



The Children's Hospital

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Inherited Metabolic Diseases Clinic

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Inherited Metabolic Disorders Visual Aids

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With Support From VITAFLO



Innovation in Nutrition

Introduction

Inherited Metabolic Disorders: Visual Aids

is offered as a supplement to your teaching aids collection. As physicians, genetic counselors, dietitians, nurses, and other providers in the field of biochemical genetics, we hope these visual aids complement your teaching with patients, families, students, and other clients.

The book is comprised of two sections: general metabolic concepts and specific biochemical pathways. The slides are not designed to stand alone, but rather depend on provider expertise for successful explanation. In some cases, overlays are utilized to allow a stepwise approach to a complex pathway.

With the advent of expanded newborn screening and ongoing research, our understanding of biochemical genetics continues to grow and change. This book reflects one approach at one point in time in a dynamic field. On many levels, it is intended as a starting point.

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Key



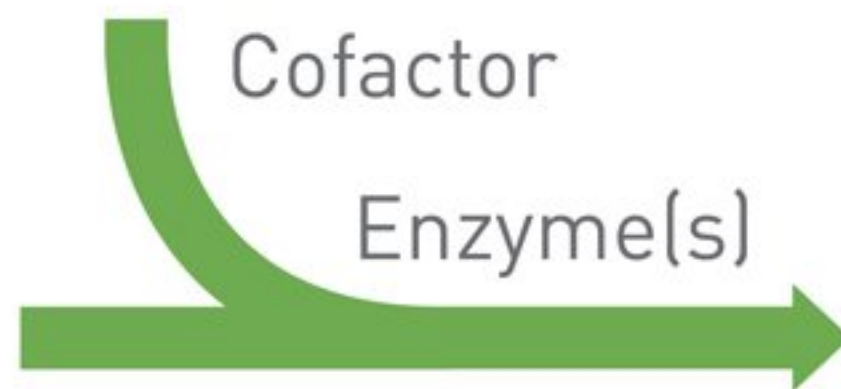
Fat from food
and/or the body



Carbohydrates from
food and/or the body



Protein from food
and/or the body



Normal pathway

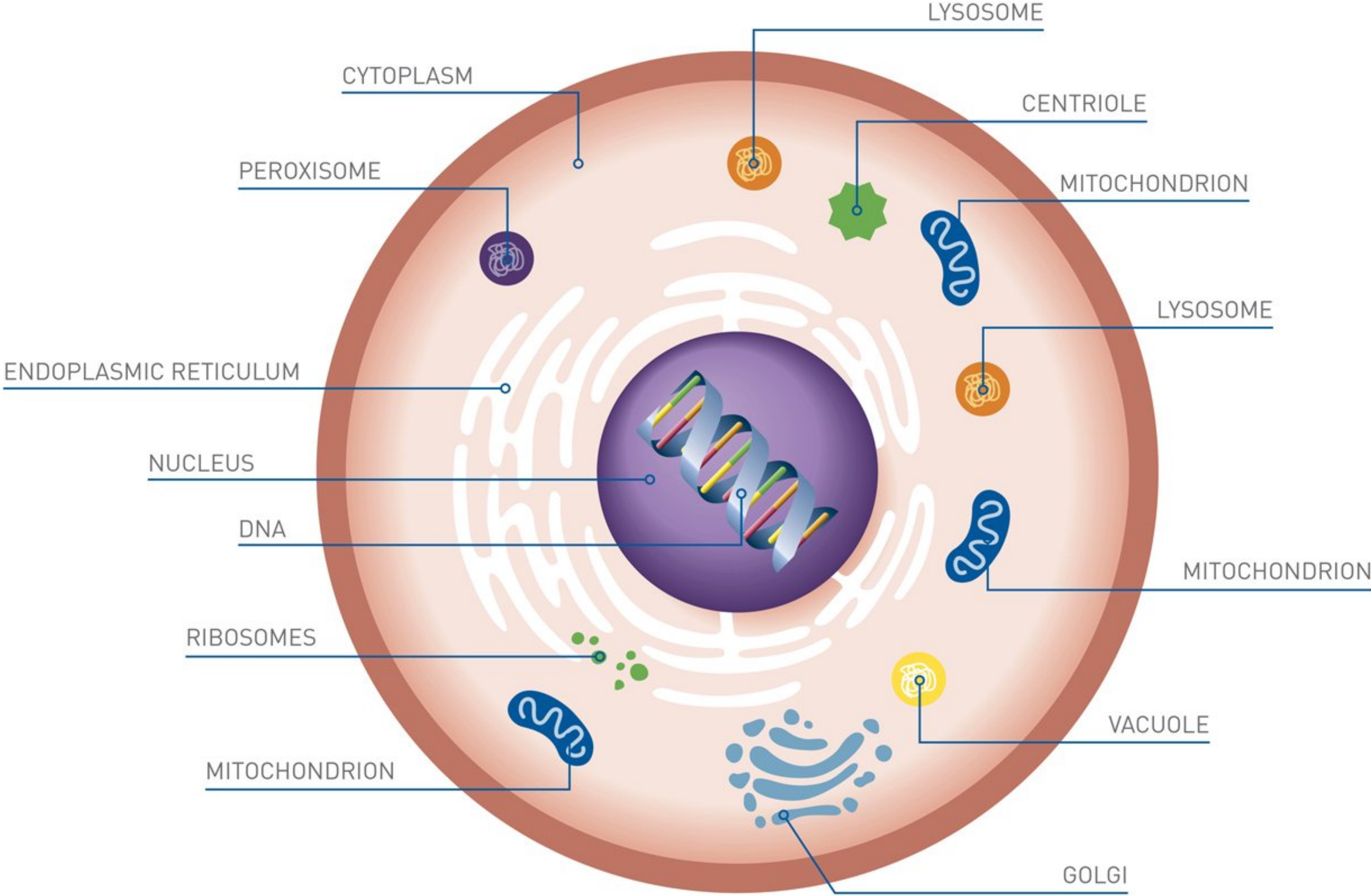


Metabolic block and
disease name

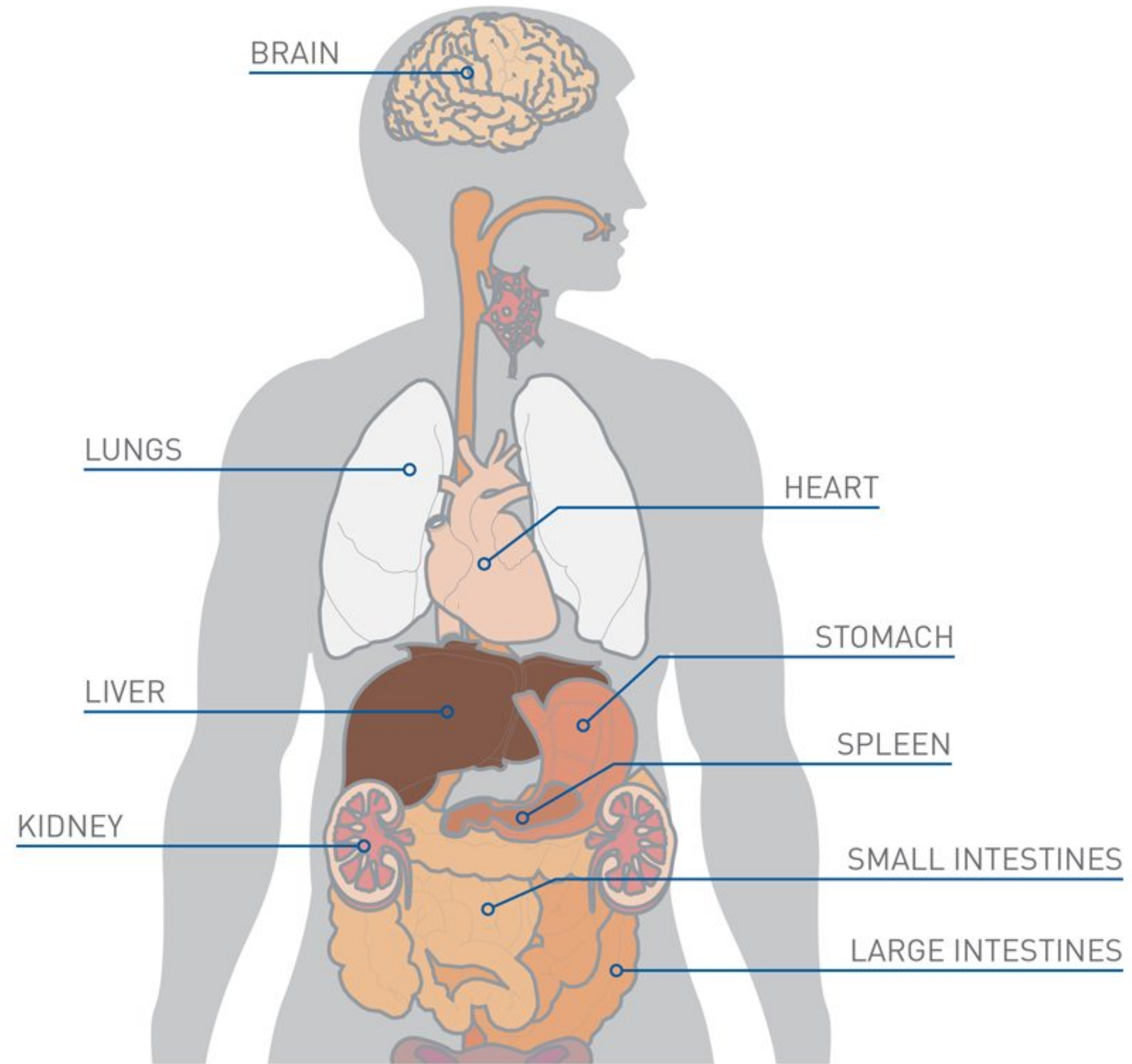


Abnormal
metabolite levels

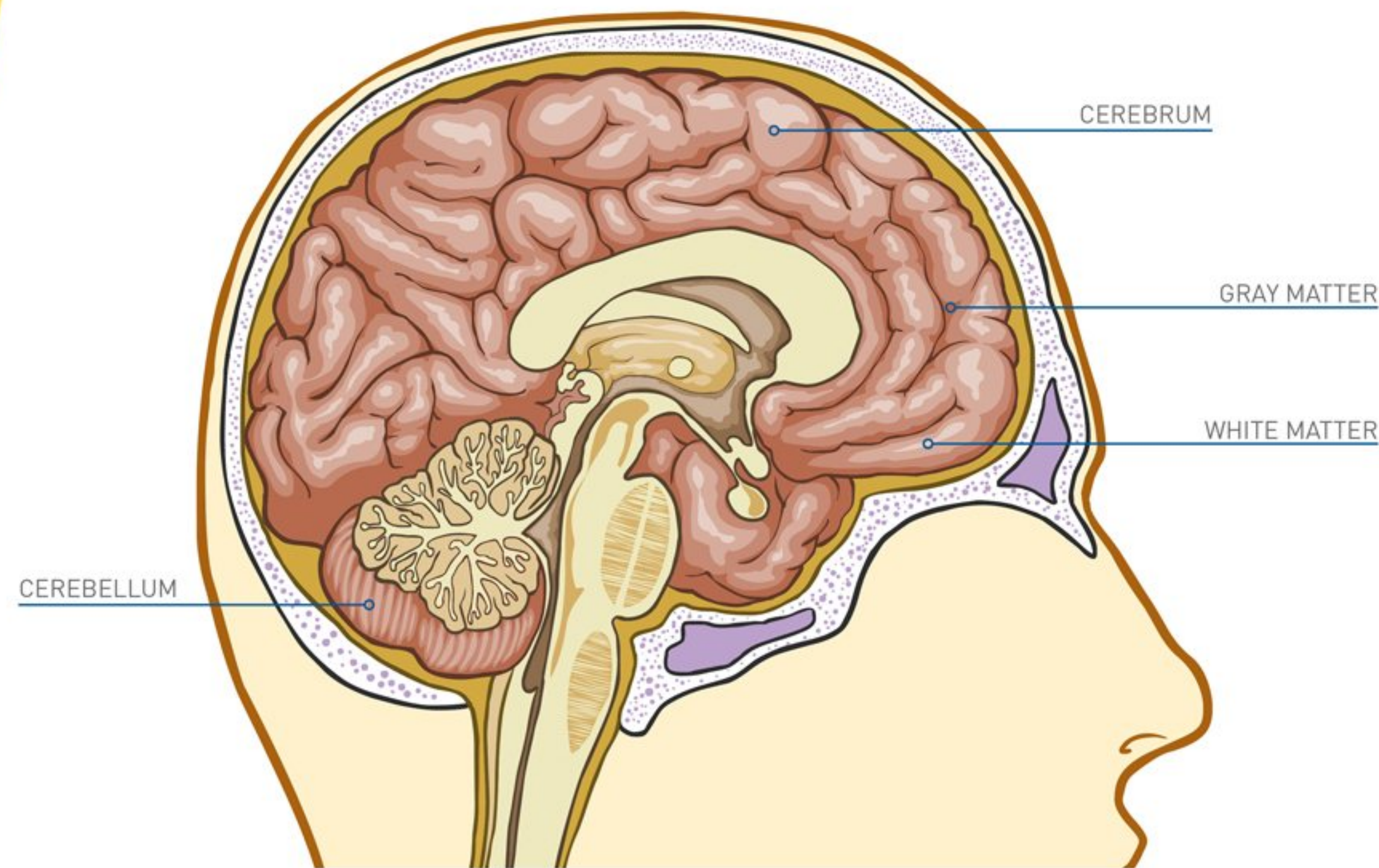
Parts of a Cell



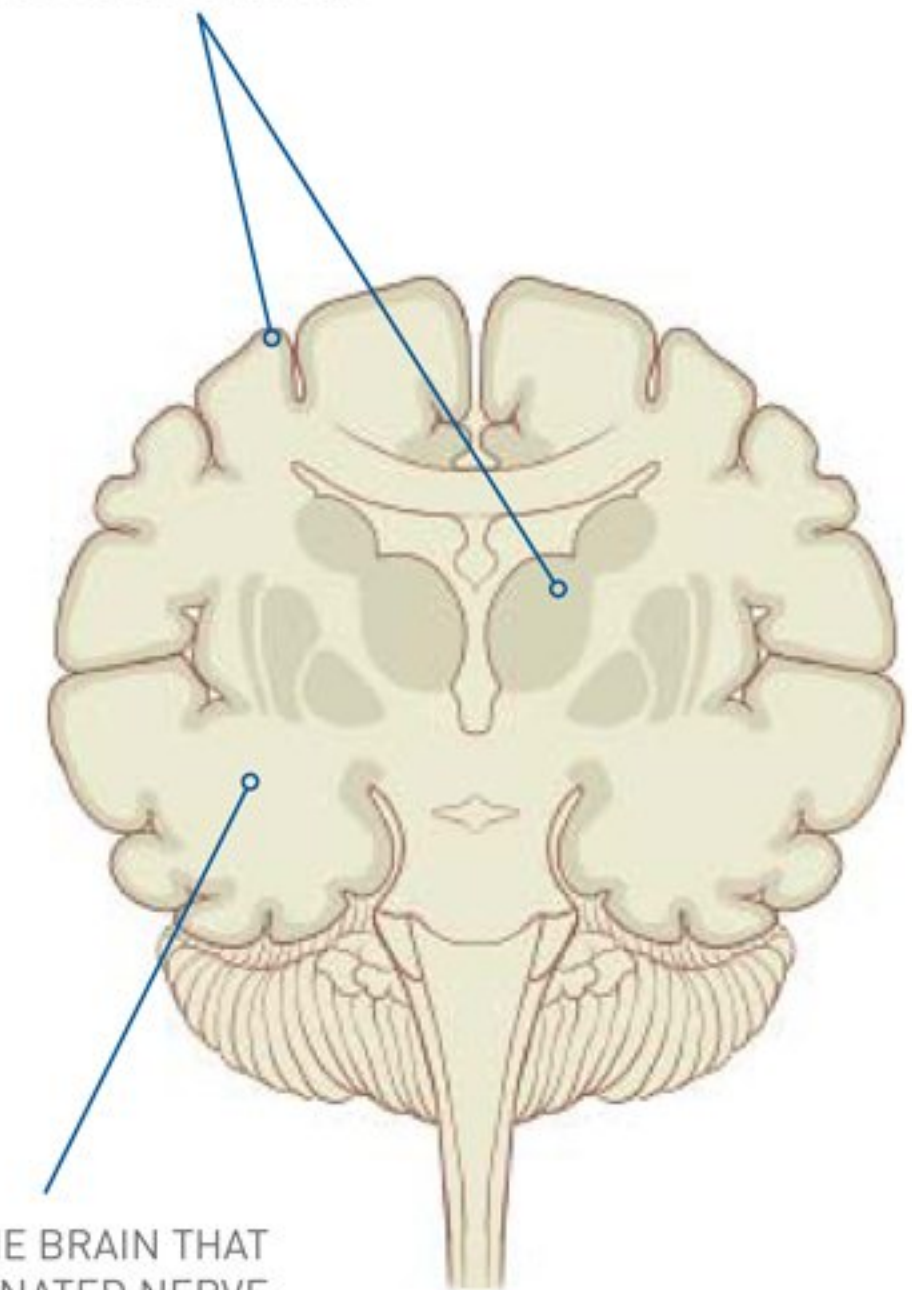
Human Anatomy



Brain Anatomy

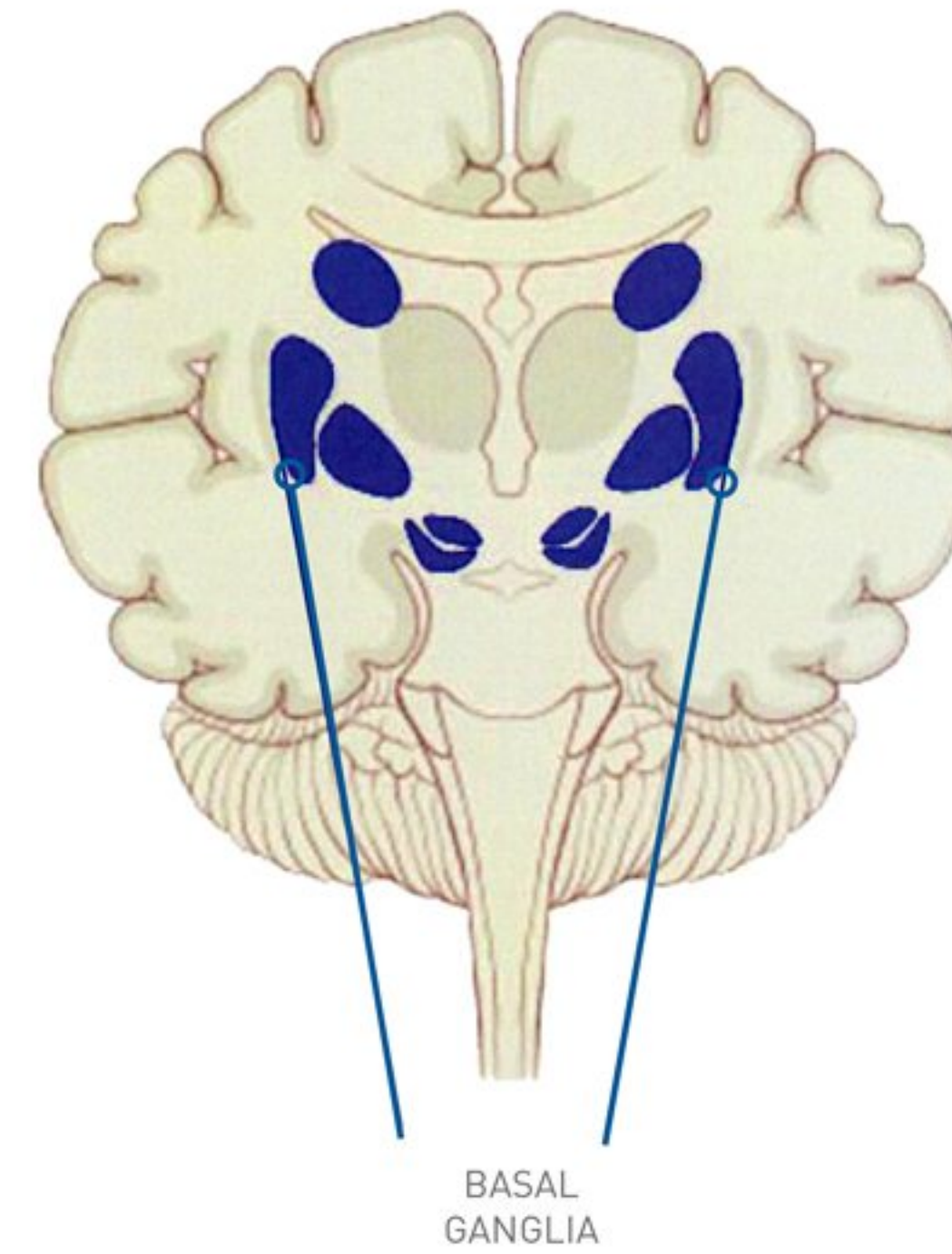
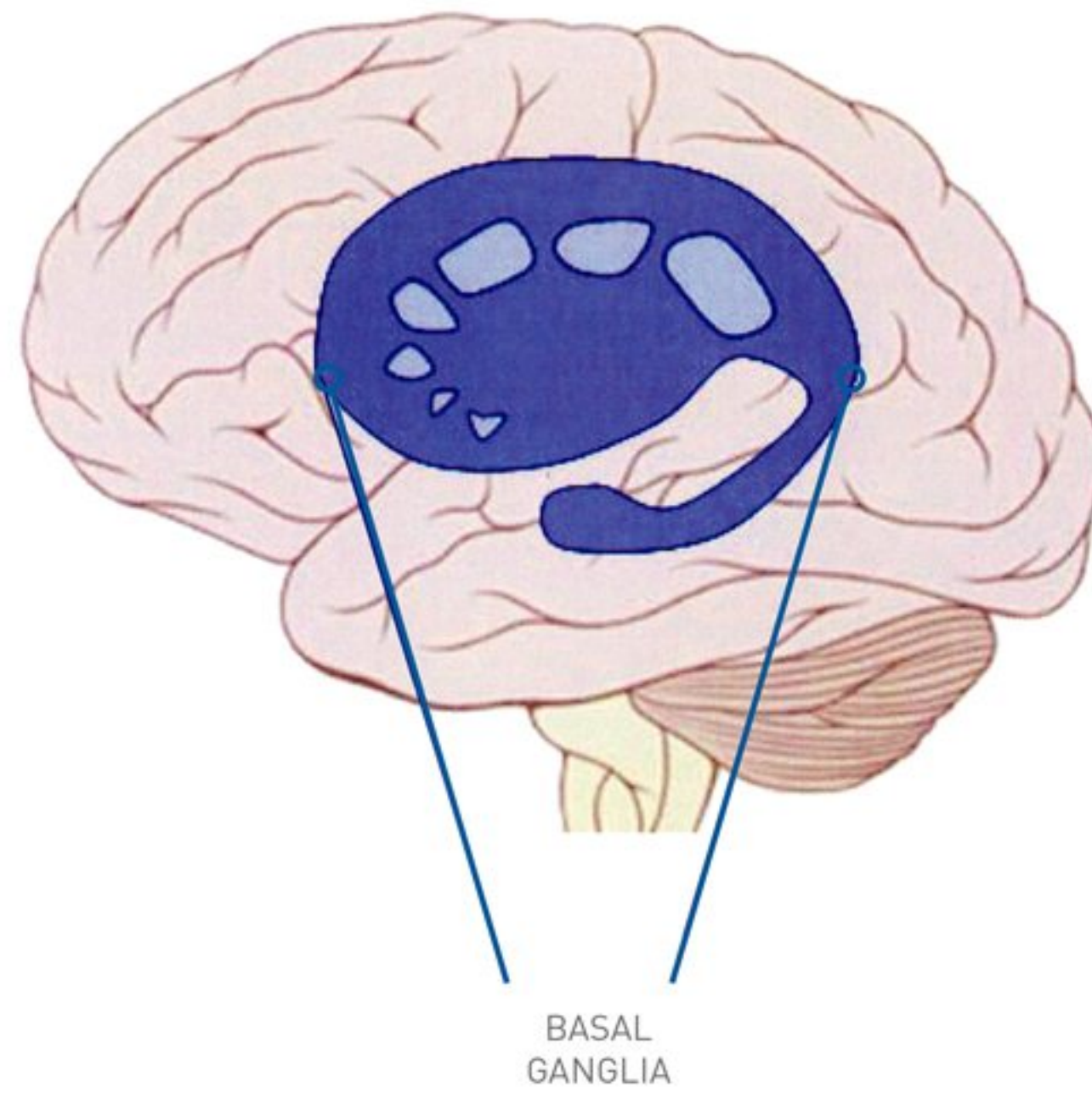


GRAY MATTER: PART OF THE BRAIN THAT IS COMPOSED OF NERVE CELL BODIES (NEURONS).

