

Physiotherapy in Alkaptonuria

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DevelopAKUre

Contents

- Manifestations of AKU on the MSK system
- Physiotherapy in AKU
- The Role of Exercise
- Pain Management
- Resources
- Questions



Musculoskeletal manifestations of AKU

- Perry et al (2006) identified that disability is common and severe in AKU
- Disability correlates well with physical findings of joint restriction and pain
- Chronic pain is complex and far reaching in its impact
 - Depression
 - Anxiety
 - Loss of social role
 - Social isolation

Musculoskeletal manifestations of AKU

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Alkaptonuria

[Giulia Bernardini](#) , [Daniela Braconi](#), [Andrea Zatkova](#), [Nick Sireau](#), [Mariusz J. Kujawa](#), [Wendy J. Intronc](#), [Ottavia Spiga](#), [Michela Geminiani](#), [James A. Gallagher](#), [Lakshminarayan R. Ranganath](#) & [Annalisa Santucci](#)

Nature Reviews Disease Primers **10**, Article number: 16 (2024) | [Cite this article](#)

- AKU is a progressive metabolic disorder characterised by homogentisic acid accumulation leading to
- Cartilage degradation
- Tendon rupture
- Ligament weakening`
- Early severe osteoarthropathy / arthritis
- The musculoskeletal manifestations resemble severe early-onset osteoarthritis and commonly affect the spine, hips, knees, and shoulders.

These physiological changes can lead to

- Pain
- Loss of range of movement and joint stiffness
- Loss of muscle strength and flexibility
- Reduced overall function including mobility, gait and balance
- Deconditioning and reduced exercise tolerance
- Possible need for arthroplasty
- Impact on personal independence
- Bernardini et al (2024)

Physiotherapy in Alkaptonuria

- The evidence base for physiotherapy in alkaptonuria (AKU) remains limited,
- Largely expert consensus, observational studies, biomechanical research, registry data, and case reports rather than gold standard randomised controlled trials
- Contemporary literature consistently supports **ongoing physiotherapy and appropriately prescribed exercise as core components of multidisciplinary AKU management**, alongside pharmacological treatment (particularly nitisinone), pain management, and orthopaedic intervention when required.

THE ROLE OF PHYSIOTHERAPY

- | Detection and diagnosis
- | Assessment
- | Management
 - | Physiotherapy addresses the secondary biomechanical consequences of (stiffness, weakness, reduced mobility) rather than the metabolic defect itself
- | Monitoring/Evaluation
- | Research





The Robert Gregory National Alkaptonuria Centre

Assessment at the NAC

- Questionnaires
 - KOOS, Oxford Shoulder Score, HAQ, SF36
- Physical Assessment
 - ROM axial and peripheral joint, TUAG, functional reach, timed walk, timed sit to stand
- Discussion – advice and education
- Report
- Liaison with your GP
- Quality of Life Reporting



Research Articles

The effects of age and sex on active and passive hip range of motion in individuals with alkaptonuria

Sophie Taylor , Nick Dobbin, Richard Imrich, Jean-Baptiste Arnoux & Lakshminarayan R. Ranganath
Pages 4256-4261 | Received 31 Jul 2024, Accepted 22 Dec 2024, Published online: 08 Jan 2025

 Cite this article  <https://doi.org/10.1080/09638288.2024.2447363>  Check for updates

- 2025 study (**Taylor et al.**)examined hip ROM in 123 individuals with AKU using data from an international (Sonia 2)clinical trial cohort.
- Significant reductions in hip ROM compared with normative healthy populations.
- Progressive limitation with age.
- Marked impairments in hip flexion and abduction.
- Clinical value of ROM measurement as an indicator of disease progression and functional decline.
- ROM assessment should be part of routine rehabilitation monitoring.
- Physiotherapists can use ROM changes to identify deteriorating function and guide interventions.
- Early identification of restriction may allow targeted exercise programmes before severe disability develops

Management

- | Advice and education regarding
 - Maintaining and improving exercise tolerance
 - Recognition of symptom & function changes
 - Pain management
 - Pacing and planning

- | Working with patients, their family and carers to promote self-management and optimise function

- | Liaise with other agencies/services/disciplines to optimise patient management

EXERCISE

Exercise is the single most important thing you can do to help yourself



- **Being physically active is easier than you think, especially if you make activity part of your daily routine**
- Pick activities you like and that fit easily into your daily routine.
- Work out what time is best for you to exercise and stick to it.
- Be active with friends and family to keep you motivated.
- Reduce the amount of time you are inactive during the day.
- The amount of activity you need to do each week depends on your age.



