UNIVERSIDADE DE ÉVORA

Physical parameters associated with the ability to exercise in women with achondroplasia

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A case control pilot study

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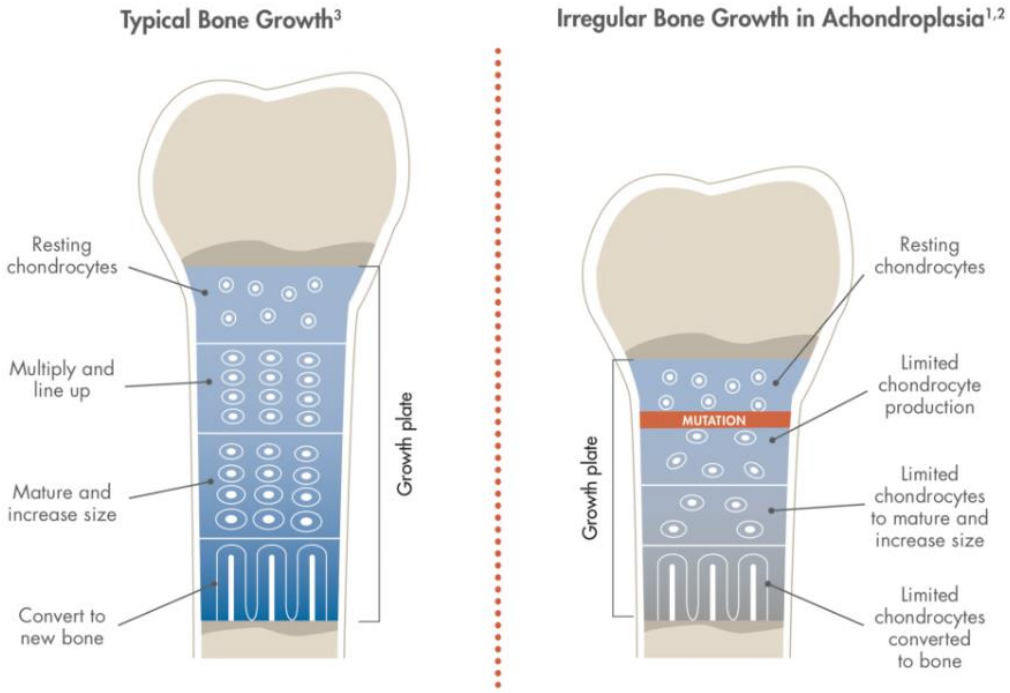
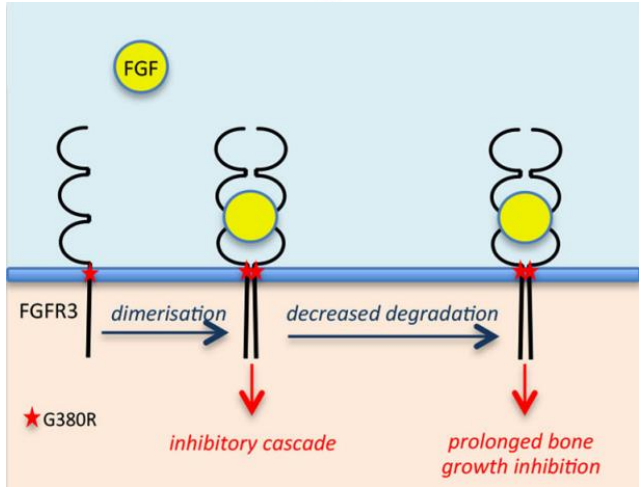
Achondroplasia

