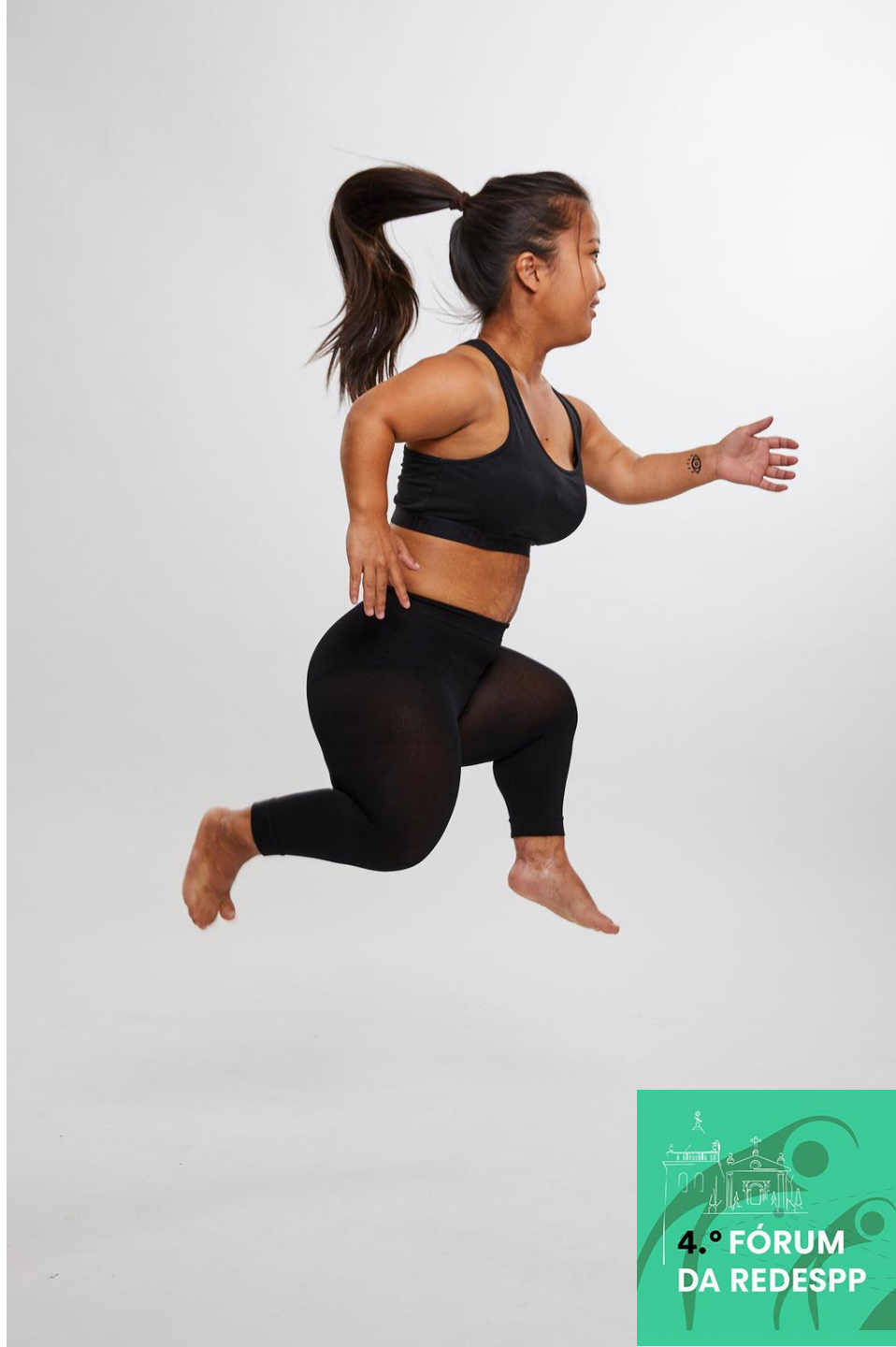
UNIVERSIDADE DE ÉVORA

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

