

# ALBINISM, GOUT

BY

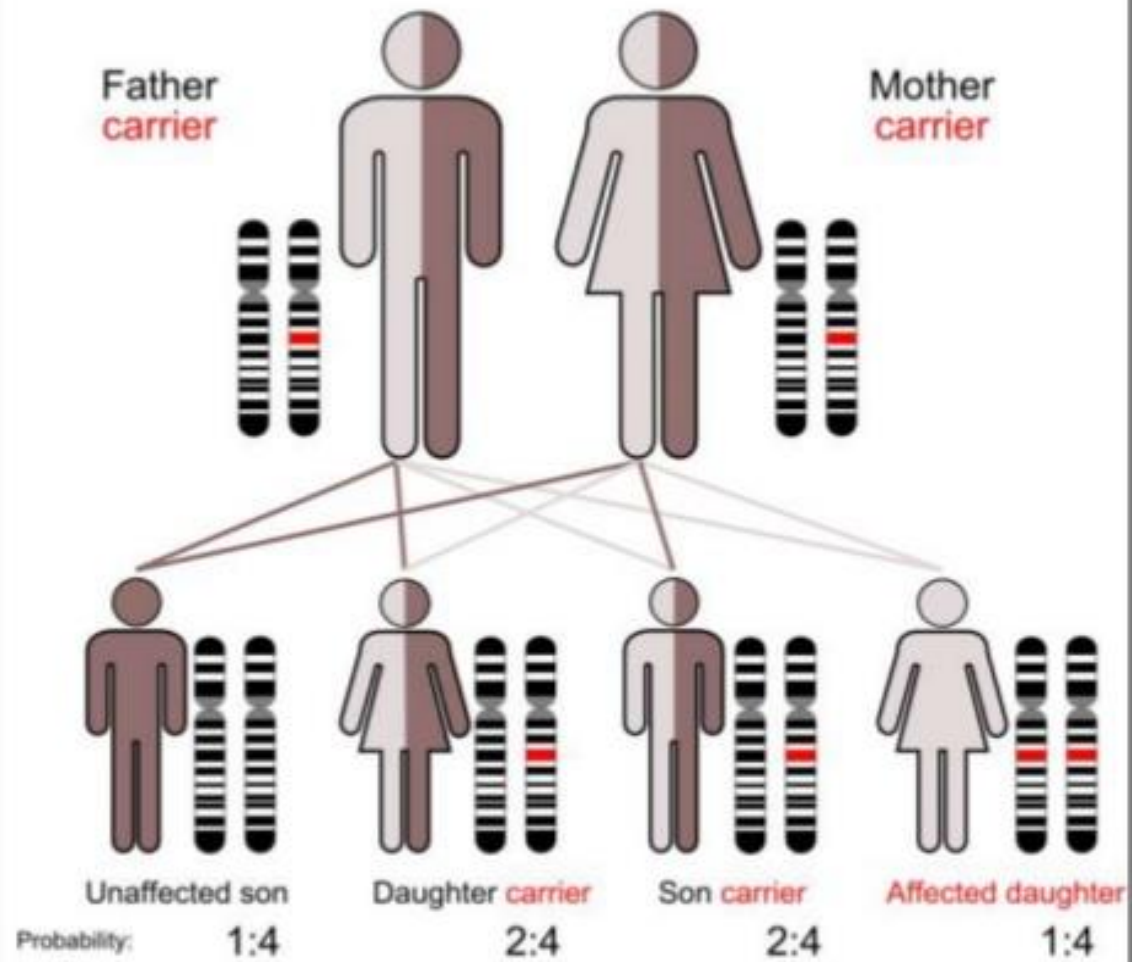
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## WHAT IS ALBINISM?

