Hartnup Disease

Hartnup disease is an autosomal recessive metabolic disorder caused by a defective neutral amino acid transporter on renal and intestinal epithelial cells. The most important amino acid affected by the poor absorption is nonpolar tryptophan. This amino acid is particularly important in the body because it is a precursor for the synthesis of serotonin, melatonin, and niacin. A defect of this transporter causes decreased absorption in the gut and increased tryptophan excretion in urine. Tryptophan deficiency can lead to pellagra, resulting in diarrhea, dermatitis, and dementia, often preceded by a period of poor nutrition.

PLAY PICMONIC

Pathophysiology

Autosomal Recessive

Recessive-chocolate

This metabolic disorder is inherited in an autosomal recessive fashion.

Neutral amino acids

Neutral-Swiss-guy and Amigo Acidic-lemon

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Defective Transporter

Broken Transporter-enzyme

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Renal and Intestinal cells

Kidney and Intestine

This amino acid transporter is found on renal and intestinal epithelial cells.

Signs and Symptoms

Causes tryptophan excretion in urine

Tri-toe-fan being Washed-away

Tryptophan is a neutral amino acid. Defect in the neutral amino acid transporter causes decreased reabsorption of tryptophan in the kidneys, leading to excretion in the urine.

Leads to Pellagra

Pelican

Pellagra is the constellation of symptoms associated with severe niacin deficiency. It includes 3 D's of diarrhea, dermatitis, and dementia.

Diarrhea

Toilet

Diarrhea is a characteristic symptom of niacin deficiency.

Dermatitis

Dermatologist Examining Rash

Dermatitis is a characteristic symptom of niacin deficiency.

Dementia

Demented-D-man

Dementia, which is a serious loss of global cognitive ability, is a characteristic symptom of niacin deficiency. However, all kinds of neuropsychiatric symptoms are possible including anxiety, depression, delirium, sleep changes, and delusions.