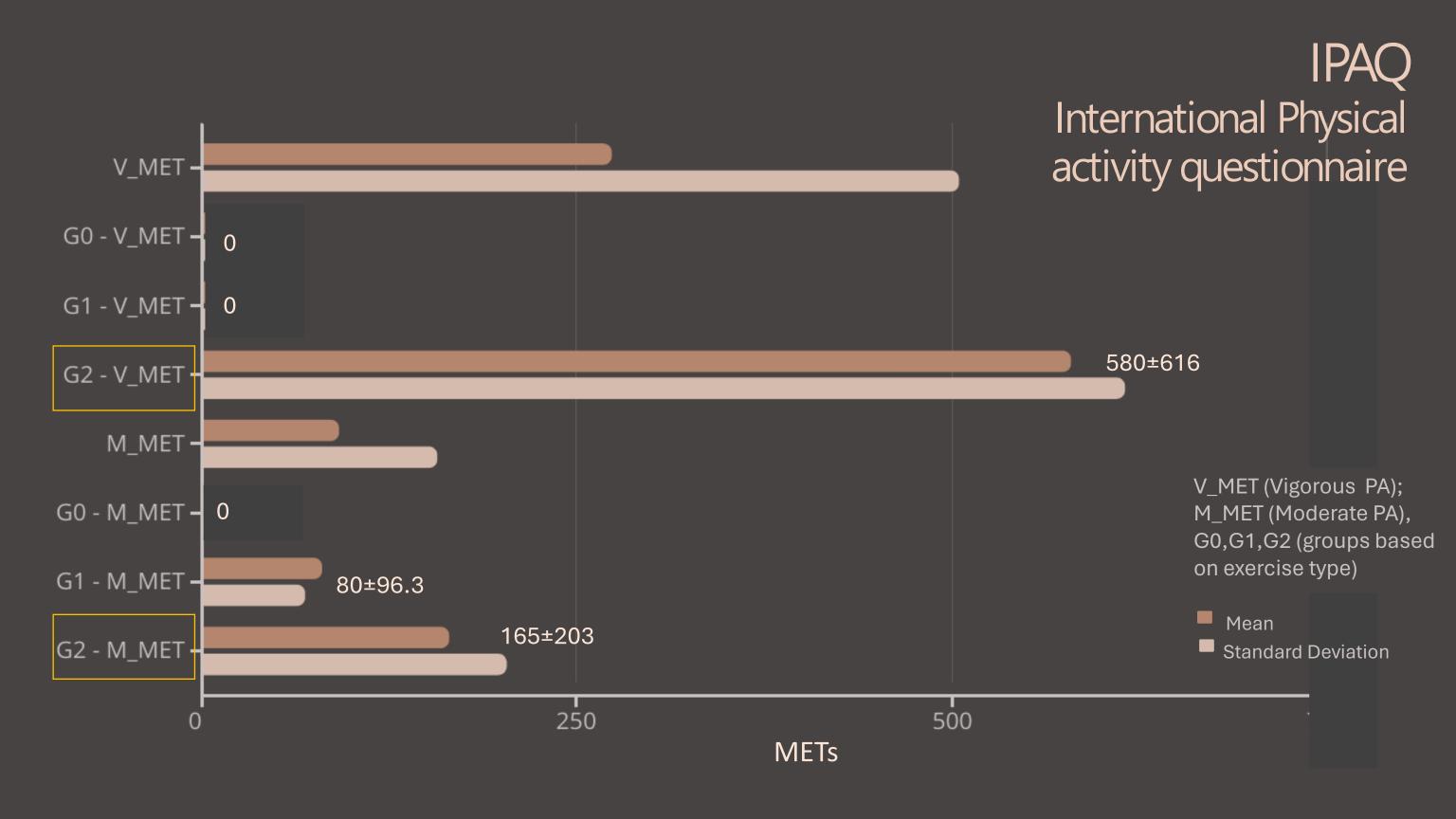
Fat mass %

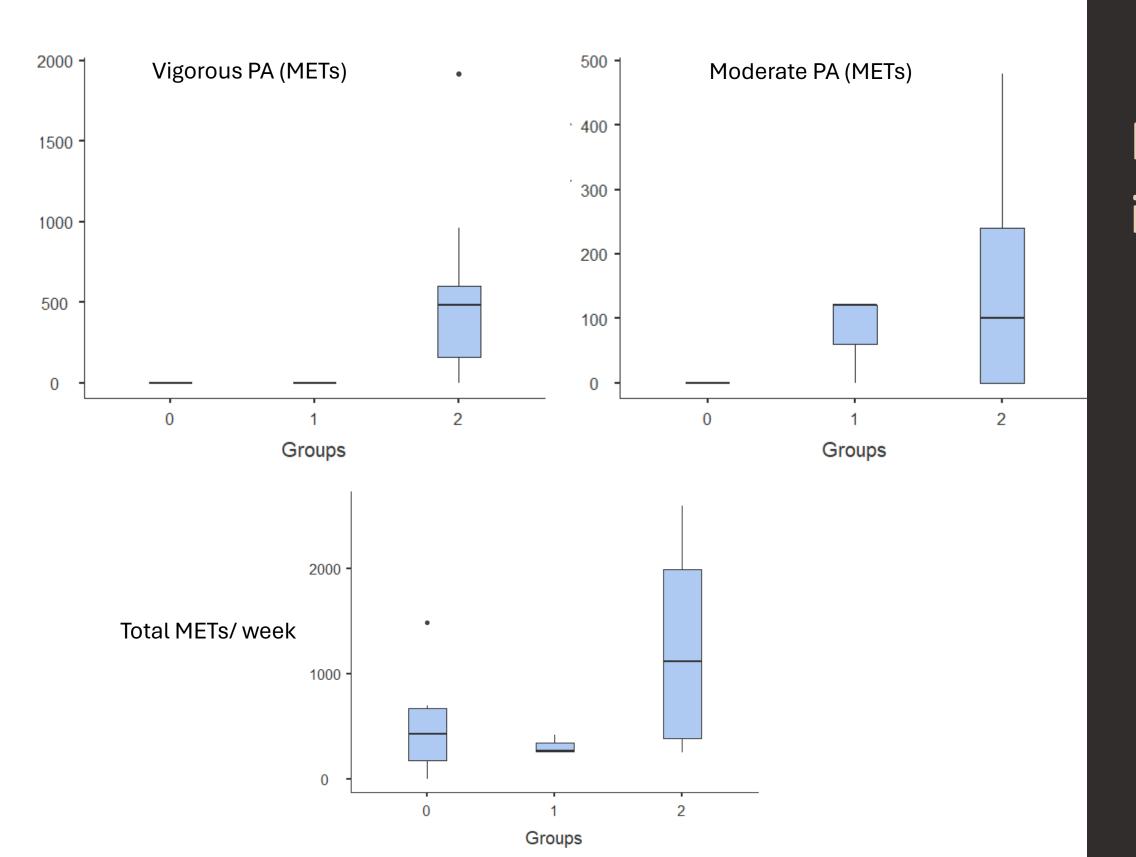
Lean mass %