- 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

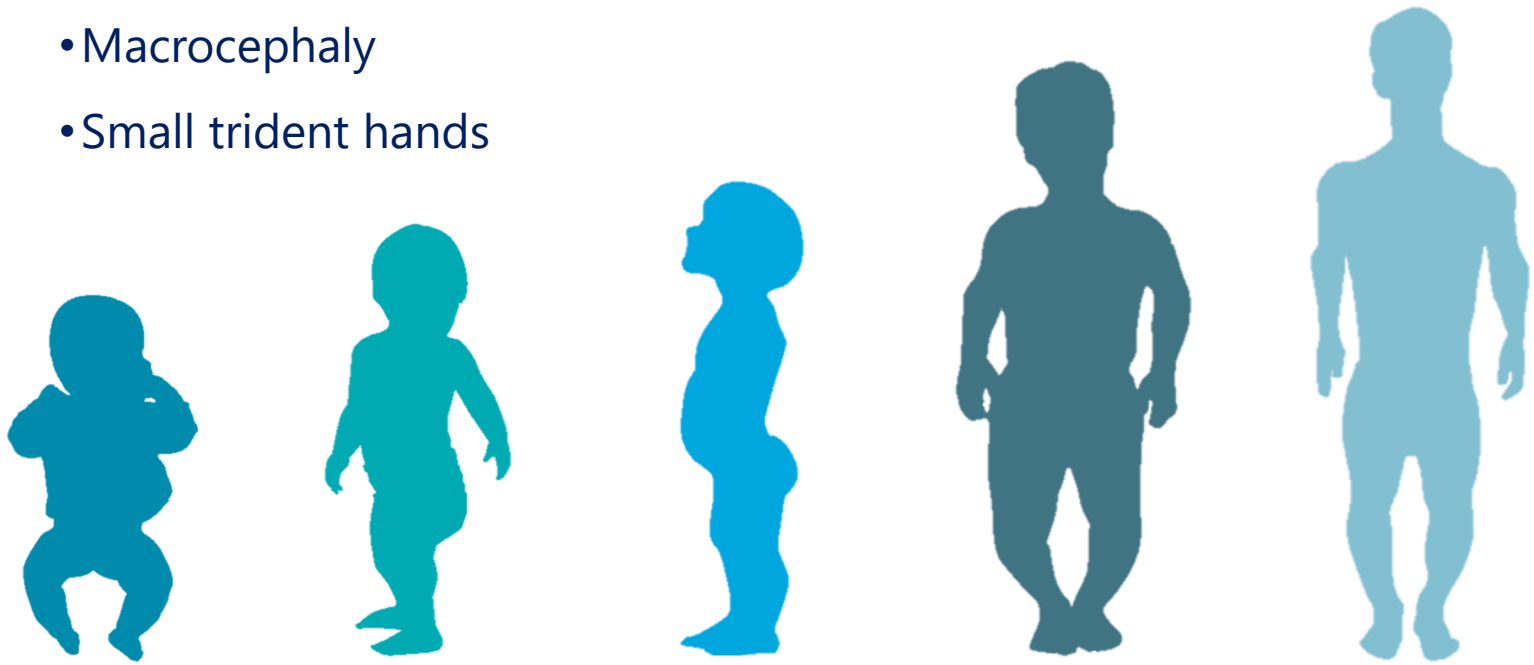
Prevalence **1:25 000 births**

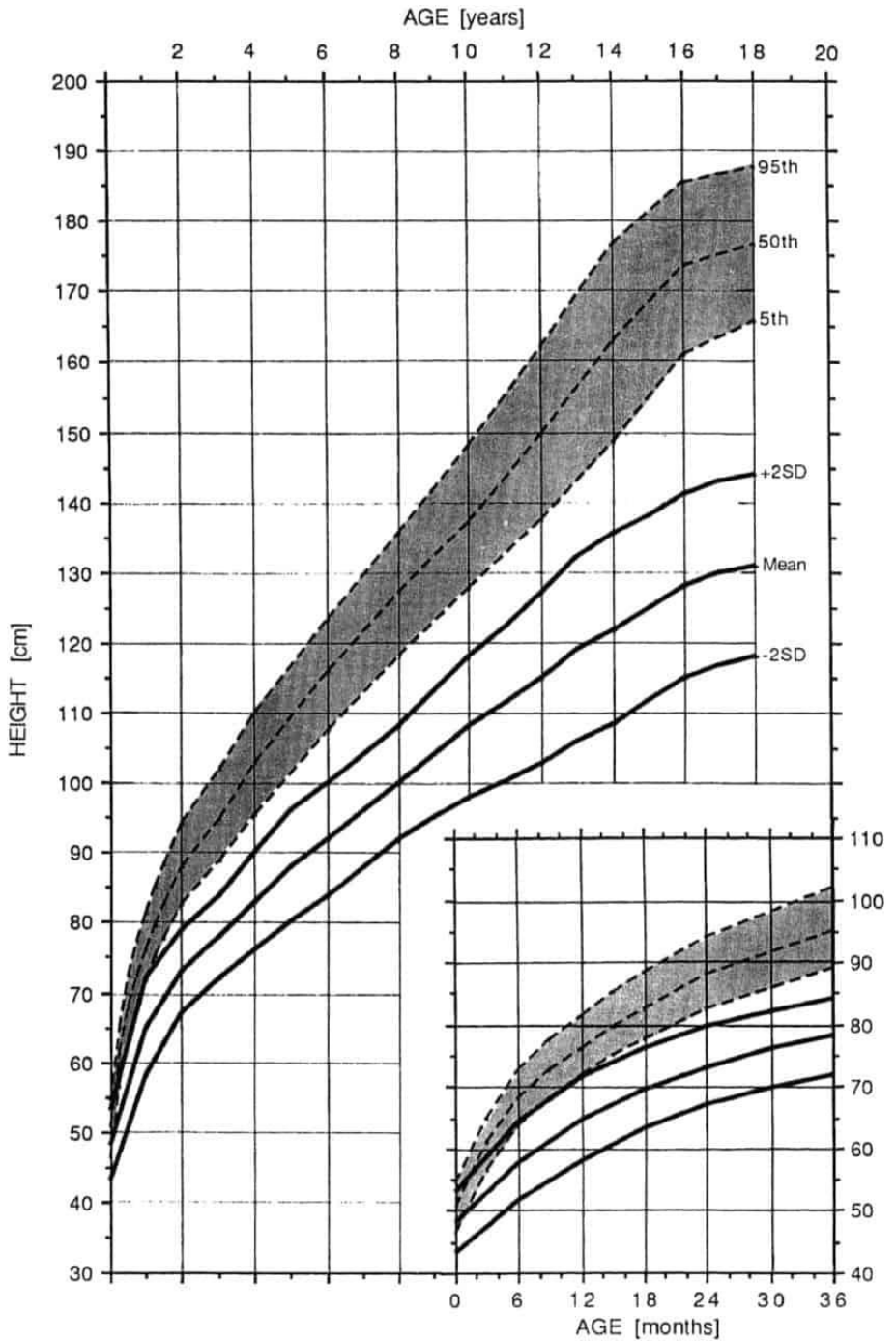


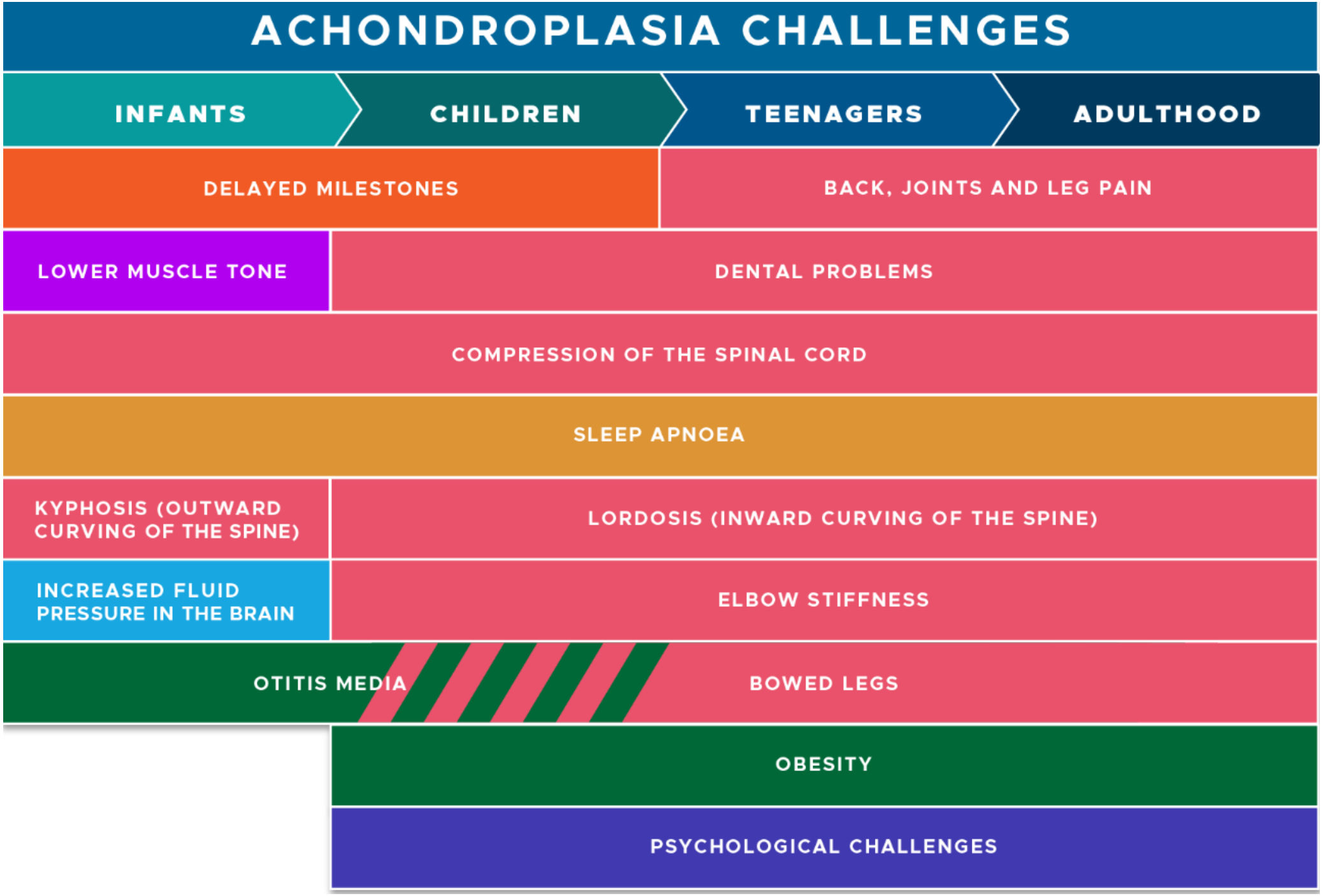


Physical impact

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






ACHONDROPLASIA CHALLENGES
INFANTS
CHILDREN
TEENAGERS
ADULTHOOD
DELAYED MILESTONES
BACK, JOINTS AND LEG PAIN
LOWER MUSCLE TONE
DENTAL PROBLEMS
COMPRESSION OF THE SPINAL CORD
SLEEP APNOEA
KYPHOSIS (OUTWARD CURVING OF THE SPINE)
LORDOSIS (INWARD CURVING OF THE SPINE)
INCREASED FLUID PRESSURE IN THE BRAIN
ELBOW STIFFNESS
OTITIS MEDIA
BOWED LEGS
OBESITY
PSYCHOLOGICAL CHALLENGES