HM Government Guidelines

- **Adults (19–64 years)**
- 1. Adults should aim to be active daily.
- Over a week, activity should add up to at least 150 minutes (2½ hours) of **moderate** intensity activity in bouts of 10 minutes or more
- – one way to approach this is to do 30 minutes on at least 5 days a week.
- 2. Alternatively, comparable benefits can be achieved through 75 minutes of **vigorous** intensity activity spread across the week or combinations of moderate and vigorous intensity activity.



Guidelines for adults

**At least 150 mins
moderate activity a
week**

- [walking](#)
- water aerobics
- ballroom and line [dancing](#)
- [riding a bike](#) on level ground or with few hills
- playing doubles tennis
- pushing a lawn mower
- canoeing
- volleyball

**75 mins of vigorous
activity a week**

- [jogging or running](#)
- aerobics
- [swimming](#) fast
- [riding a bike](#) fast or on hills
- singles tennis
- football
- hiking uphill
- energetic [dancing](#)
- martial arts

HM Government Guidelines

- **Moderate intensity** physical activities will cause adults to get warmer and breathe harder and their hearts to beat faster, but they should still be able to carry on a conversation.
- Brisk walking
- Cycling
- **Vigorous intensity** physical activities will cause adults to get warmer and breathe much harder and their hearts to beat rapidly, making it more difficult to carry on a conversation.
- Running
- Sports such as swimming or football





Adults should undertake
physical activity to improve
muscle strength at least twice
a week

activity t



- Physical activities that **strengthen muscles** involve using body weight or working against a resistance. This should involve using all the major muscle groups.

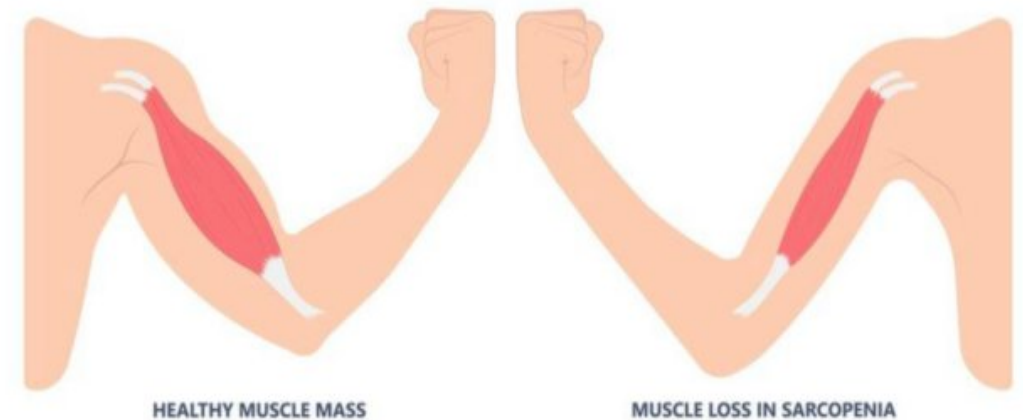
- Examples include:
- Exercising with weights
- Yoga, pilates



- **Muscle strength** is necessary for:
- all daily movement
- to build and maintain strong bones – site specific resistive exercise
- to regulate blood sugar and blood pressure
- to help maintain a healthy weight

Sarcopenia

- Sarcopenia is gradual loss of muscle mass and strength over time
 - Can lead to impairments in mobility and quality of life if not tackled.
-
- Slow, progressive skeletal muscle disorder
 - Loss of muscle mass and strength
 - Typically begins in midlife, with muscle mass declining on average by around 1–2% per year from about age 50
 - Risk factors including a sedentary lifestyle, smoking and alcohol use, poor dietary intake, multimorbidity, polypharmacy, recent extended hospital stays and social isolation
 - Not an inevitable but modifiable condition
 - Supervised resistance training can lead to improvements in muscle function and personal independence
 - Early intervention and education around the modifiable risk factors that can accelerate the process such as nutrition, alcohol and smoking can significantly enhance disease prevention. (BSJM, 2026)