Bone mass %



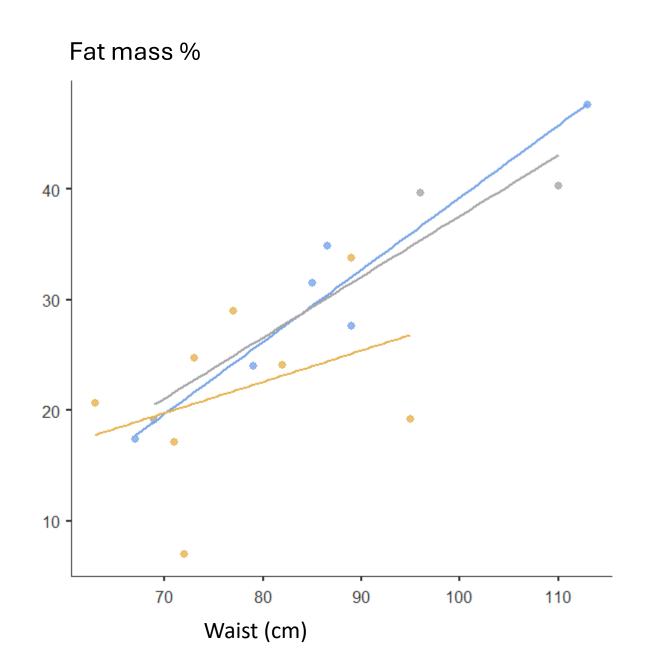
# Mean comparison between groups (95% CI, p<0.05)