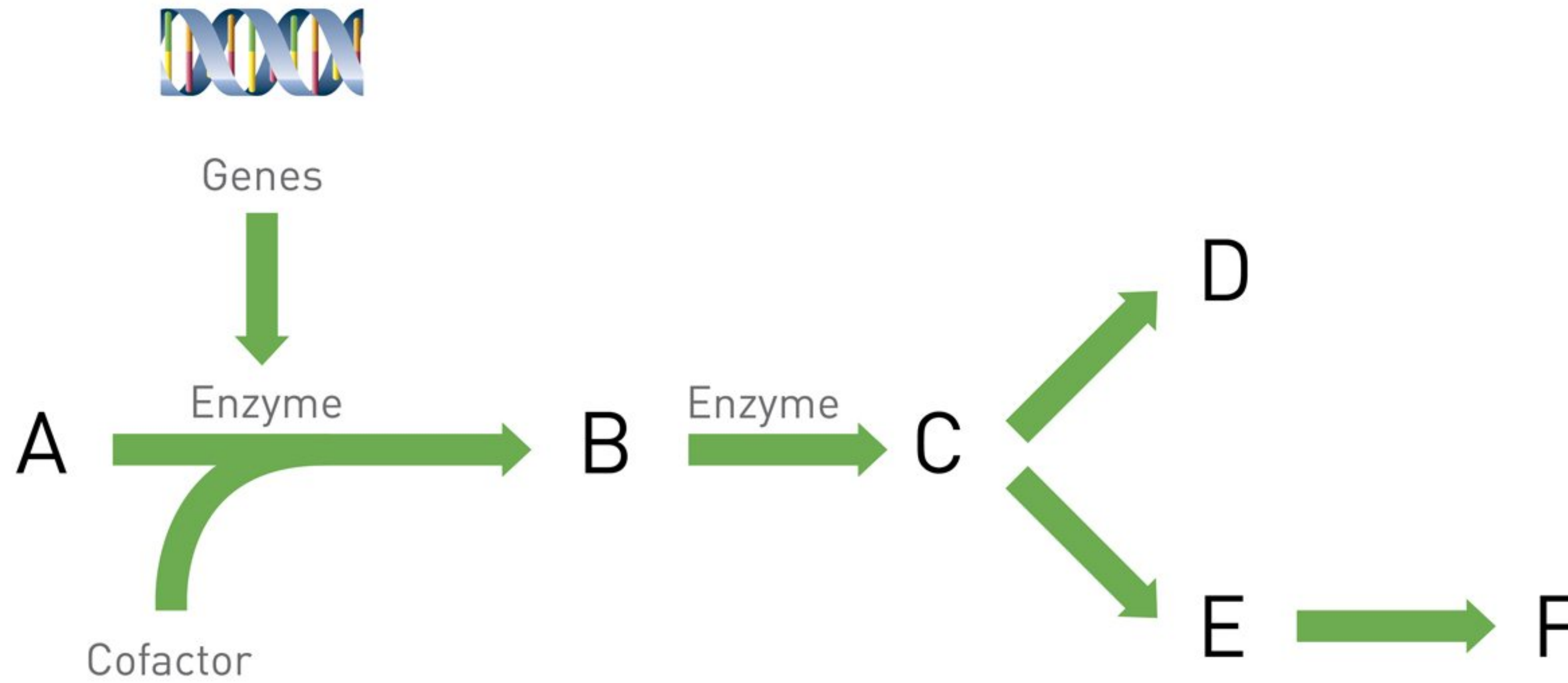


WHITE MATTER: PART OF THE BRAIN THAT IS COMPOSED OF MYELINATED NERVE CELL PROCESSES (AXONS).

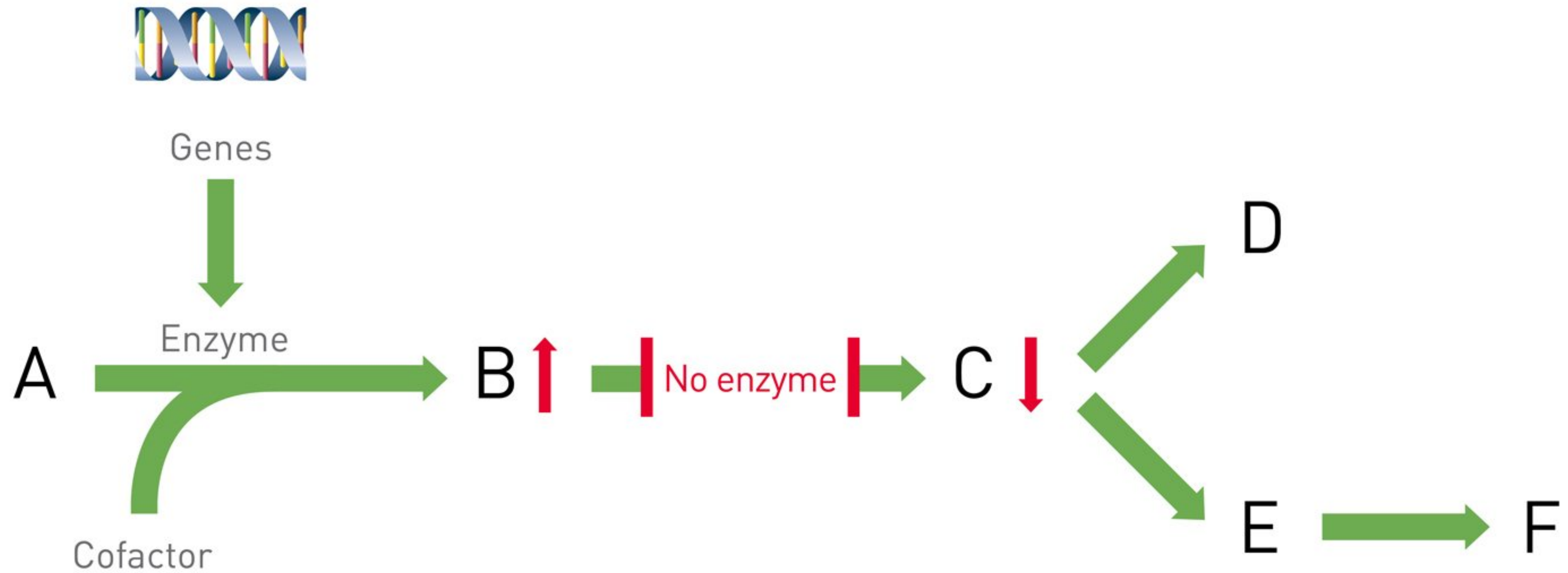
Basal Ganglia

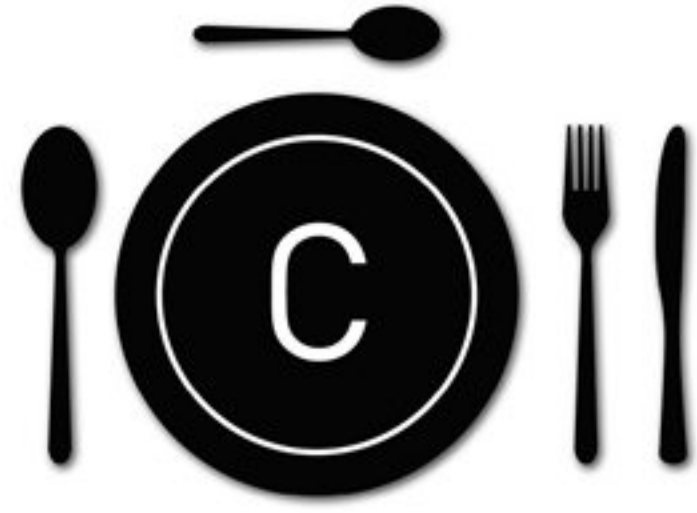


Enzyme



Enzyme Disorders





Carbohydrates

Monosaccharides (simple sugars)