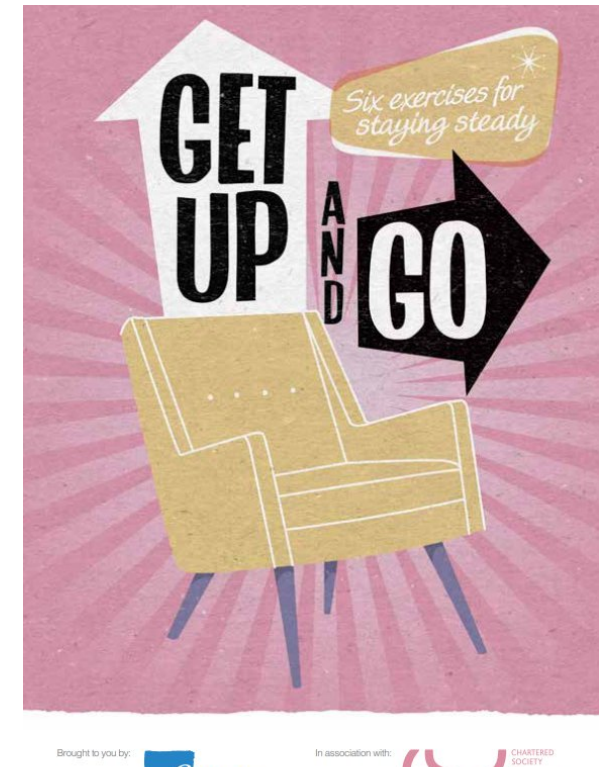
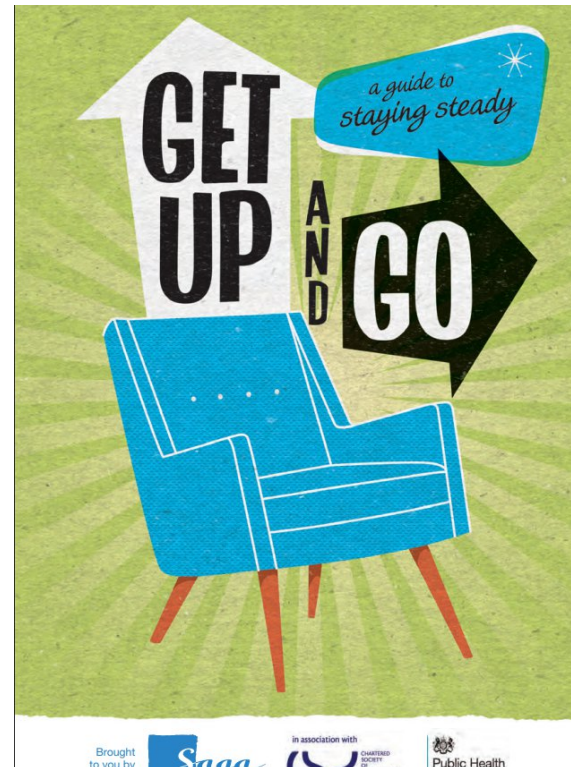


Fall Prevention

Try to break up long periods of sitting with light activity, as sedentary behaviour is now considered an independent risk factor for ill health, no matter how much exercise you do.

Regular exercise on at least two days of the week helps adults at risks of falls to improve strength, balance and co-ordination

Examples include walking tai chi and line dancing



Benefits of Exercise

- Increased Flexibility
- Increased Strength
- Decreased Stiffness
- Better Cardiovascular Fitness
- Improved Balance
- Reduced risk of falls
- Improved Posture
- Decreased Pain
- Improved function & physical ability
- Exercise results in improved vitality, physical role and mental health scores (Perry et al, 2006).