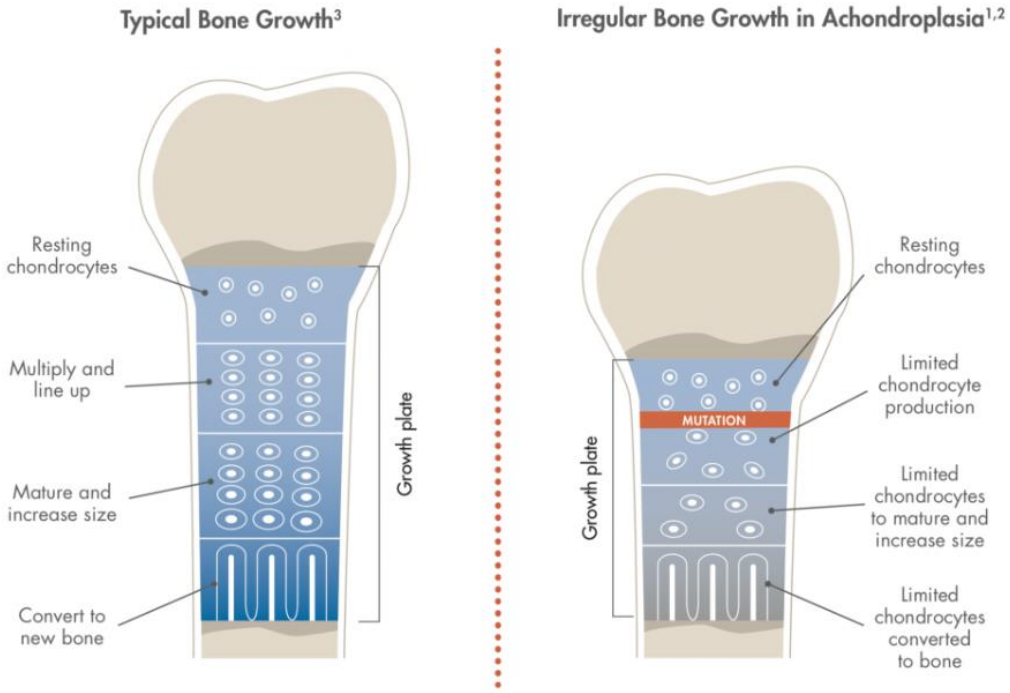


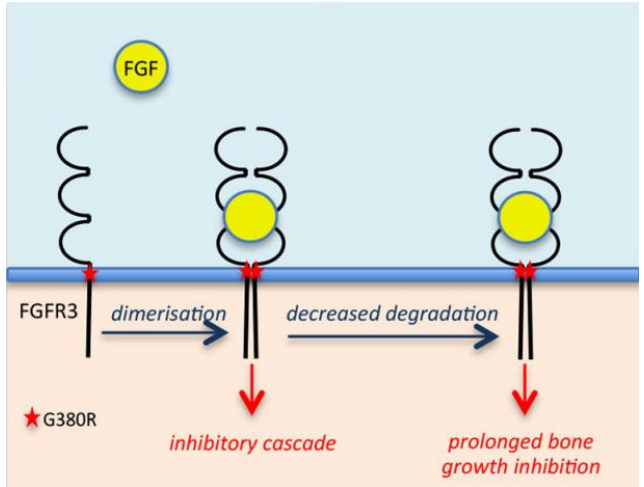
Inês Alves
Maria António Castro
Sofia Tavares
Orlando Fernandes