- ⦿ Albinism is a recessive trait that is normally inherited from two heterozygous parents
- ⦿ Also called achromia, achromasia, or achromatosis
- ⦿ Deficit in the production in melanin and by the partial or complete absence of pigment in the skin, hair and eyes
- ⦿ This hereditary disease can be found in humans (affecting all races), mammals, birds, fish, reptiles and amphibians



## Autosomal recessive



## SYMPTOMS

- ◉ Absence of color in the hair, skin, or iris of the eye
- ◉ Lighter than normal skin and hair
- ◉ Patchy, missing skin color
- ◉ Crossed eyes (strabismus/heterotropia)
- ◉ Light sensitivity (photophobia)
- ◉ Rapid eye movements (nystagmus/dancing eyes)
- ◉ Vision problems, or functional blindness

## DEGREE OF PIGMENTATION

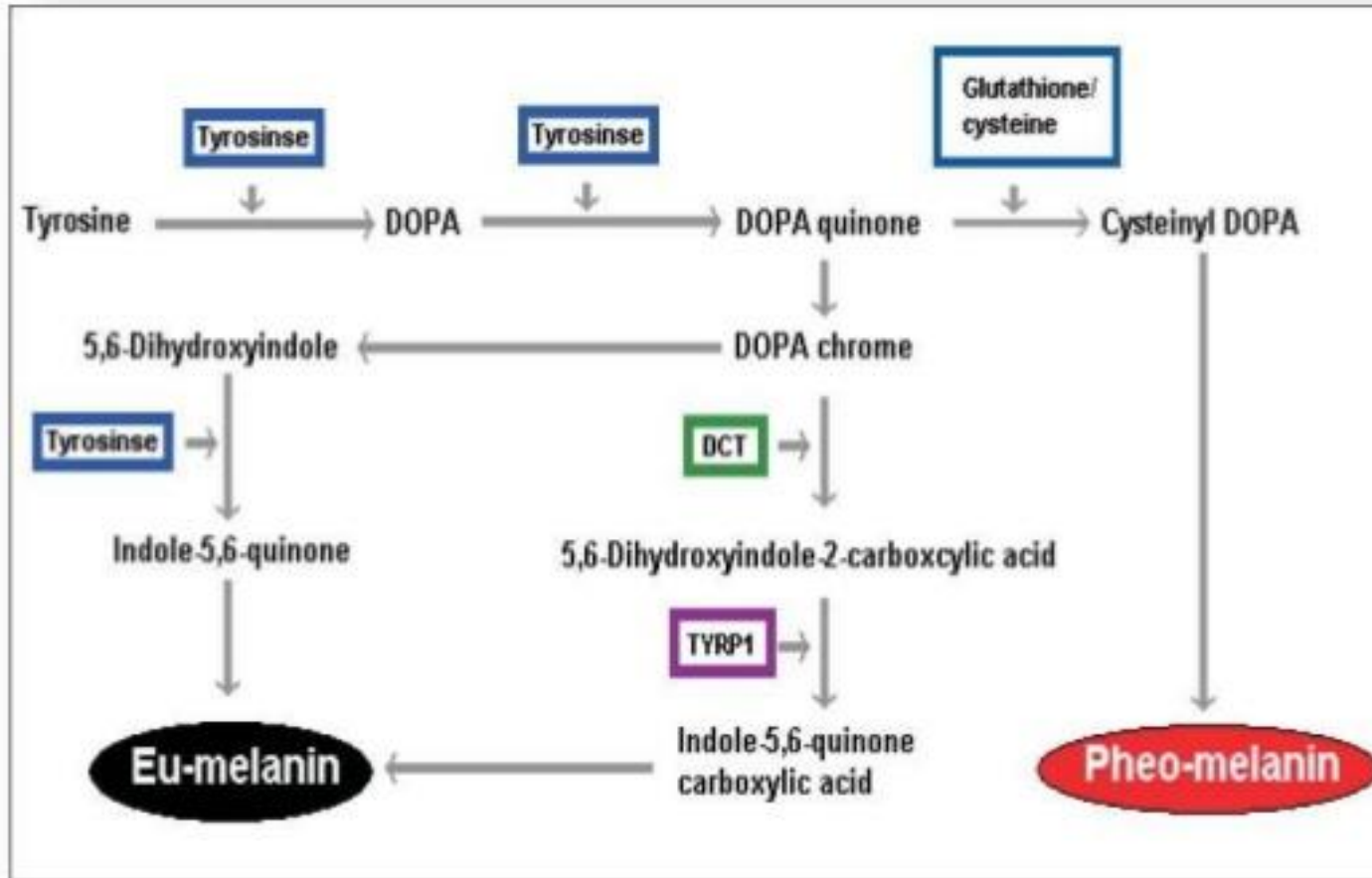
- ◉ complete absence of melanin is called an albino.
- ◉ small amount of melanin is described as albinoid.