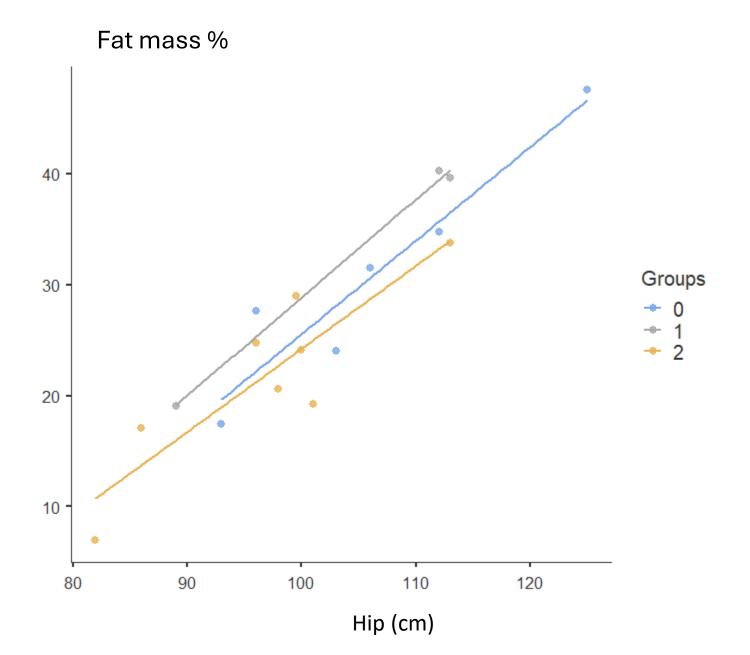




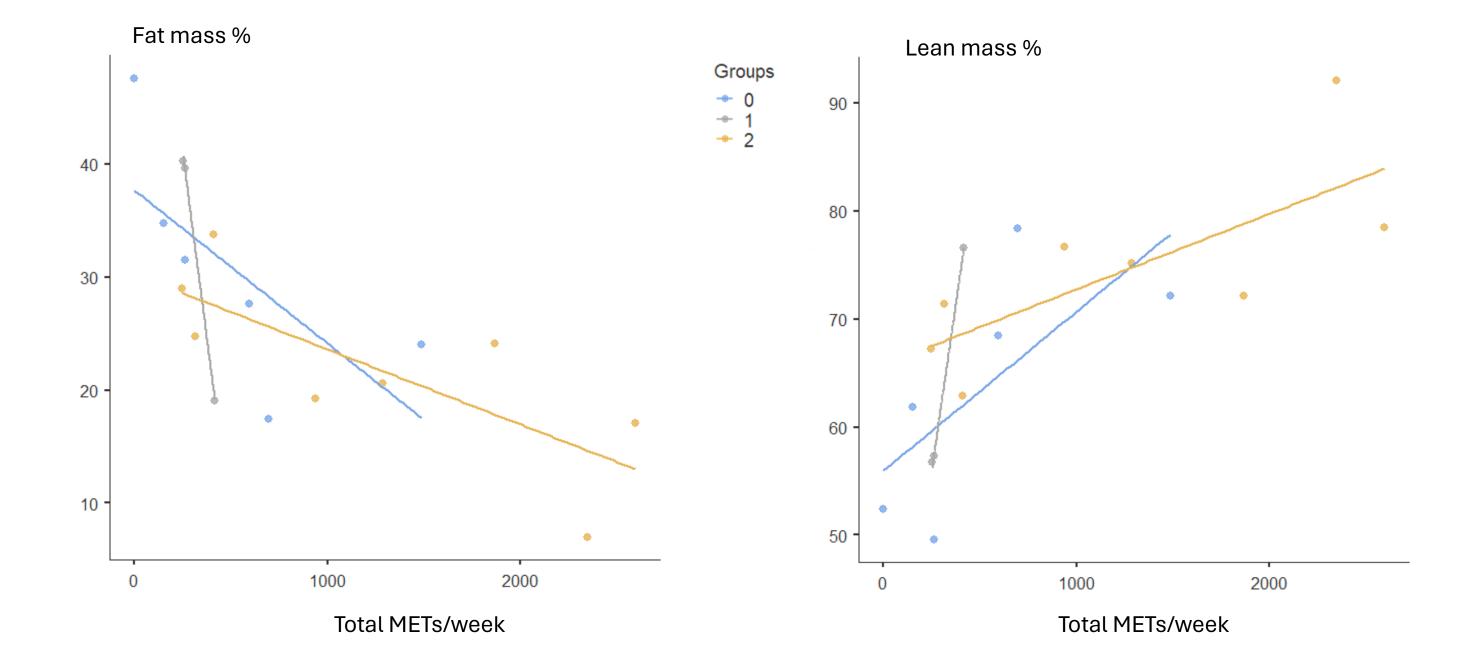
# Physical activity in METs

# Correlations anthropometric x Body composition

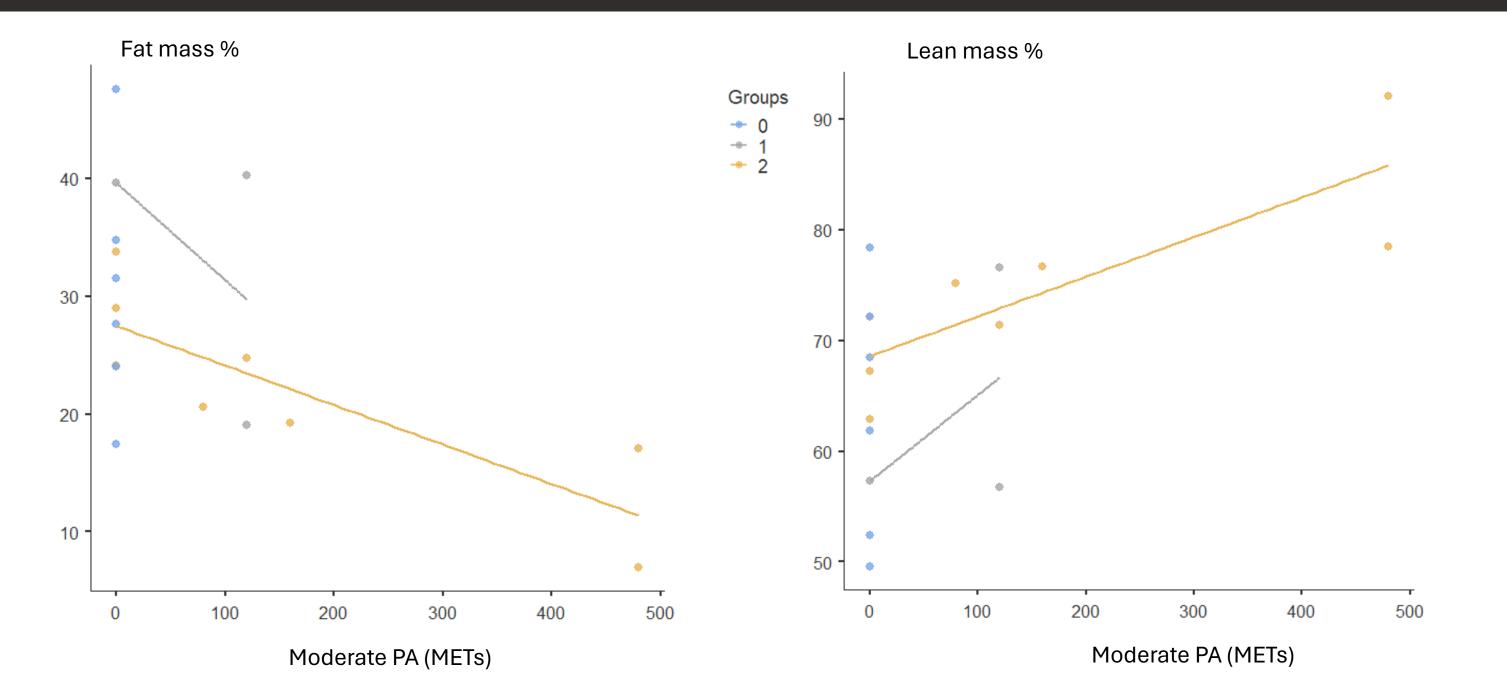




# Correlations Body composition x Physical activity



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# Results summary

21.90 Fat mass %

Lower in G2 vs G0 G2 vs G1