4.º FÓRUM DA REDESPP

Achondroplasia

- 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

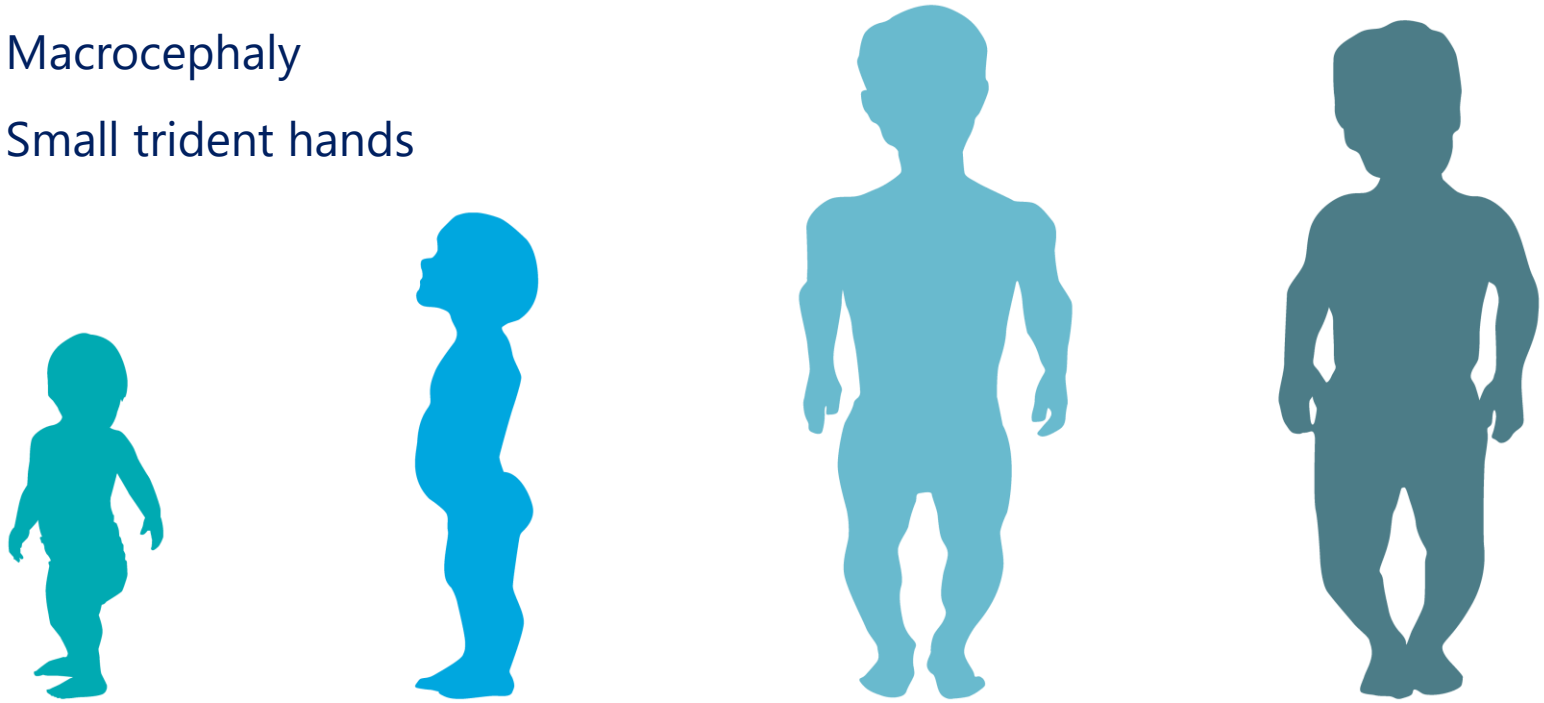


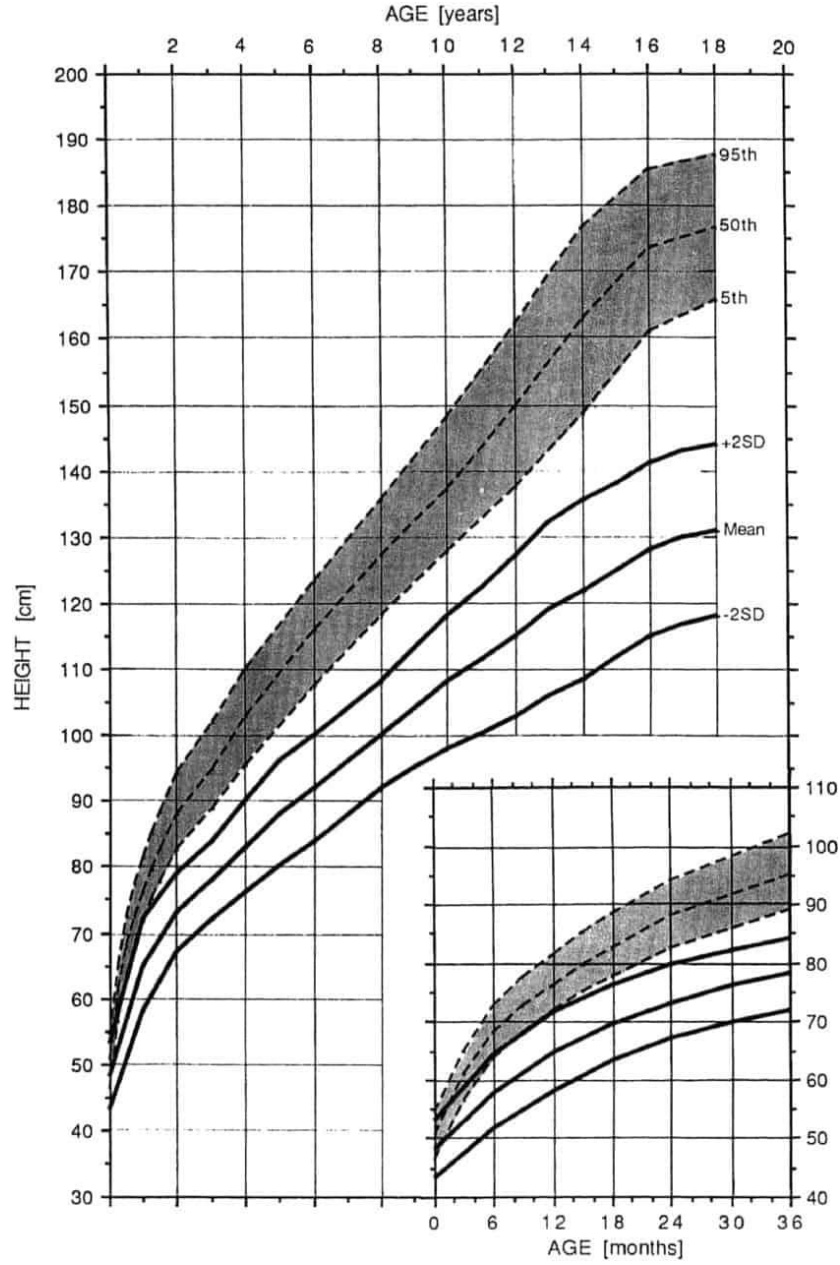


Prevalence **1:25 000 births**

Physical impact

- Genu varus
- Joints hyperlaxity
- Hyper lordosis
- Small chest
- Facial hypoplasia
- Macrocephaly
- Small trident hands





