Hartnup’s Disease

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Etiology

- Defective - Neutral Amino acid Transporter
  - **Mainly = Tryptophane**
- Genetic Disorder.
- Autosomal Recessive Disorder.

- Glycine
- Alanine
- Valine

Neutral Amino Acids:

- Leucine
- Isoleucine
- Proline

- **Aromatic:**
  - Phenylalanine
  - **Tryptophan**
  - Tyrosine.
- **Sulfur-containing:**
  - Methionine
  - Cysteine.

- **Hydroxyl/Amide-containing:**

Pathogenesis

- Defective transport of **Neutral Amino Acids** across
  - Renal tubular epithelium
  - Intestinal mucosal epithelium.
- No Intestinal absorption of Neutra Amino Acid
  - Same will be deficient in Blood
- No Renal Re-absorption of Neutra Amino Acid
  - Amino acids are excreted in the urine.

Clinical Features

- Due to Tryptophane Deficiency
  - Niacin deficiency
  - Pellagra
- Pellagra like symptoms = **3D**
  - Dermatitis
  - Diarrhoea
  - Dementia
  - Ataxia.

AminoacidUria Niacin deficiency

Ataxia NUtral Amino Acid Deficiency

Hereditary **HARTNUP** Pellagra

Photosensitive Rash

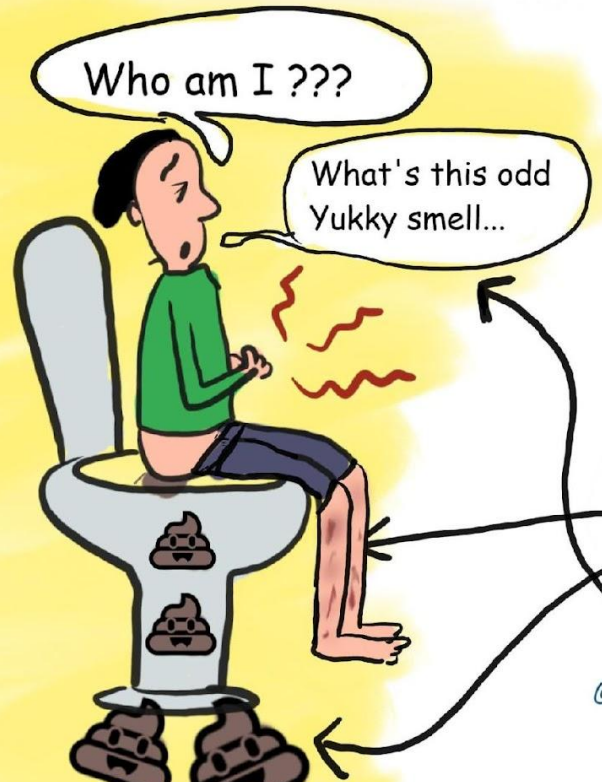
Protein diet helps

Autosomal Recessive

Transport of Neutral AA defect

(at PCT and Enterocytes)

Obermeyer test- indole group



Tryptophan Deficiency

Niacin deficiency

Pellagra

3D

Dermatitis

Diarrhea

Dementia

Serotonin deficiency

Neurological Problem

Ataxia

Diagnosis

Increased amino acid levels in Urine

Treatment

Protein diet

Niacin supplements

Creative-Med-Doses

Diagnosis

- Aminoaciduria
- Increased excreation of Indole compounds
  - Stool
- It is detected by the Obermeyer test.
- Indican >> indoxyl with HCL >> indigo
  - Blue colored compound with ferric chloride.

Treatment

- High Protein Diet
- Essential Amino Acid
- Niacin

Fanconi syndrome

- Renal tubule disorder
- Fail to reabsorb essential nutrients
  - Glucose > 3P (Polyuria , Poly Dispsia , Poly Phagia)
  - Amino acids > Growth Retard
  - Phosphate > Osteoporosis
  - Bicarbonate > Metabolic Acidosis
- All substance are osmotically active
  - Diuresis > Loss of Water > Lost of Micro-Nutrients
    - Decrease Magnesium , Phosphate, Calcium , Sodium , Potassium

Hartnup Disease

Etiology

- Genetic Disorder
- Autosomal Recessive

Pathogenesis

- Failure of Amino Acid (Non-Polar Amino acid) Transport
  - In Intestine & Kidney
- Decrease absorption of Tryptophan
- Decrease Reabsorption of Tryptophan
- Increase Loss of Tryptophan

Clinical Feature

- Pellagra

Fanconi Syndrome

Etiology

- Congenital or Acquired

Pathogenesis

- Inadequate absorption in Proximal Renal Tubules
- Reabsorption of Amino acid ,Glucose, Uric acid , Phosphate , Bicarbonate

Clinical Feature

- Polyuria , Polydipsia & dehydration
- Hypophosphatemia
- Rickets.
- Osteomalacia (in adults)
- Growth failure
- Metabolic Acidosis
- Hypokalemia
- Hyperchloremia

1). In Hartnup disease, Urinary indican increased due to:

A. excess hepatic conversion of tryptophan to indican

B. excess bacterial degradation of unabsorbed tryptophan in intestine

C. excess renal conversion of tryptophan to indican

D. All of above.

2). Which is correct about urinary biochemical in Hartnup disease?

A. Increased Neutral amino acid and Glucose in Urine

B. Increased Indican and Glucose in Urine

C. Increased Neutral amino acid and Tryptophan in Urine

D. Increased Indican and Glucose in Urine

3). Pellagra like symptoms in Hartnup disease is due to

A. defect in absorption of tryptophan and niacin

B. defect in re-absorption of tryptophan and niacin

C. defect in conversion of Tryptophan to Niacin

D. All of above

E. None of above

4). In Hartnup disease, niacin deficiency develops primarily because:

A. Tryptophan is excessively converted to serotonin

B. Tryptophan cannot be absorbed from intestine

C. Tryptophan degradation is increased in liver

D. Niacin absorption is impaired

5). Niacin supplementation can improve symptoms in Hartnup disease. Which biochemical reason is correct about it ?

A. It bypasses defective renal transport

B. It restores tryptophan absorption

C. It compensates for decreased endogenous niacin synthesis

D. It reduces indole formation

6) In fanconi syndrome , reason for rickets and osteomalacia is mainly due to

A. decrease magnesium

B. decrease calcium

C. decrease phosphorus

D. decrease vitamin D