Glucose



Galactose



Fructose

Disaccharides



Glucose

+



Fructose



Sucrose

Table Sugar



Glucose

+



Galactose



Lactose

Milk Sugar



Glucose

+

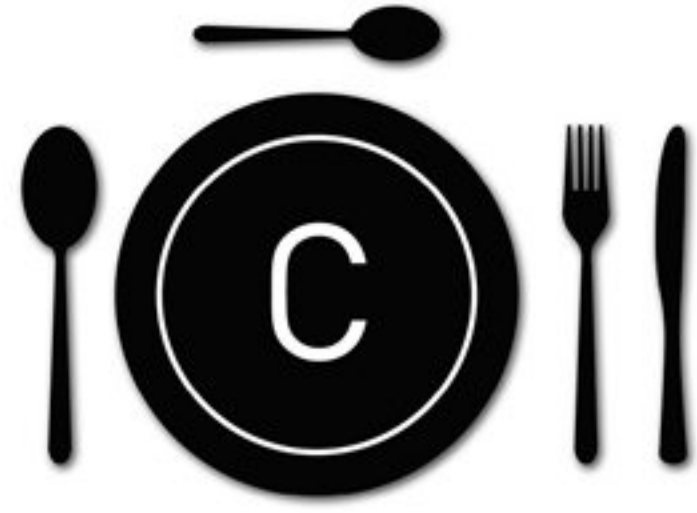


Glucose



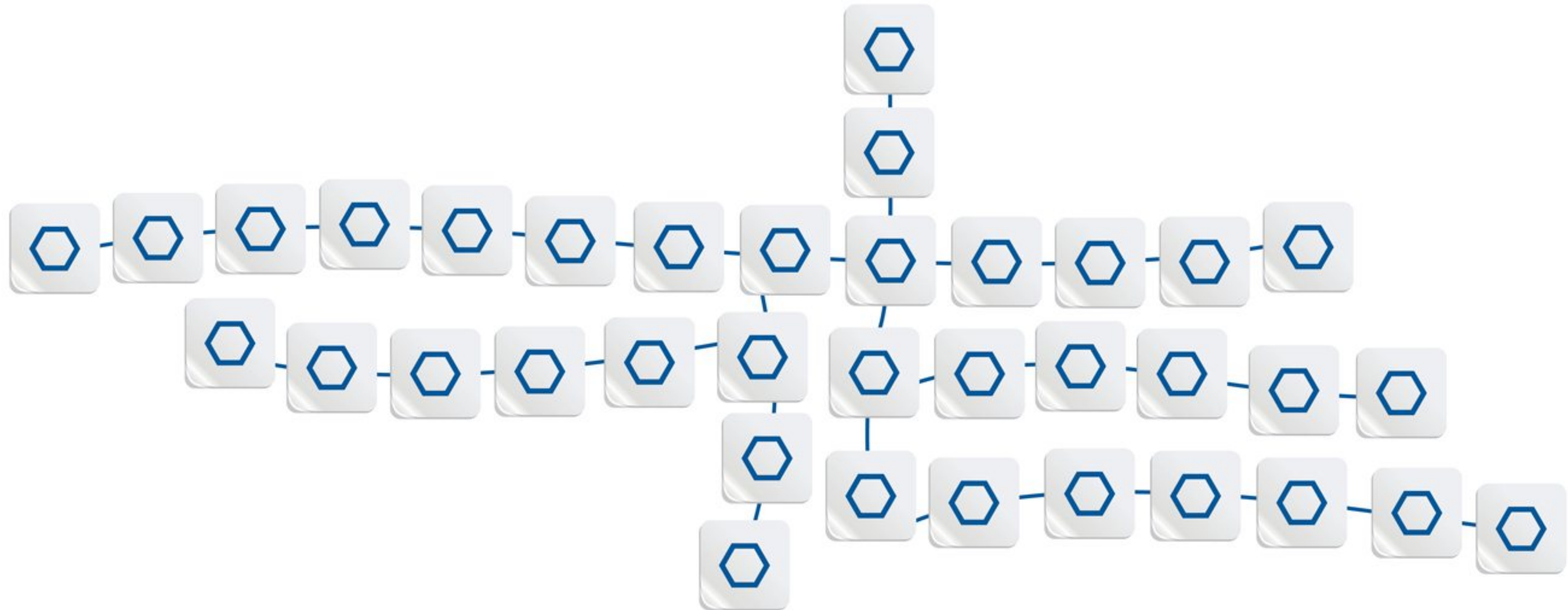
Maltose

Malt Sugar



Carbohydrates

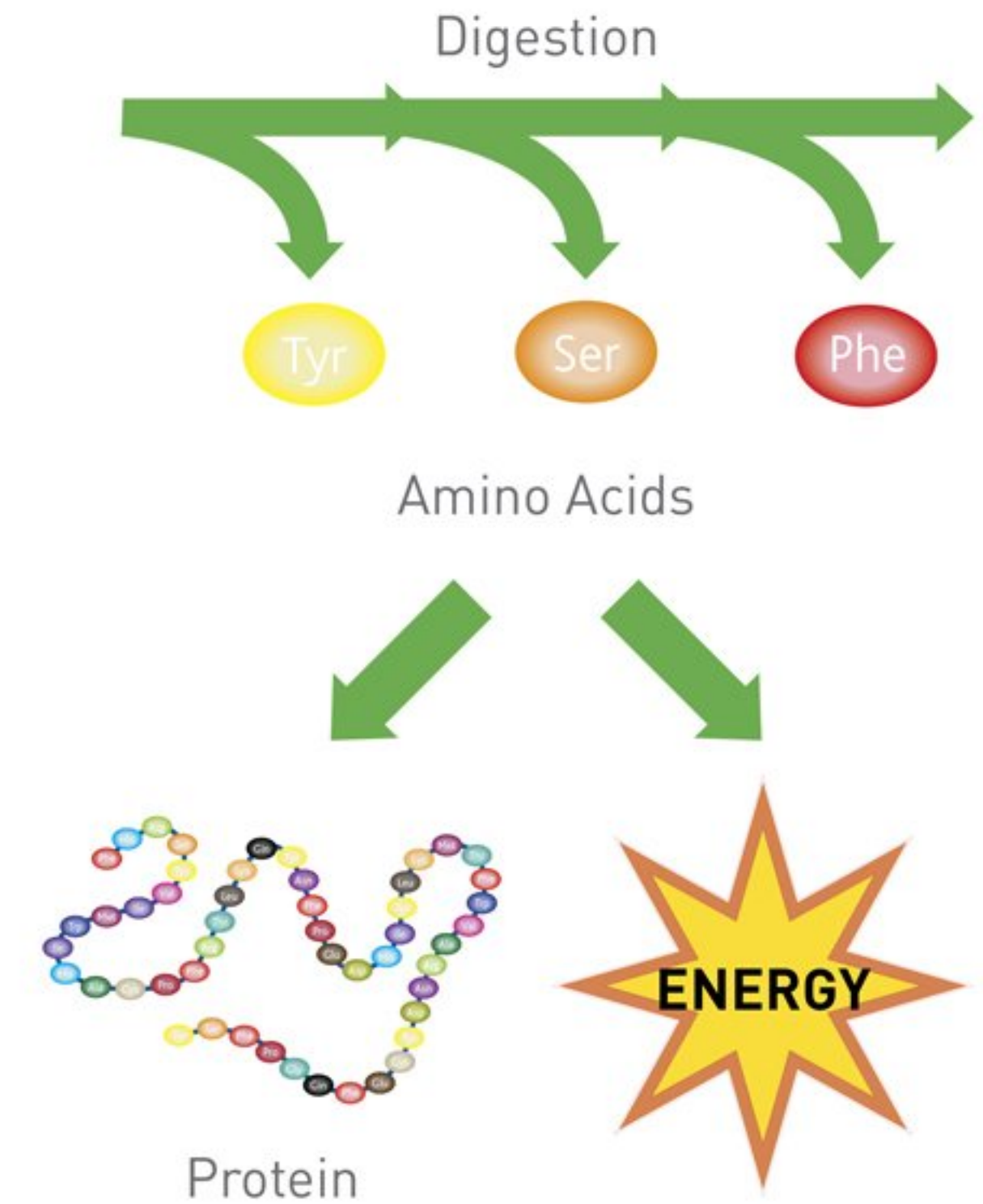
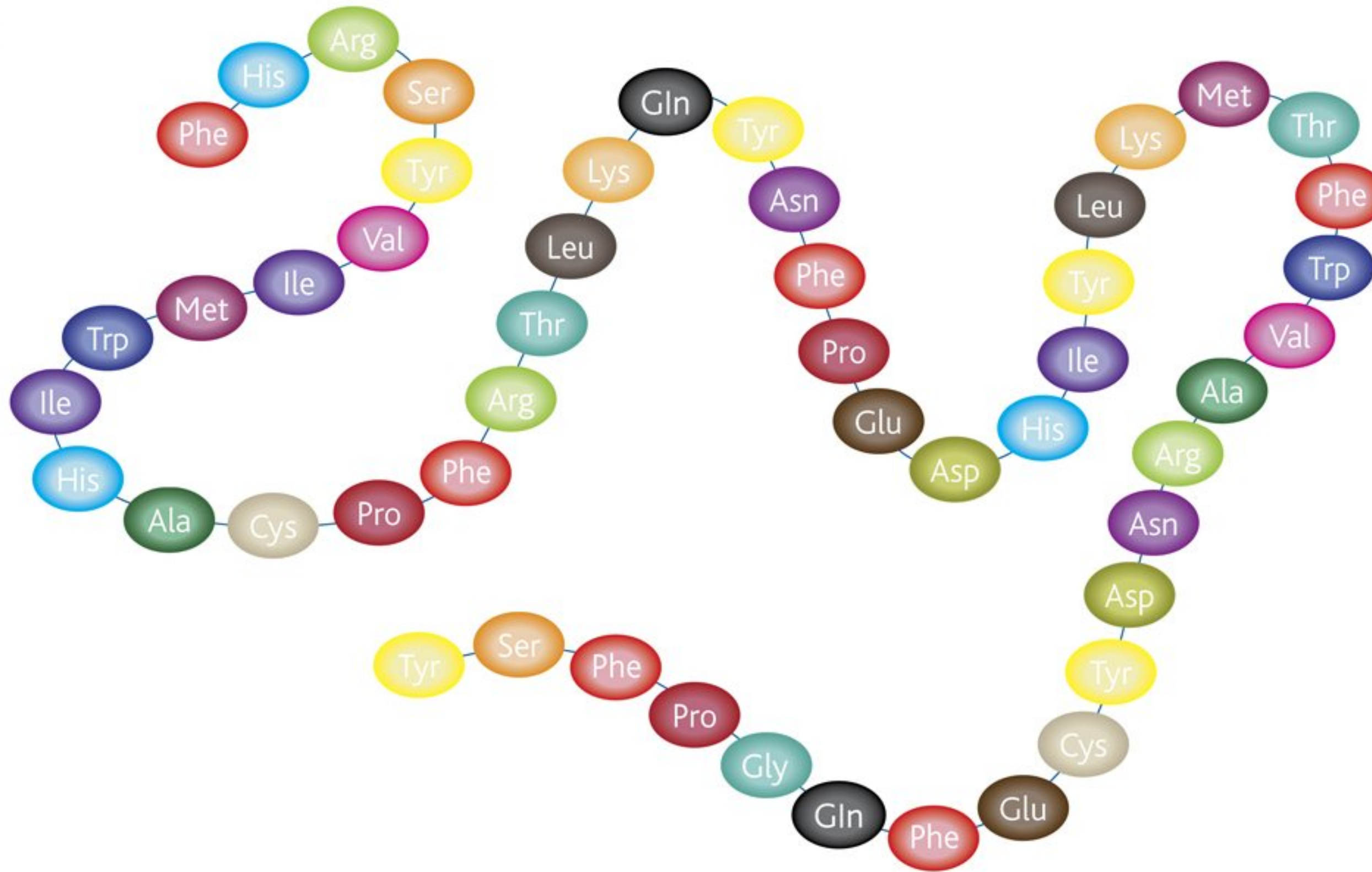
Polysaccharides (complex carbohydrates)



Carbohydrates are many molecules of simple sugars joined together (such as glycogen and starch) that the body uses for energy. They can be eaten as food (potatoes, corn) or can be produced by the liver.



Protein



Protein is found in foods such as cheese, eggs, fish, and meat. Our muscles also contain protein.



Amino Acids

20 basic building blocks of protein

Essential Amino Acids

(Must get from our food)

His	Histidine
Ile	Isoleucine
Leu	Leucine
Lys	Lysine
Met	Methionine
Phe	Phenylalanine
Thr	Threonine
Trp	Tryptophan
Val	Valine

Non-Essential Amino Acids

(Made by our bodies)

Ala	Alanine	Ser	Serine
Arg	Arginine	Tyr	Tyrosine
Asn	Asparagine		
Asp	Aspartate		
Cys	Cysteine		
Glu	Glutamate		
Gln	Glutamine		
Gly	Glycine		
Pro	Proline		

