AMINO ACID
FIRST STAGE

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MEDICAL BIOCHEMISTRY

AMINO ACID

AMINO ACIDS ARE THE BUILDING BLOCKS OF PROTEINS.

EACH AMINO ACID CONTAINS: AMINO GROUP (NH_2),

CARBOXYL GROUP (COOH), HYDROGEN ATOM, AND SIDE CHAIN (R GROUP). THESE GROUPS DETERMINE THE

CHEMICAL BEHAVIOUR OF AMINO ACIDS.

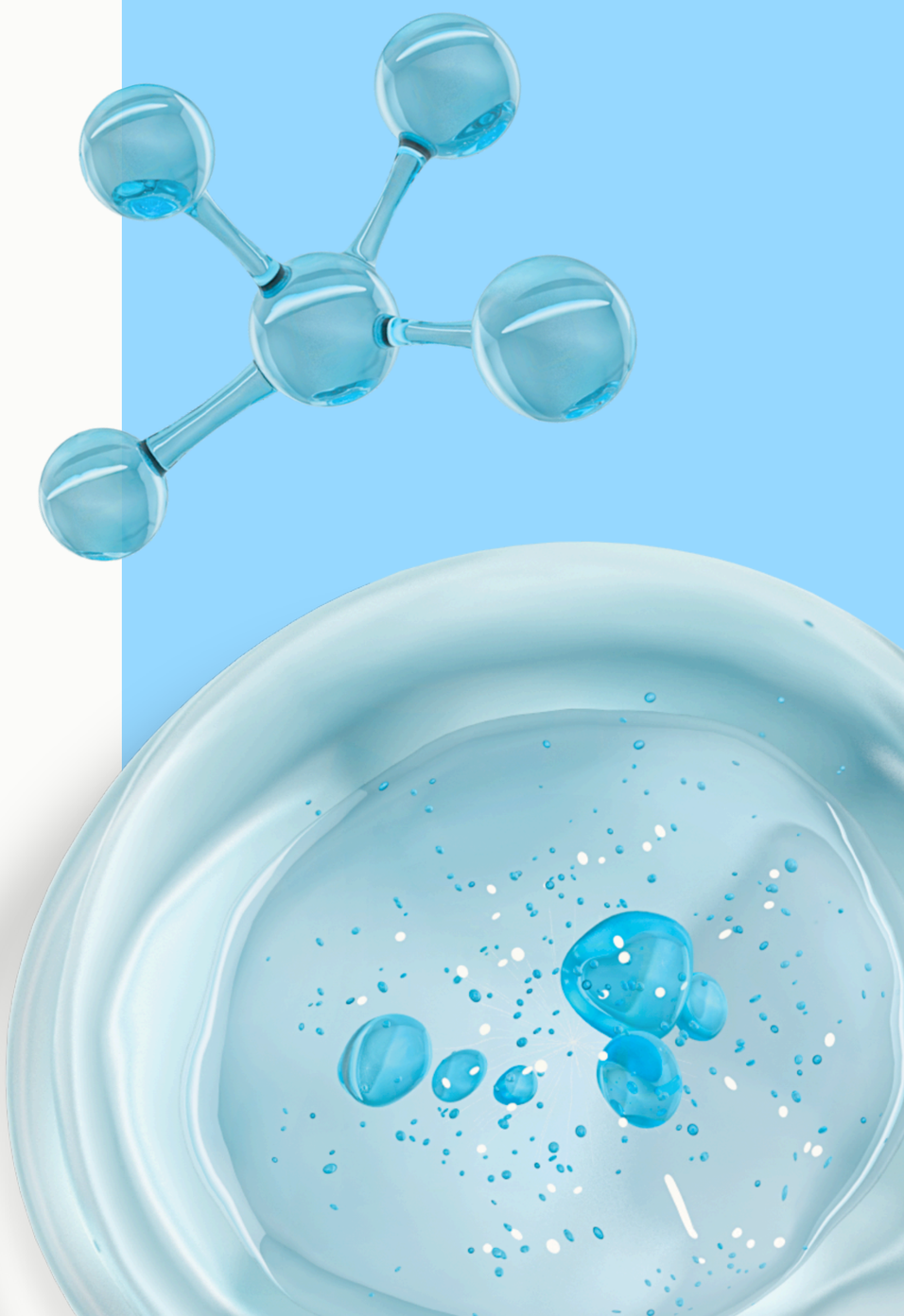
Classification of Amino Acids :

amino acids are classified into two types:

1. Essential amino acids

2. Non-essential amino acid.

this classification depends on whether the body can synthesize them or not.