## PRECAUTIONARY MEASUREMENTS

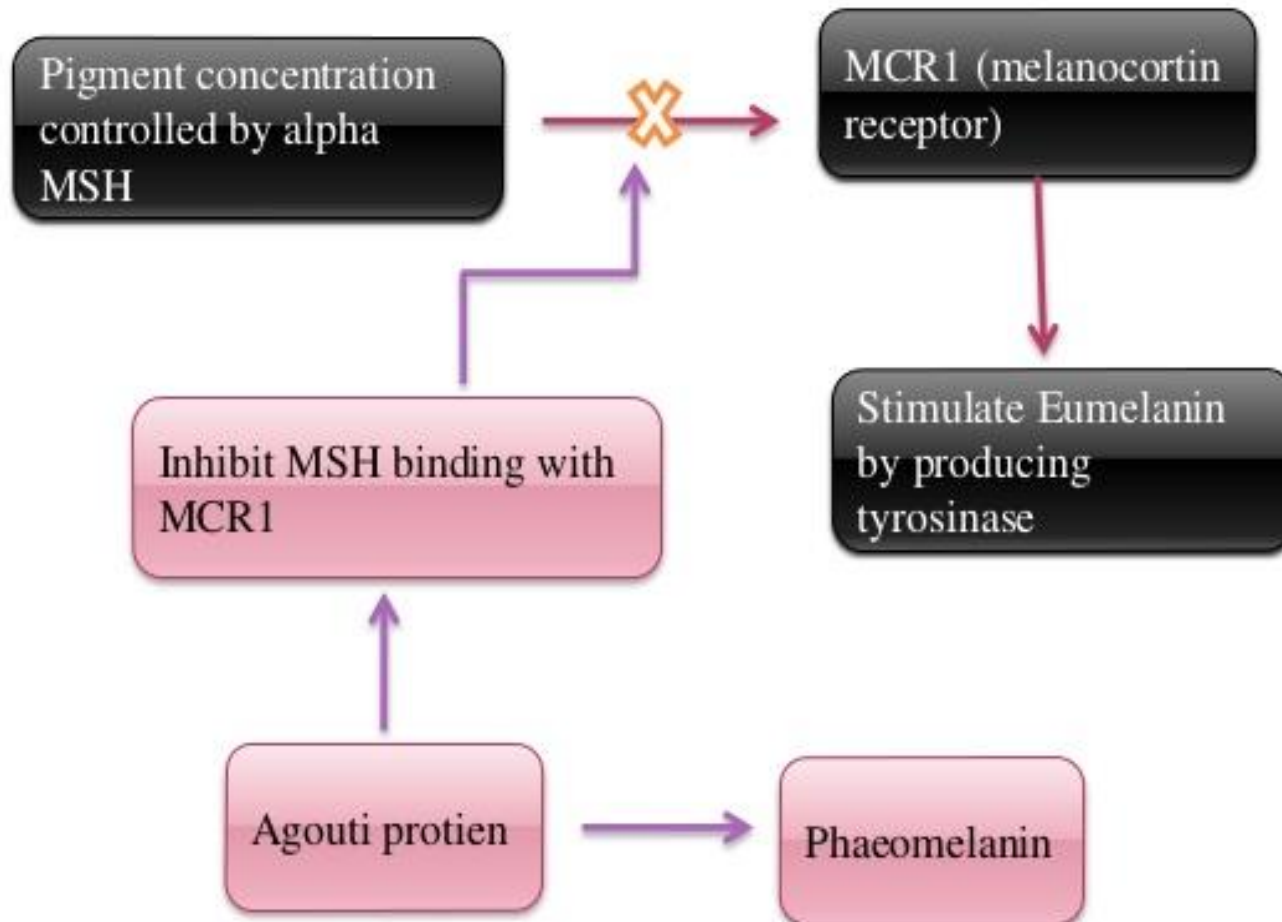
- ◉ Reduce sunburn risk by avoiding the sun, using sunscreen, and covering up completely with clothing when exposed to the sun.
- ◉ Sunscreen should have a high sun protection factor (SPF).
- ◉ Sunglasses (UV protected) may relieve light sensitivity.