Disadvantages of Exercise

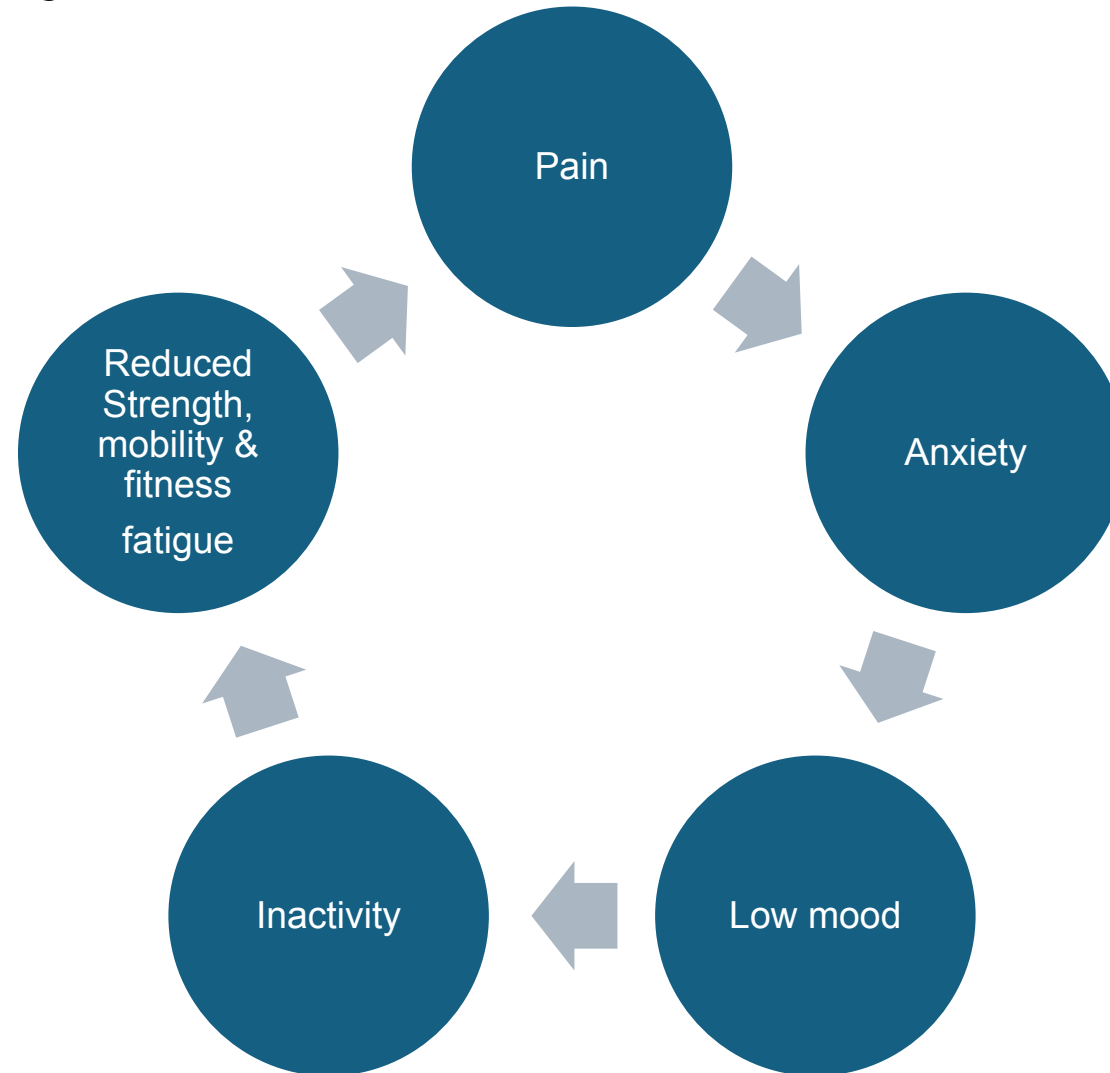


Barriers to Exercise

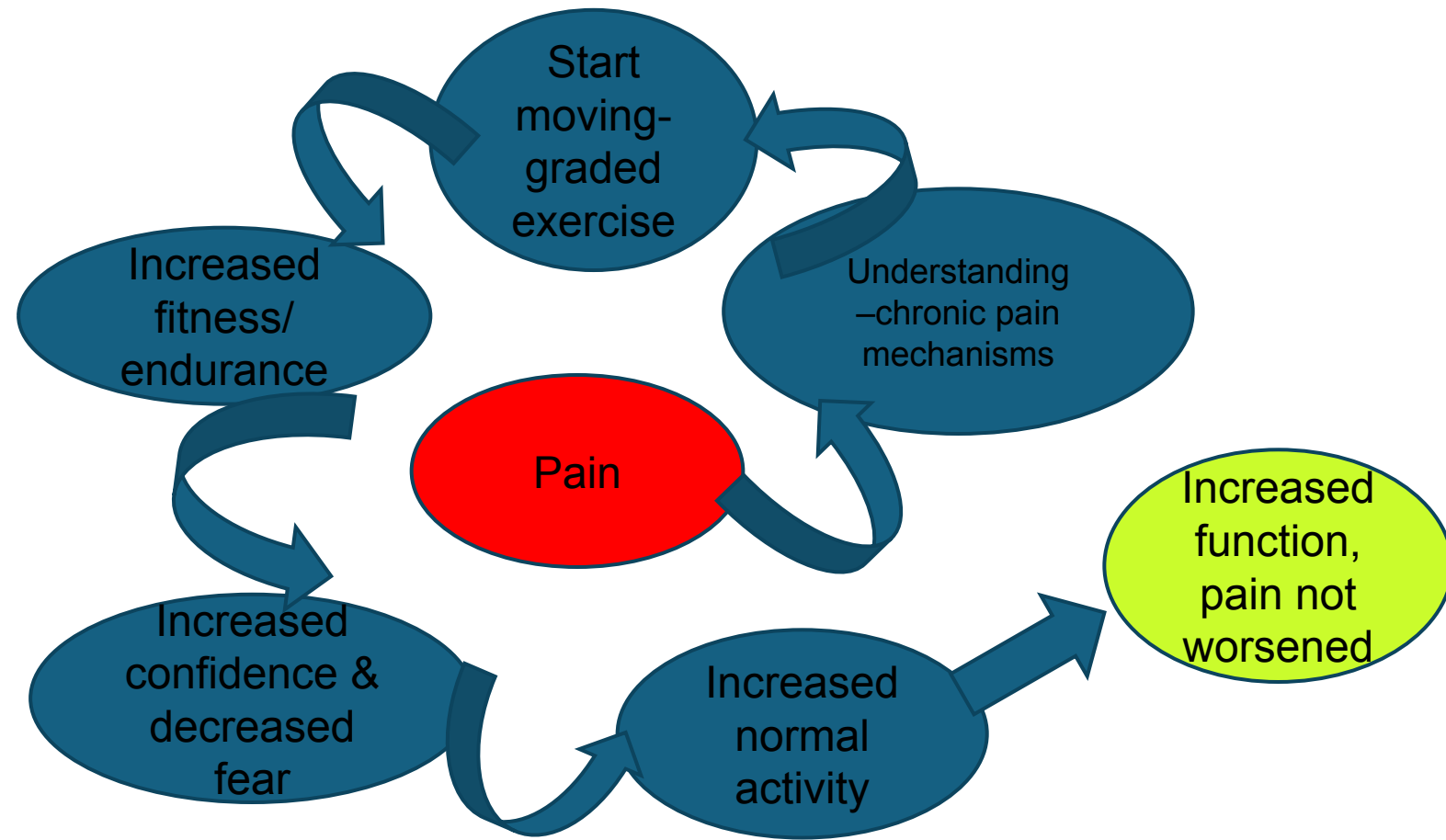
- Pain
- Fear of injury
- Reduced energy levels
- Fatigue
- Motivation
- Lack of self-efficacy
- Lack of confidence
- Lack of time- competing priorities e.g caring responsibilities
- Work
- Social support and isolation
- Cultural and language barriers
- Cost
- Facilities
- Environment, weather



The pain cycle



Breaking the cycle





What type of exercise?

- Stretching
- Strengthening Exercises
- Breathing Exercises
- Chair based exercise
- Walking / Nordic walking
- Jogging
- Cycling
- Pilates / Yoga / Tai Chi
- Swimming / Water aerobics/Hydrotherapy
- Golf
- Wii Fit
- Gym classes e.g. body balance, body pump, abs strengthening

HYDROTHERAPY

- Exercise in water gives you freedom of movement!



Activities to generally avoid



Contact sports



High impact running



**Repetitive high load
exercise esp on spine, hips
and knees**



**Maintaining joint mobility
movement and muscle
strength and power is
important , but excessive
loading may accelerate
symptoms in already
compromised joints.**

Getting started



- Discuss your plans with your Physiotherapist
- Decide what you want to achieve
- Set a SMART goal
- Start low and go slow
- Don't expect results overnight
- Warm up and stretch
- Cool down and stretch

- You will feel stiff and sore – Delayed Onset Muscle Soreness (DOMS) after exercise- particularly if you haven't exercised for a while
- If you feel you have done too much, reduce the intensity and build up again gradually
- Do something you enjoy which is fun – you'll look forward to it and stick to it
- Involve someone else
- **A small amount of activity regularly can make a huge difference**
- **'exercise snacks'**



Resources

- <https://www.nhs.uk/live-well/exercise/about-our-videos/>
- <https://www.nuffieldhealth.com/about-us/our-impact/healthy-life/joint-pain-programme>
- https://www.arthritis-uk.org/media/edyho5vv/a5-keep-moving-booklet_digital-1.pdf
- <https://www.paintoolkit.org/>

RECOMMENDATIONS



Comprehensive clinical assessment



Regular re-assessment



Bespoke exercise programme



Swift access to advice during a flare-up or deterioration

Thank you for listening