ESSENTIAL AMINO ACIDS :

Essential amino acids: cannot be synthesized by the body and must be obtained from diet.

Examples: Leucine, Isoleucine, Lysine, Methionine, Phenylalanine, Threonine, Tryptophan, Valine, Histidine.

NON-ESSENTIAL AMINO ACIDS :

Non-essential amino acids: can be synthesized in the body and do not need to be obtained from diet.

Examples: Alanine, Glycine, Aspartate, Glutamate, Serine, Proline.

AMINO GROUP :

the amino group (NH_2): contains a nitrogen atom, has a lone pair of electrons, and participates in several biochemical reactions.

REACTIONS DUE TO THE AMINO GROUP :

important reactions include:

1. Transamination

2. Deamination

3. Formation of carbamino compounds. these reactions are important in amino acid metabolism.

TRANSAMINATION :

Definition: transfer of the amino group from an amino acid to a keto acid. General reaction: Amino acid + α-keto acid produces New amino acid + new keto acid.

Enzymes and Cofactor :

Transamination requires:

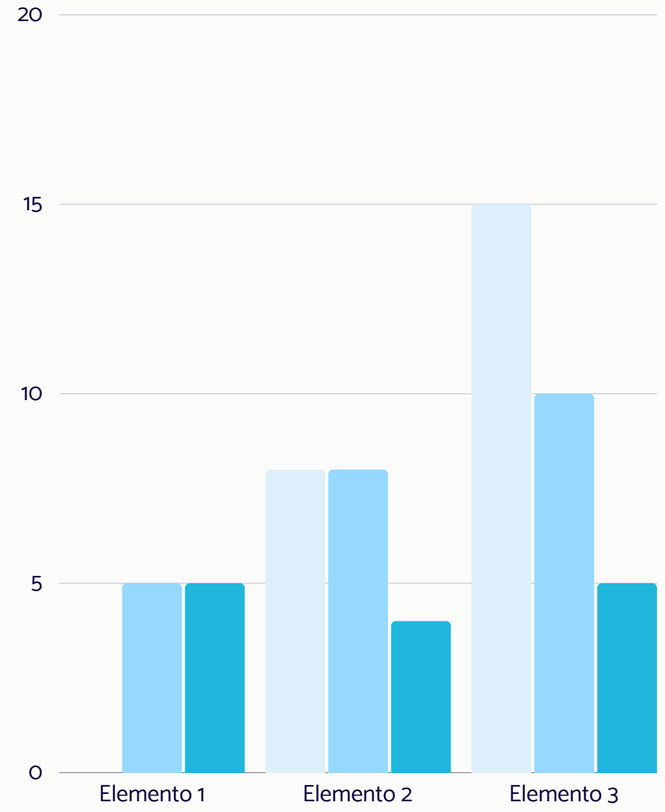
Enzymes: Aminotransferases.

Examples: ALT and AST.

Cofactor: Pyridoxal phosphate (PLP) derived from Vitamin B6.

Deamination :

Definition: removal of the amino group from an amino acid. Result: Amino acid produces Keto acid + Ammonia (NH_3). occurs mainly in the liver.



Carbamino Compound Formation :

amino groups can react with carbon dioxide (CO_2). Example: Hemoglobin + CO_2 produces Carbaminohemoglobin. this reaction contributes to CO_2 transport in blood.

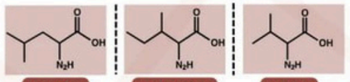
MAPLE SYRUP URINE DISEASE

PMF IAS

- Gene therapy offers new treatment options for **Maple Syrup Urine Disease (MSUD).**

MSUD

1. A rare **Genetic disorder** caused by **BCKDH** deficiency, **preventing** the **metabolism** of **BCAAs** like leucine, isoleucine, and valine.



2. **Inheritance Pattern:** **Autosomal recessive;** requires mutated genes from both parents.

Types of MSUD

Classic MSUD
Most severe and common; symptoms emerge within days of birth.

Intermittent MSUD
Triggered by illness or stress; higher BCAA tolerance than classic MSUD.

Intermediate MSUD
Milder than classic; symptoms appear between 5 months and 7 years.

Thiamine-Responsive MSUD
Improves with high-dose B1 and dietary restrictions.

Traditional Treatment Approaches

Dietary Management
Lifelong **low-protein diet** to restrict BCAA intake.

Liver Transplant
BCKDH enzyme therapy offers a cure but involves surgical risks.

Symptoms of MSUD

Distinct Odor
Sweet, maple syrup–like odor in urine, sweat, or earwax.

Neurological Symptoms
Lethargy, irritability, and poor feeding in infants.