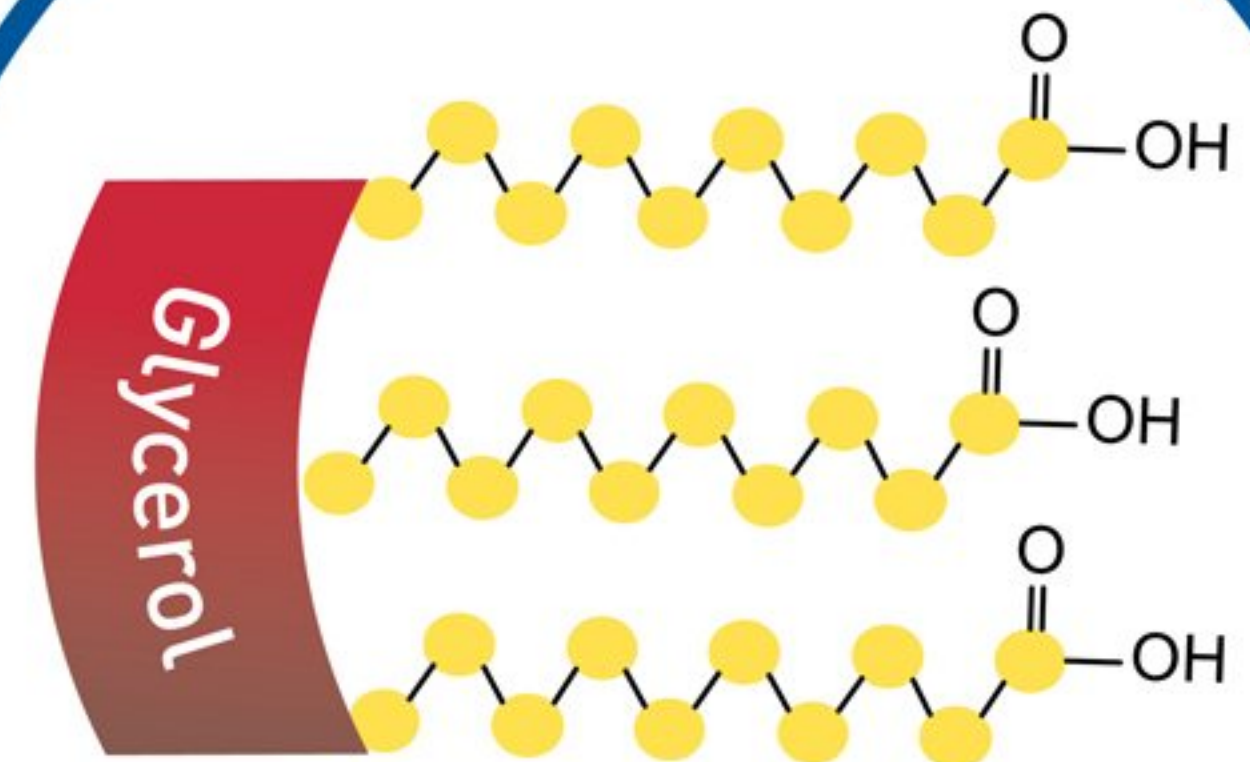
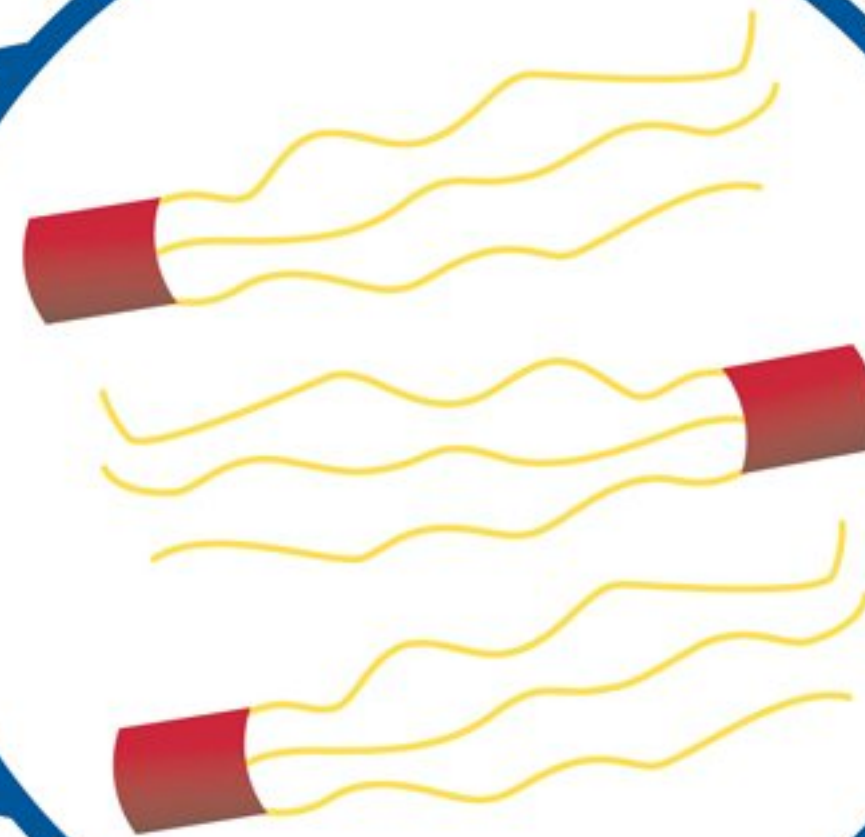


Fats

A triglyceride is made up of glycerol and three fatty acids.

...contains molecules called triglycerides.

Fat in food and in our bodies...



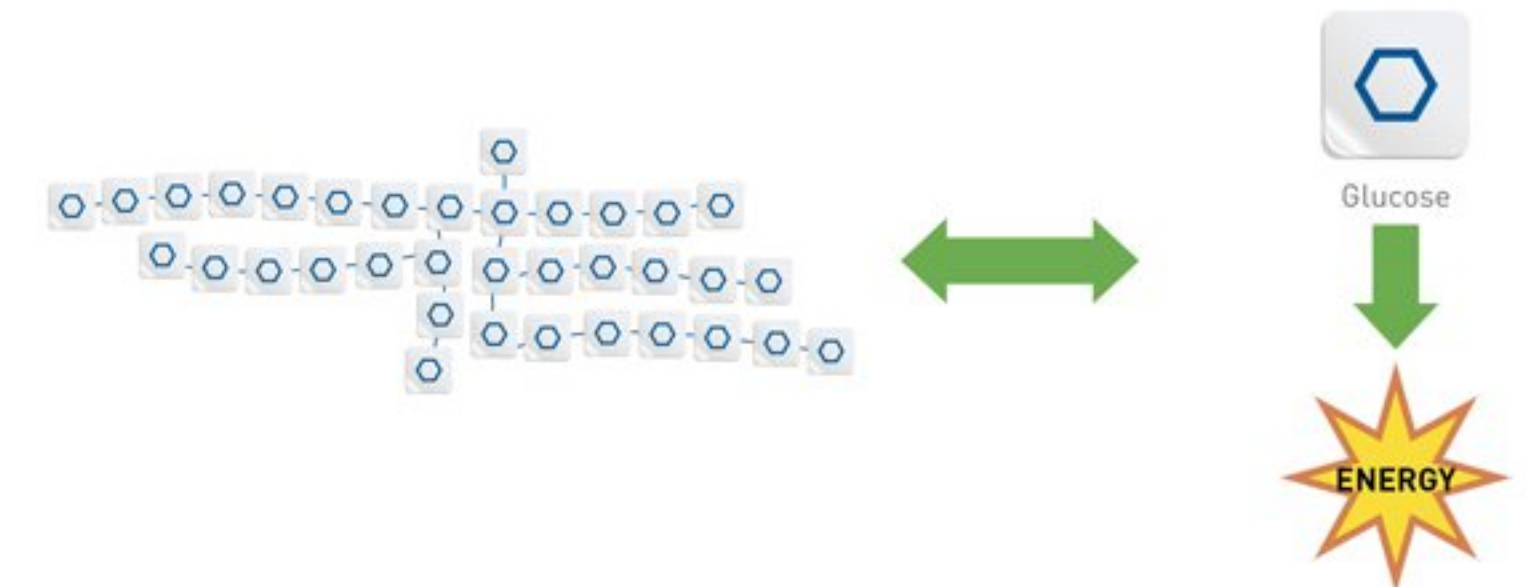
Saturated Fats: solid at room temperature