Objective

QoL in adults with achondroplasia

Comparing to the general population:

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

Functionality
Strength
QoL

Correlations and predictors

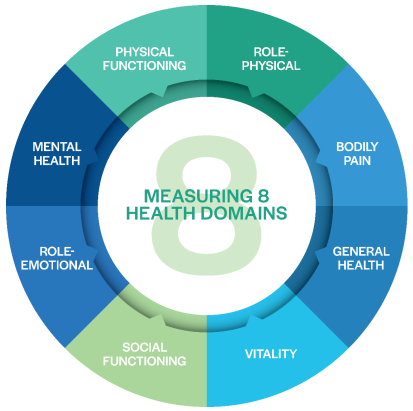
Physical activity

Methods

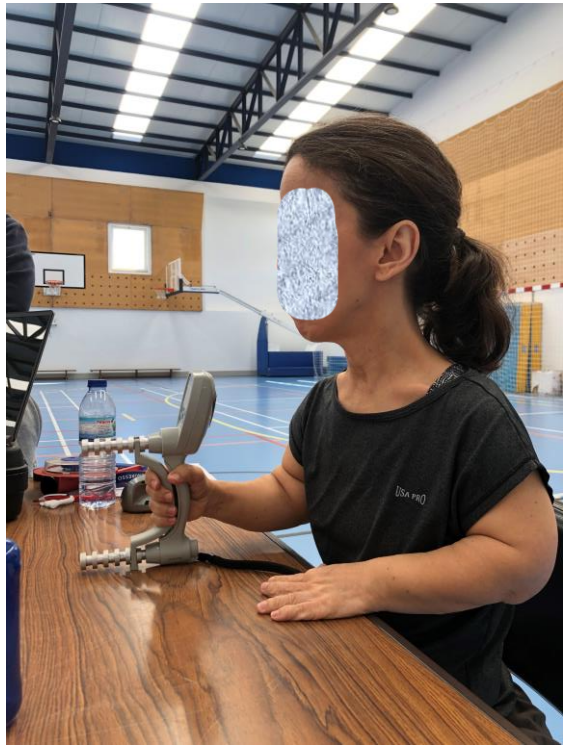
16 adults with achondroplasia (10 women and 6 men)