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

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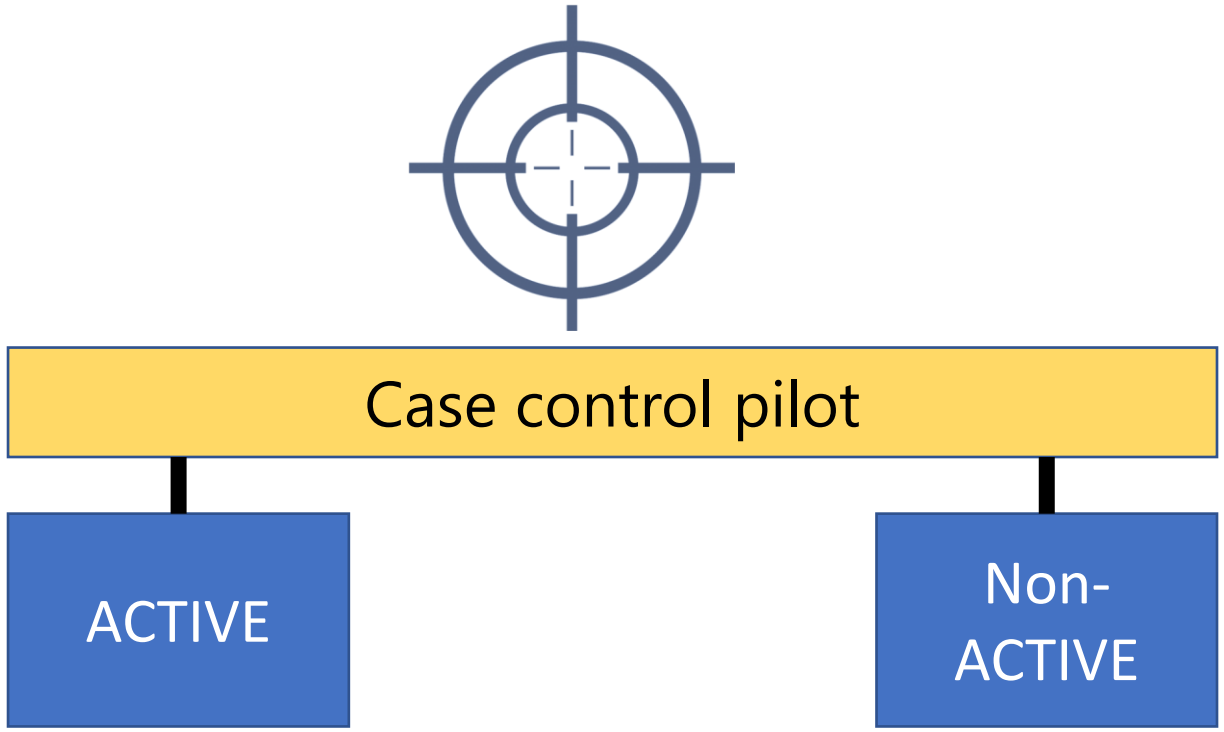
Benefits of PA and exercise for adults with achondroplasia