74.50 Lean mass%

Higher in G2 vs G0 G2 vs G1

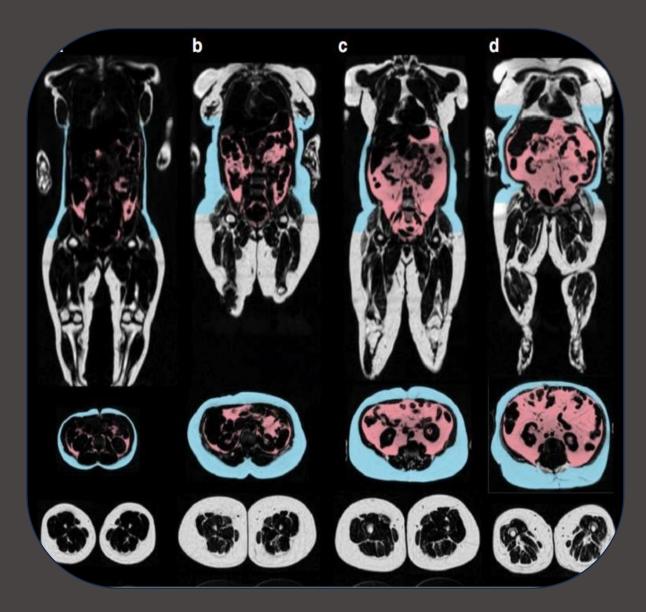
4.08

Bone mass %

Higher in G2 vs G0 and G2 vs G1



### Conclusions



Higher adiposity associated with lower MET exercises and physical activity



Improving body composition, may facilitate physical activity participation









# Thank you

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