Gender	Age (years)	Height (cm)	Weight ()kg)	Fat mass (%)	Waist circunference	Hip circunference
Women (n=10)	34.9 ± 14.6	123.0 ± 11.6	49.3 ± 10.0	27.8 ± 7.2	80.3 ± 9.2	102.0 ± 8.5
Men (=6)	41.0 ± 11.8	130.0 ± 14.5	61.1 ± 18.5	26.5 ± 15.2	91.7 ± 18.0	100.0 ± 16.2

IPAQ
International Physical Activity Questionnaire



SF-36

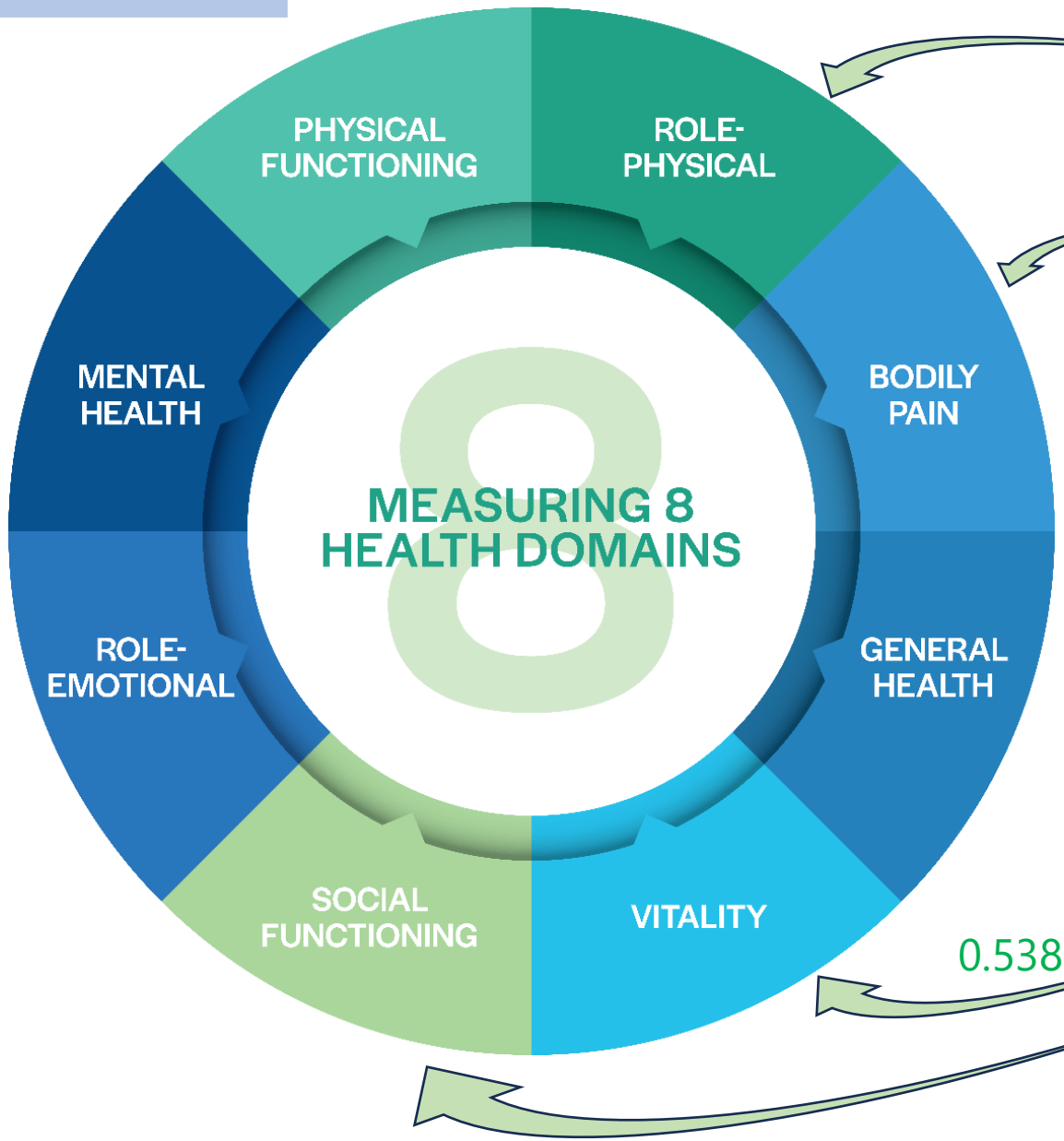


Results

Gender	Handgrip (kg)	6MWT (m)
Women (n=10)	9.1 ± 3.5	408.0 ± 57.2
Men (=6)	15.0 ± 6.4	377.0 ± 123.0

Strong correlations

QoL



0.516

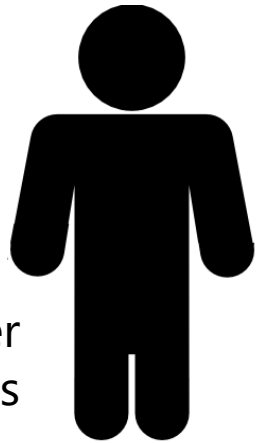
0.528

Correlations p<0.05

Moderate physical activity
(MET-min/week)