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

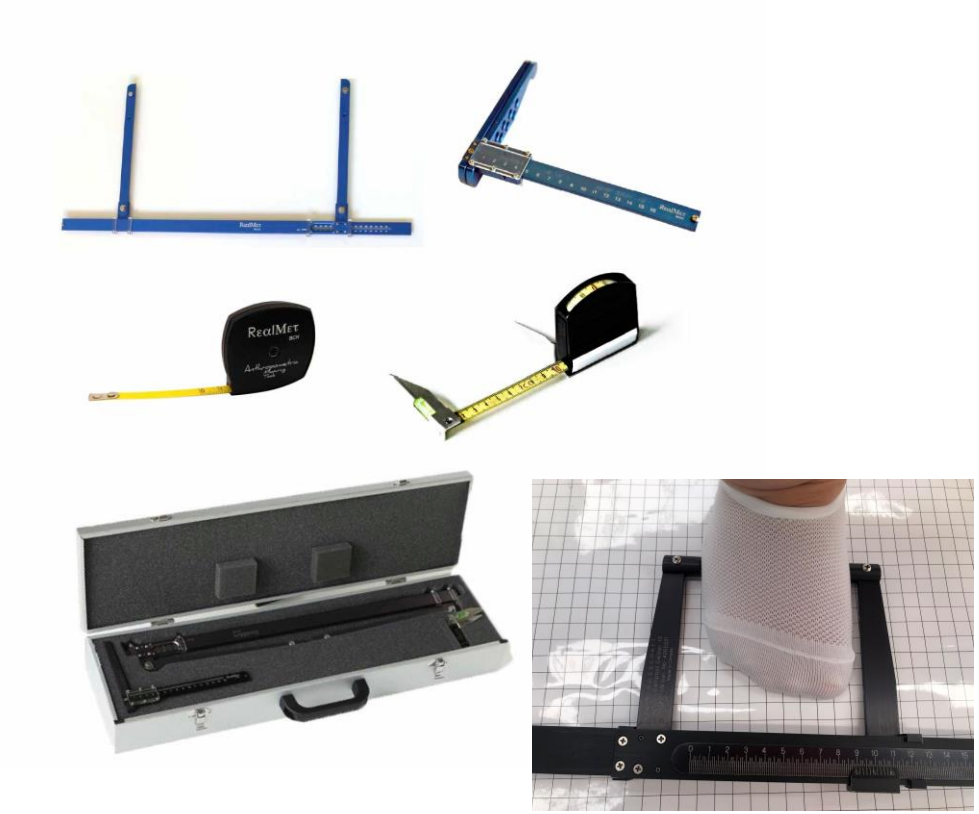
-Leg

-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)

20 cm height

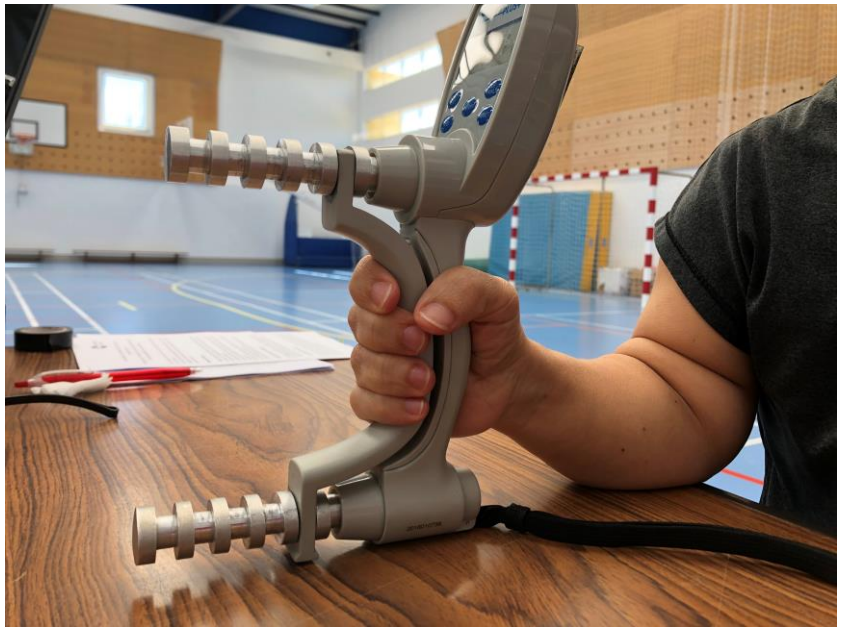
❖ Upper limbs

Hand strength

30 seconds biceps curl (2kg)

30 seconds push-up

1kg chest throw



Results

Active AG (2)		Non-active NAG (2)
48.2	**Weight (kg)**	55.4
32.8	**BMI (Kg/m2)**	44.4
77.8	**waist circumference (cm)**	92.5
26.9	**fat mass (%)**	36.7
69.3	**lean mass (%)**	60
22.5	**biceps curl**	16.5
15	**push-ups**	13
250	**chest throw**	236
16.5	**30CST**	11

Mean age 42.3 [22-51]

Correlations

95% CI p<0.05

BMI <> waist circumference

BMI <>Fat mass %

BMI<> lean mass %

waist circumference <> hand strength

weight <> push-ups

95% CI p<0.001

BMI <>biceps curl

Tendency of association between

physically active

strength

body composition

Thank you

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ANDO
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