

Rehabilitation in Pycnodysostosis

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A Multidisciplinary Approach to Improving Clinical and Mental Outcomes

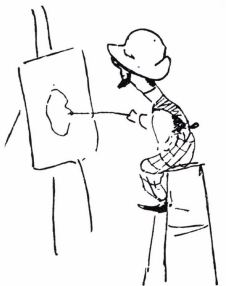
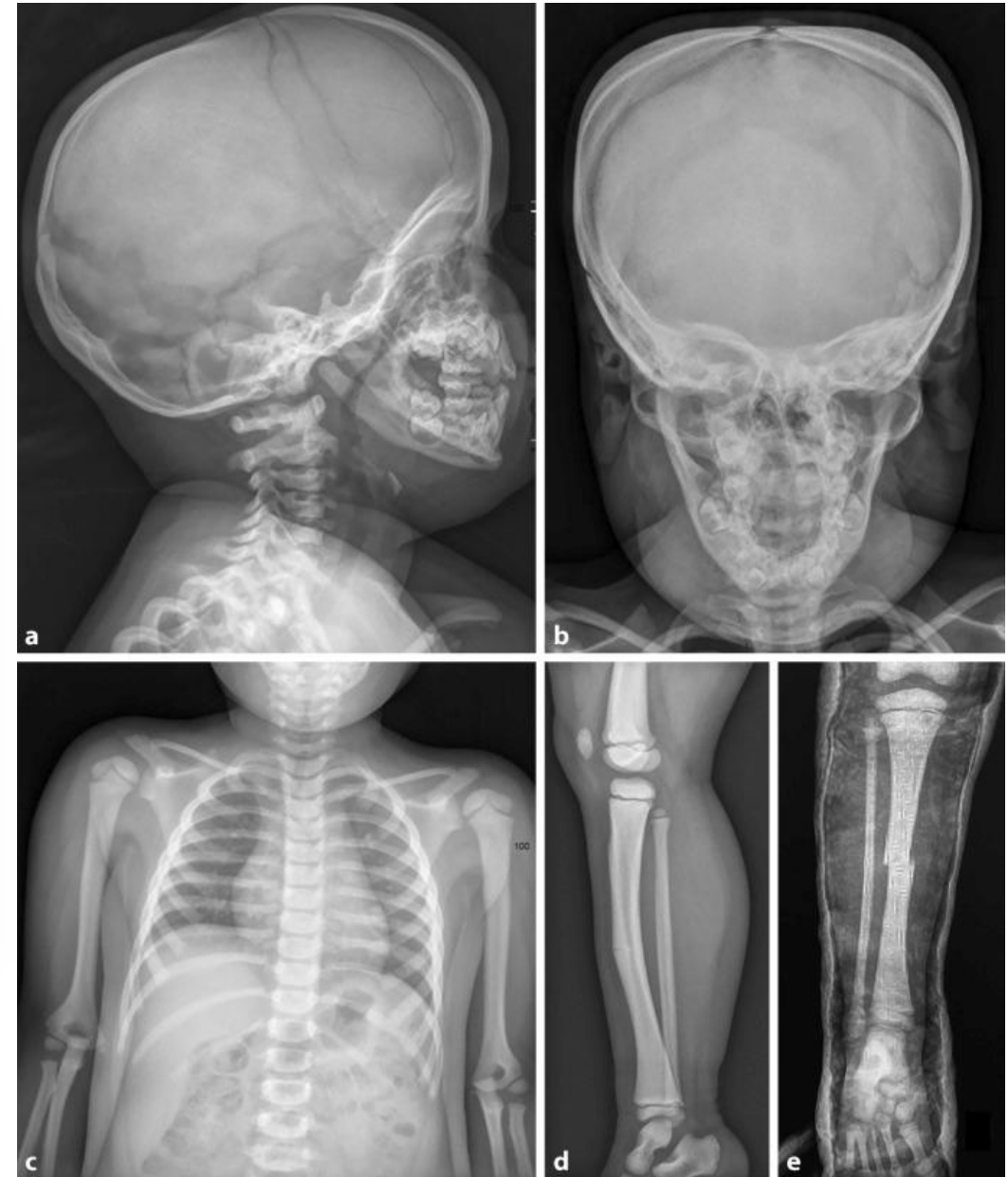
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- Autosossomic recessive skeletal dysplasia
- 1-5 : 1.000.000 births
- CTSK mutation at chromosome 1q21



- bone fragility
- reduced exercise tolerance



Benefits of cardiorespiratory rehabilitation

- Improvement of exercise tolerance
- Dyspnea control
- Weight control
- Improvement of balance and coordination
- Better sleep quality
- Reduction in the number of hospital admissions
- General improvement of QoL and functional status