# MELANIN PRODUCTION PATHWAY



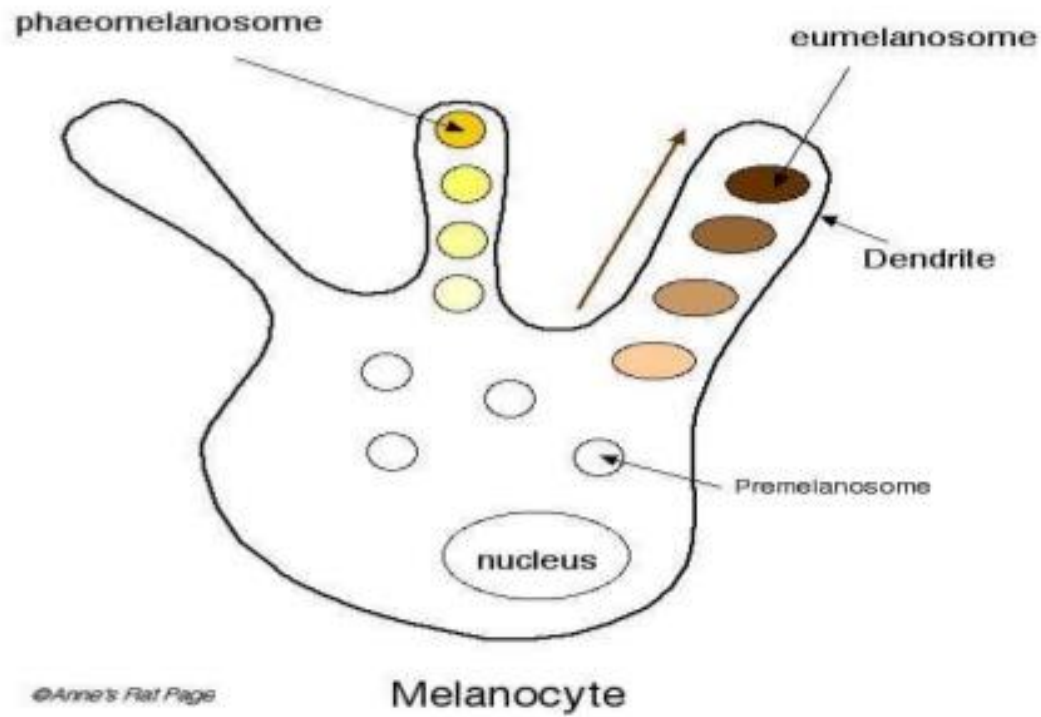


## MUTATION IN MELANOSOME



## LESS PIGMENT DEPOSITION

- Transport