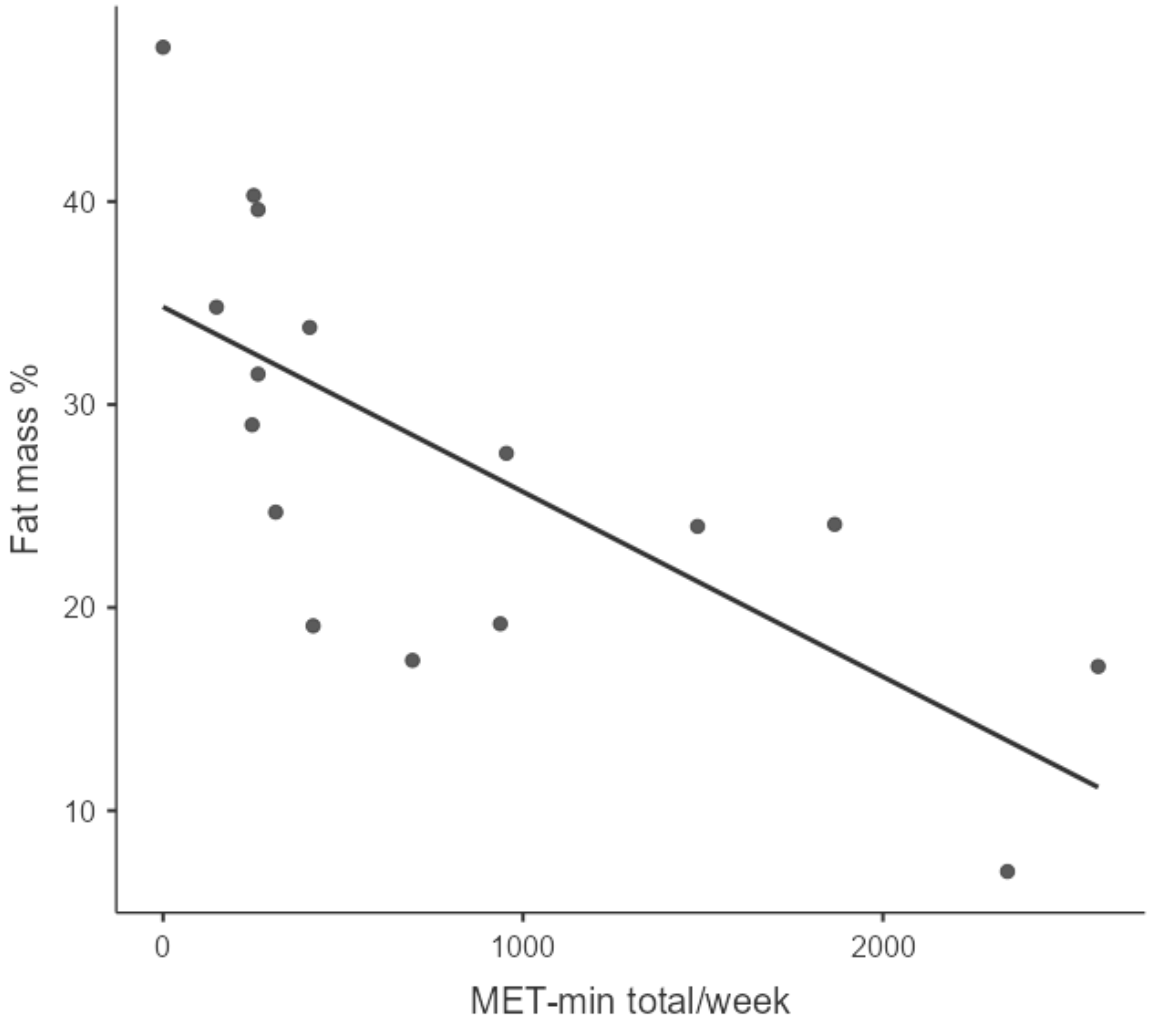
0.523

0.538



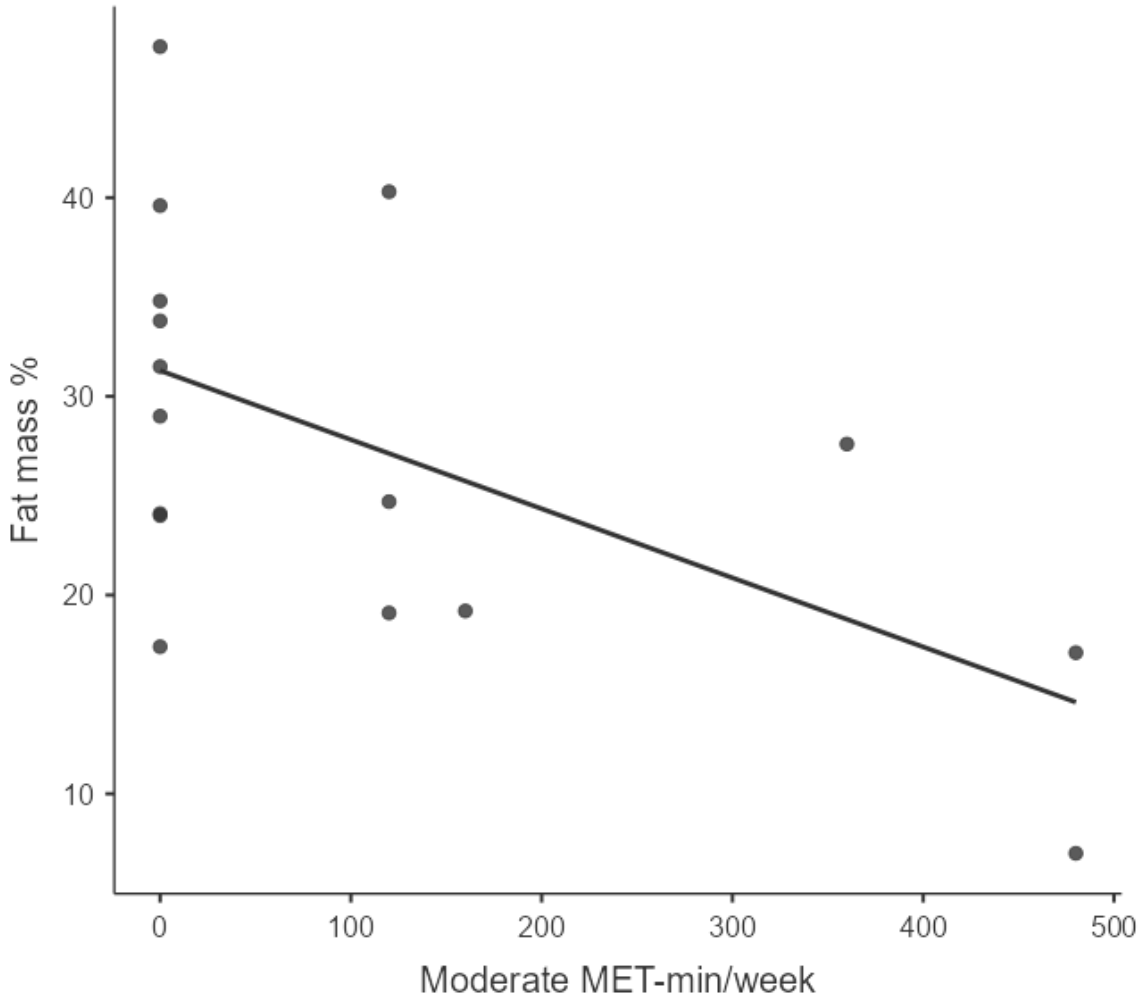
MEN scoring higher in all domains

Predictors