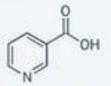
Severe Complications
If untreated, can cause seizures, coma, or death

Advancements in Gene Therapy

Recent Developments
Adeno-associated viral (AAV) vector based gene therapy delivers functional BCKDHA and BCKDHB genes.

Adeno-Associated Viral (AAV) Vector:
AAVs are favored in gene therapy for low pathogenicity and efficient gene delivery.

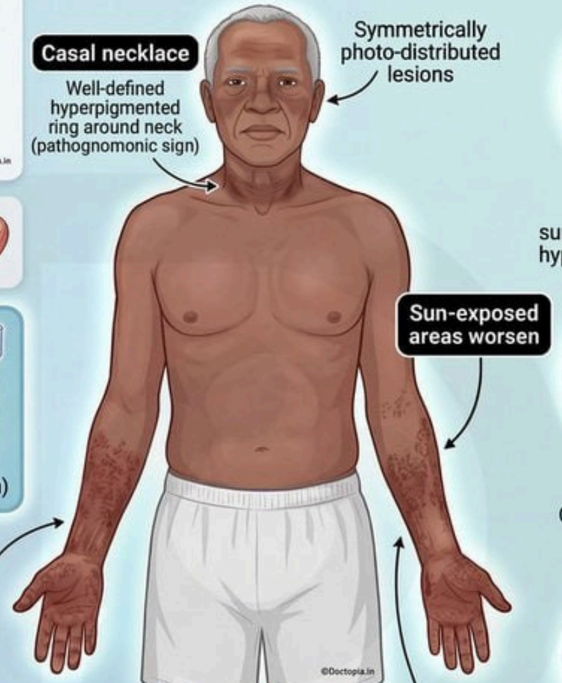
PELLAGRA



Niacin (Vitamin B3)
→ Required for NAD⁺/NADP⁺ synthesis

Deficiency causes impaired cellular respiration

Mnemonic badge:
3 Ds = Dermatitis + Dementia + Diarrhea
(with optional 4th D: Death)



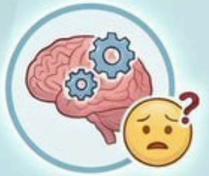
Atrophic Glossitis: beefy red, smooth, inflamed appearance

Causes:
- malnutrition
- chronic alcoholism
- isoniazid therapy (depletes B6)
- tryptophan deficiency (e.g., Hartnup Disease, Carcinoid Syndrome)

THE 3 Ds

1. Dermatitis
sun-exposed symmetric hyperpigmented lesions

2. Diarrhea
GI mucosal atrophy, malabsorption



3. Dementia
Cognitive decline, hallucinations, memory loss

Treatment:
Niacin supplementation

Diagnostic clue:
Casal necklace = pathognomonic pattern

AminoacidUria Niacin deficiency

Ataxia NUtral Amino Acid Deficiency

Hereditary **HARTNUP** Pellagra

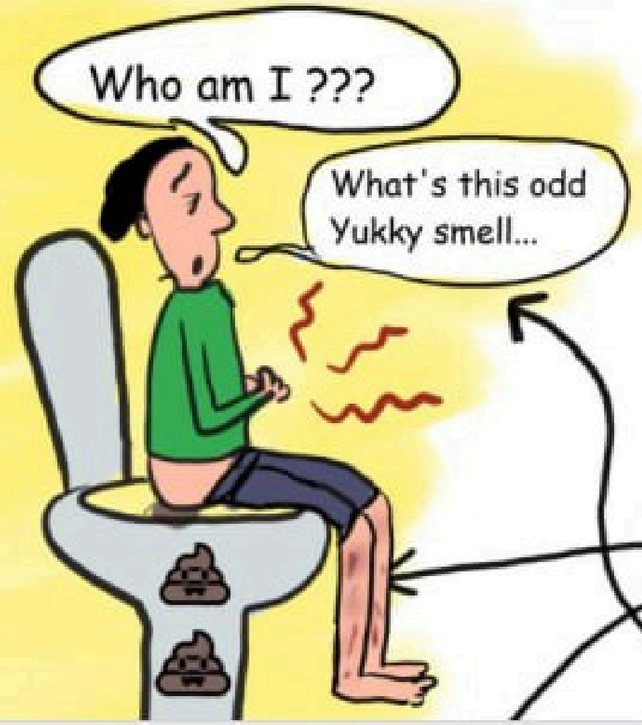
Photosensitive Rash

Protein diet helps

Autosomal Recessive

Transport of Neutral AA defect

(at PCT and Enterocytes)



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

THANK YOU FOR YOUR ATTENTION !