system of melanosome is effected

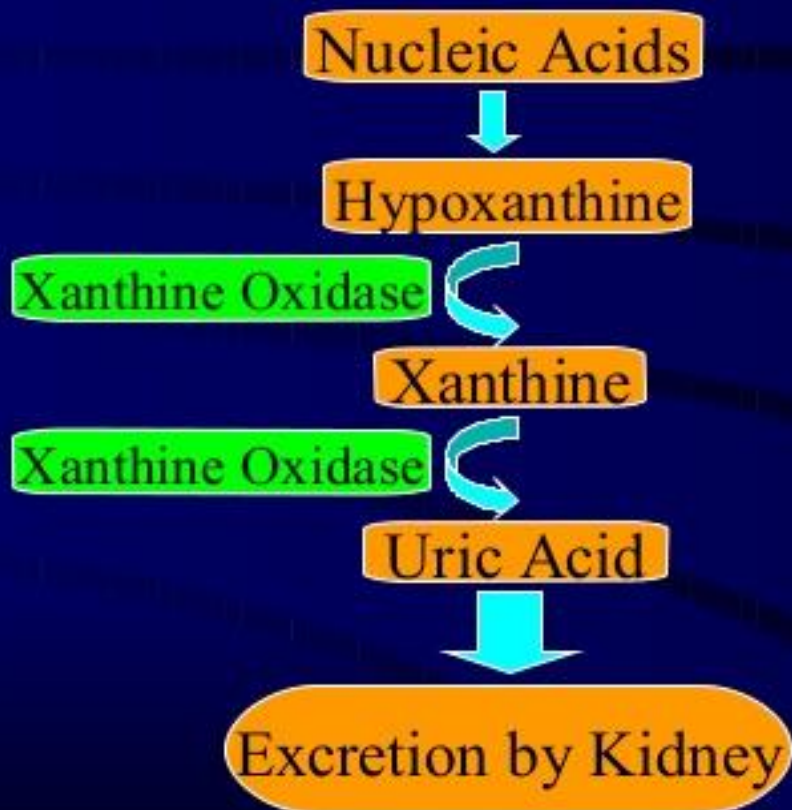


# GOUT

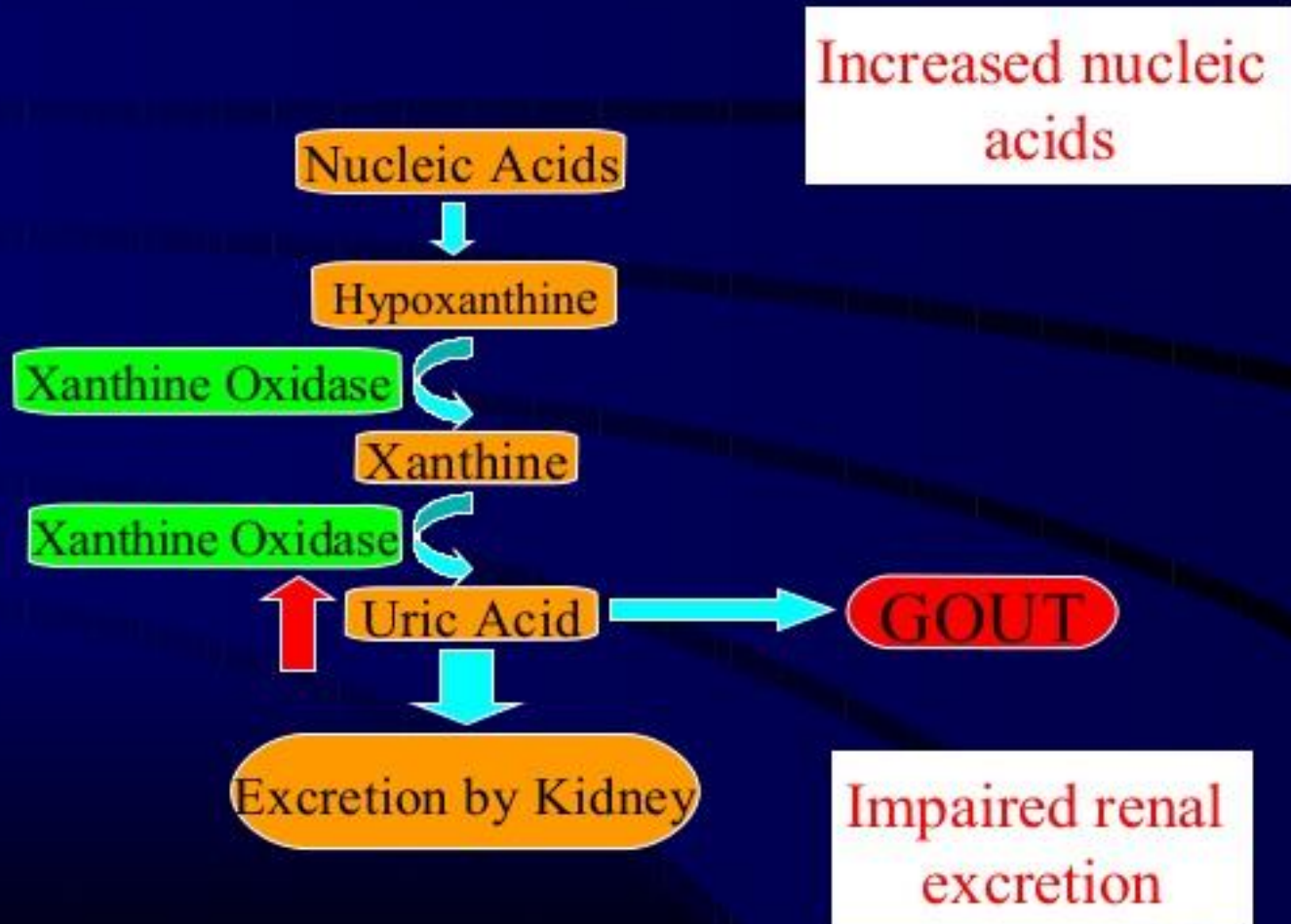
## What is Gout?