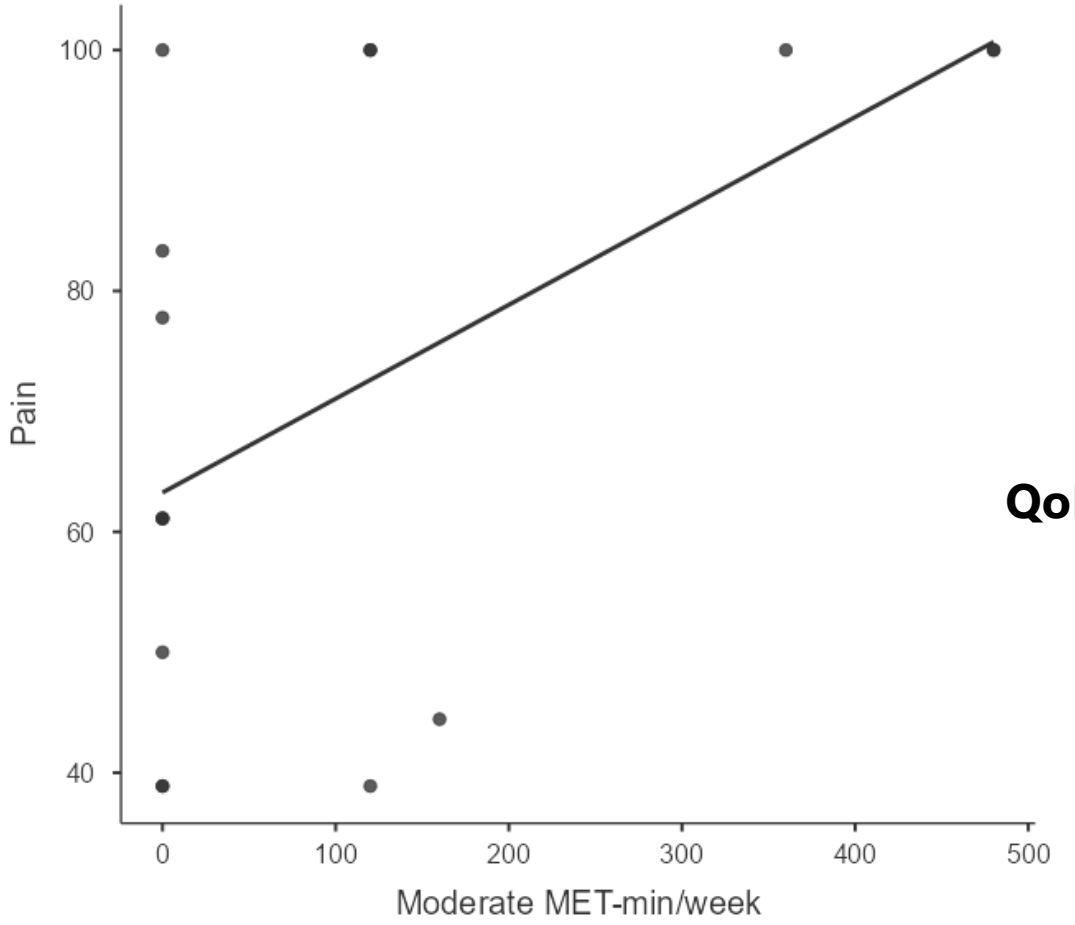
R: 0.71 (p 0.004)

Fat mass – Physical activity



R: 0.750 (p 0.001)

Predictors



QoL pain domain - Physical activity

R: 0.528 (p=0.036)

Conclusion

Physical activity

FAT MASS

QoL

UNIVERSIDADE DE ÉVORA

CHRC
COMPREHENSIVE HEALTH RESEARCH CENTRE

RoboCorp
Laboratório

Thank you

orlandoj@uevora.pt