All images shared with participant permission



Study Rationale

Evidence Gap

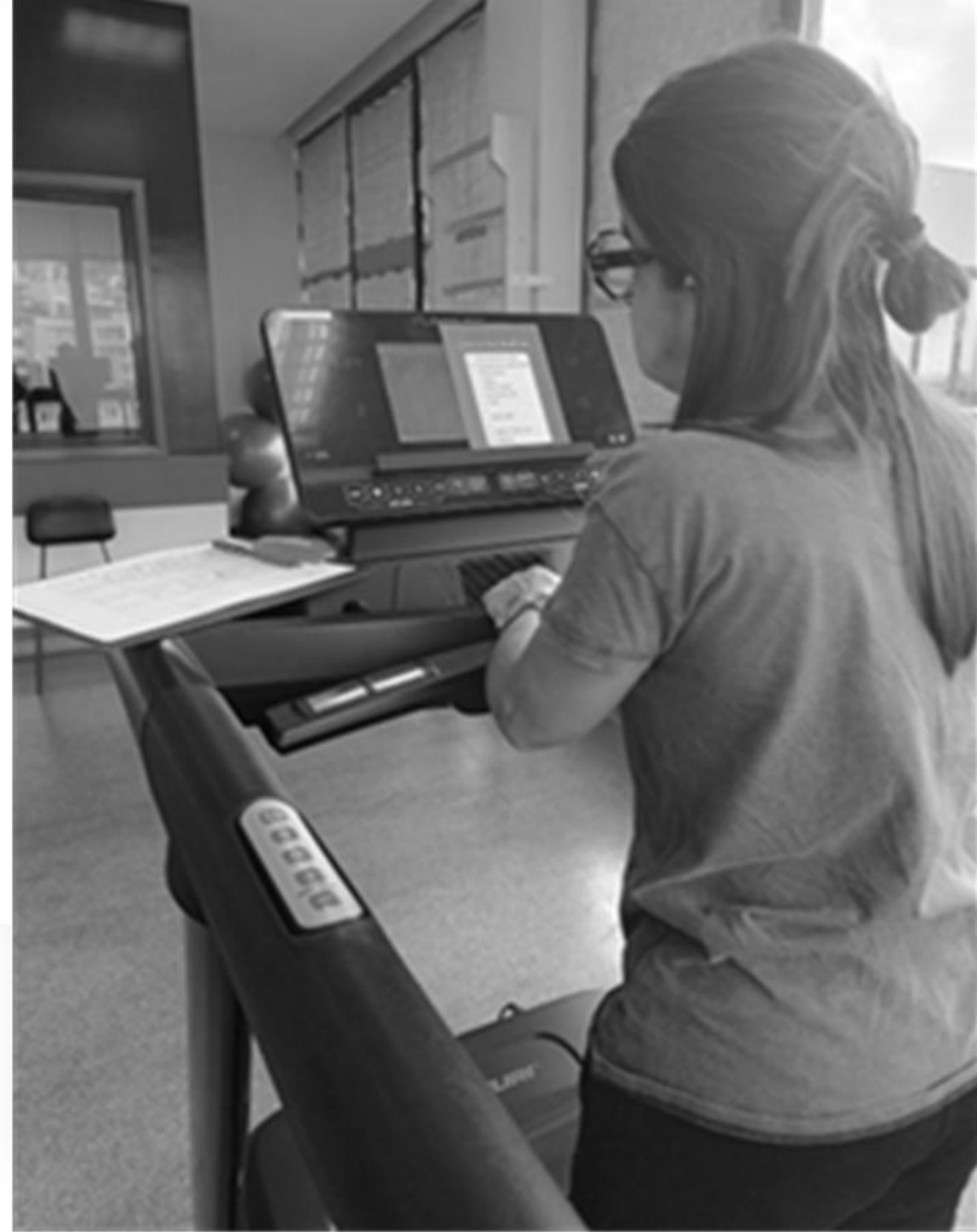
No published cardiorespiratory rehabilitation protocols for PYCD

Clinical Need

Multidisciplinary care recommended but protocols lacking

Potential Benefits

Exercise training proven effective in similar conditions



Case Presentation

- 42-year-old female, 140cm, 38 kg
- Multiple fracture history (femur, tibia, clavicle)
- Recent tibial fracture with surgical stabilization

Baseline limitations

- 6MWT
- pain
- reduced function





Challenges in a patient with PYCD

**Airway
dysfunction**

- Cranial and facial dysmorphia
- Narrowing of pharynx, larynx and trachea
- Laryngotracheobronchomalacia

**Thorax and
lung
compliance
dysfunction**

**Lung
hypoplasia**

**Bone
fractures**



Key Components Rehabilitation protocol

Duration

12-weeks, 2x sessions

Structure FITT



Monitoring

Modified Borg scale, vital signs

Assessments

6MWT, Berg, TUG,
questionnaires

Patient Health Questionnaire-9 (PHQ-9)

Hospital Anxiety and Depression Scale (HADS)

Short Form Survey (SF-36 v2)

Epworth Sleepiness Scale (ESS)

International Physical Activity Questionnaire (IPAQ)



Physical Outcomes

TEST	BASELINE	12 WEEKS	OUTCOMES
6MWT	183 m	491,5 m	↑ 68%
TUG	18s	9s	↓ 50%
Treadmill	12min	25min	↑ 51.6%
Cycle ergometer	6 min	24 min	↑ 400%



Mental Health Outcomes

TEST	BASELINE	12 WEEKS	OUTCOMES
PHQ-9	10 points Moderate depressive disorder	4 points No Depressive disorder	Moderate depression to no depression
HADS	Anxiety 10 Depression 4	Anxiety 5 Depression 1	Decrease in scoring indicating the patient's better condition. The patient appears to be aware of her current state and seems to be highly motivated
SF-36-V2	Mental health= 0 Emotional role = 0 Social Functioning = 25 Pain = 10	Mental health= 40 Emotional role = 25 Social Functioning= 100 Pain = 67	A more reliable score was found and an increase in quality of life related to emotional role, social functioning, and health change perception



Clinical Implications

- First evidence of structured rehabilitation benefits in PYCD
- Safe and effective when properly adapted
- Addresses multiple manifestations simultaneously
- Model for similar skeletal dysplasias



THANK YOU