- Crystal arthritis
- Deposition of uric acid crystals in joints
- Presents with acute monoarticular arthritis in 90 % of patients.
- Men > Women
- Onset 40-60 years
- Very treatable
- Untreated, may lead to chronic tophaceous gout.

# Uric Acid Metabolism



# What goes wrong?





# Predisposing Factors

- Gender- male > female
- Family History
- Previous attack
- Body size - large > small
- Alcohol
- Diet – purine rich
- Diuretics
- Causes of high cell turnover e.g. psoriasis, chemotherapy
- Renal insufficiency



# Investigations

- Joint aspiration - polarising light microscopy for crystals
- FBC - white cells raised
- ESR - raised
- X-ray - generally normal
- Serum urate

**HIGH SERUM URATE DOES NOT NECESSARILY  
MEAN THE PATIENT HAS GOUT!**

# Treatment of Acute Attack

**AIM- to reduce symptoms**

- NSAID
- Colchicine
  - GI side effects,
  - Most effective in 1<sup>st</sup> 24 hours
- Corticosteroid
  - If NSAID and colchicine contraindicated

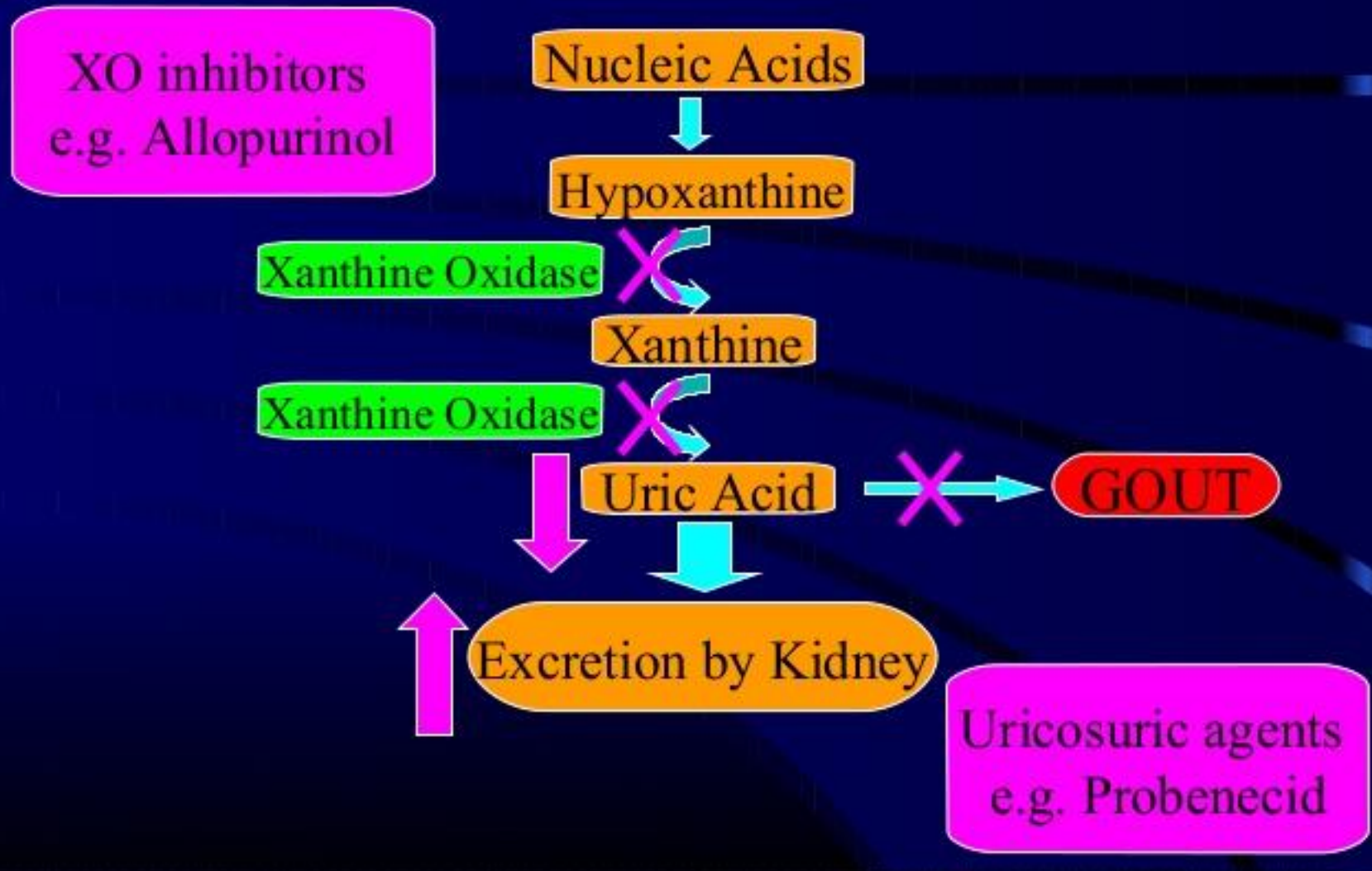
# Prevention of Further Attacks

## AIM - Prophylaxis

- Lifestyle changes – to minimise risk factors
- Drug therapies
  - Xanthine oxidase inhibitors e.g. Allopurinol
  - Uricosuric agents e.g. Probenecid
- Need to be taken for life
- Can precipitate an acute attack therefore give together with NSAID or colchicine for 2 months.



# Site of drug action



# Chronic Tophaceous Gout

- If gout untreated
- Other joint involvement
- Formation of tophi- collection of crystals in soft tissues
- Bone erosion at joints- ‘punched out’ erosions on x-ray.

# Key Points

- Gout is due to deposition of urate crystals in joints leading to inflammation.
- Diagnosis is by history and polaroid microscopy of synovial fluid.
- High serum uric acid does NOT mean the patient has gout.
- Gout is one of the most treatable arthritic conditions.
- Treat acute attack and then consider prophylaxis of future episodes.
- If untreated, can develop into chronic tophaceous gout.