Unsaturated Fats: liquid at room temperature

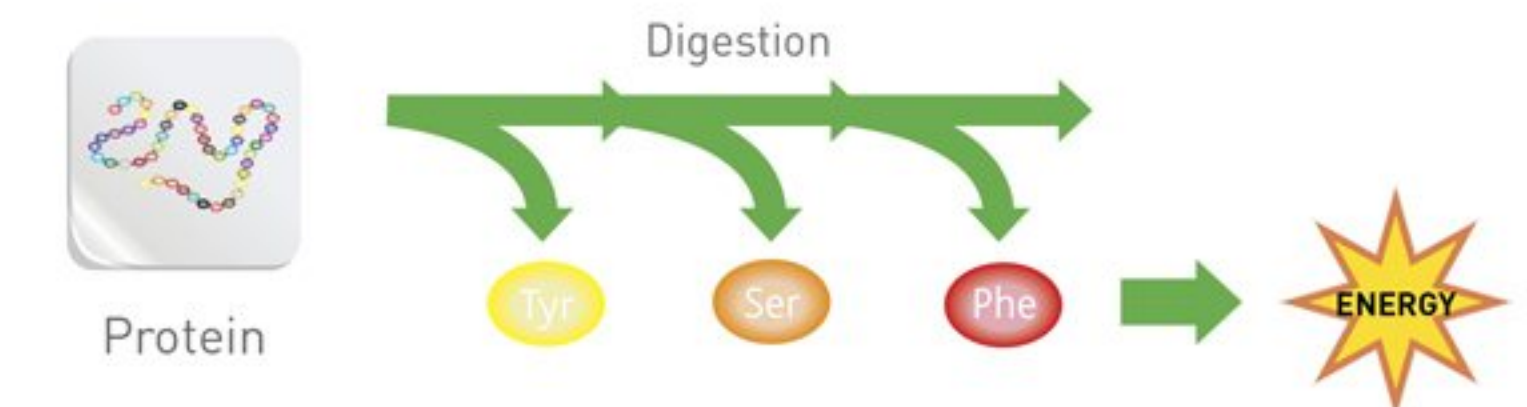
Digestion/Fasting - Up to 36 hours



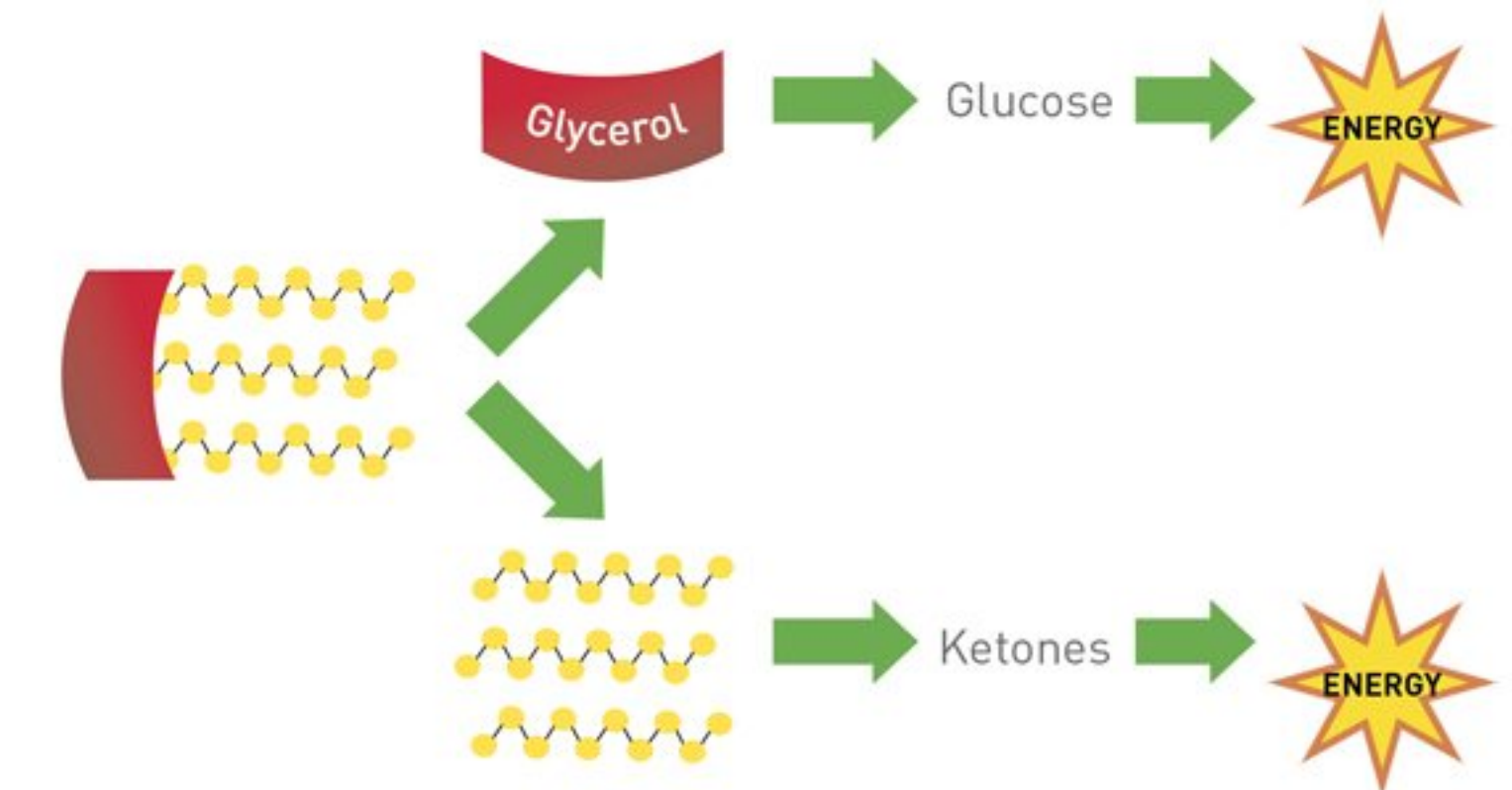
- I. Glucose from food is used for energy and is the preferred energy for the brain. Extra glucose is stored as glycogen. When glucose is gone, glycogen is broken down into glucose for energy.



- II. Protein from muscle is broken down into amino acids. Amino acids go to the liver and are broken down for energy through gluconeogenesis or the citric acid cycle.

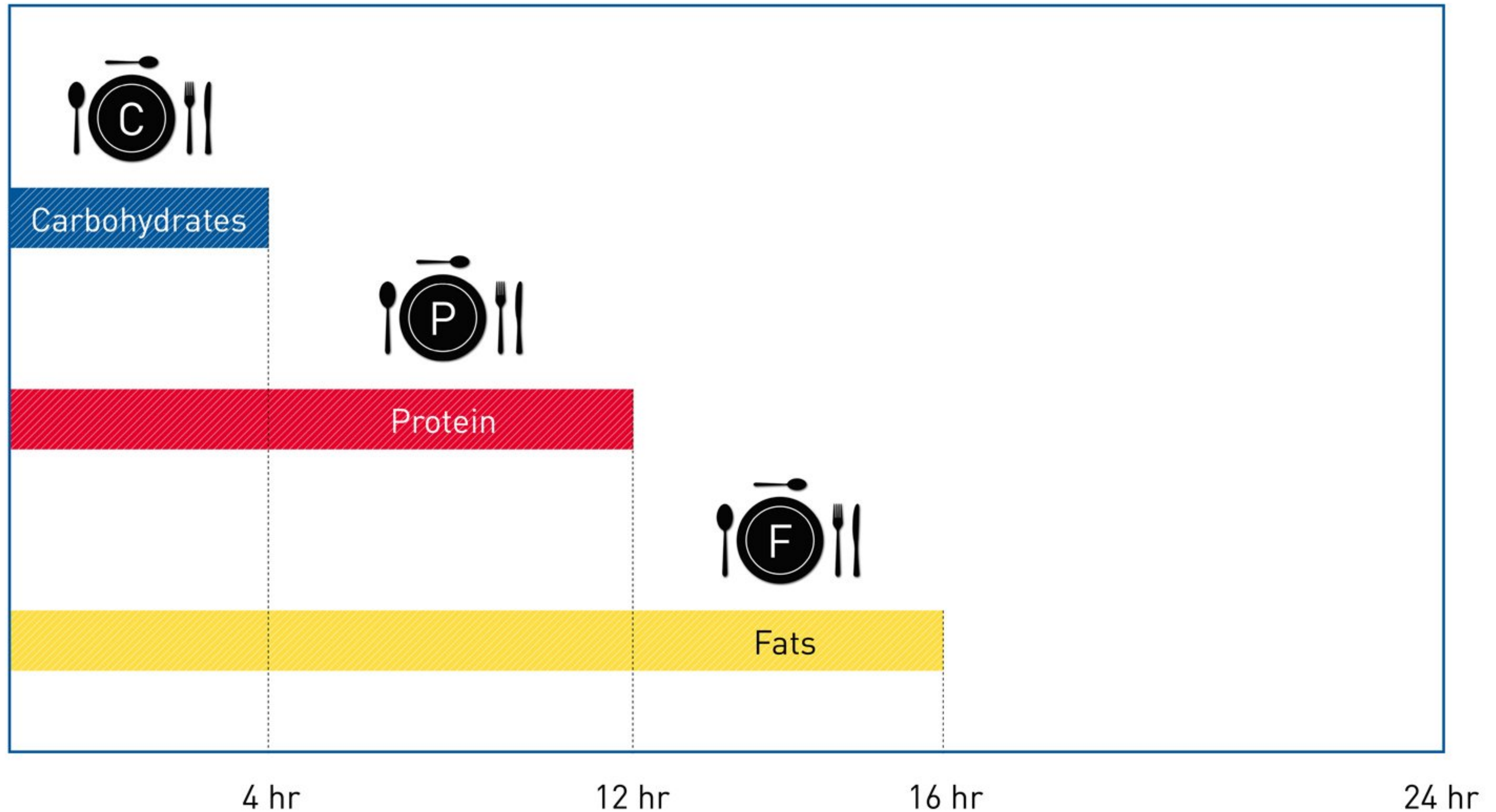


- III. Fat tissue breaks down into triglycerides. Glycerol is broken down into glucose. Fatty acids go to the liver and are broken down into energy for the body and energy for the brain in the form of ketones.

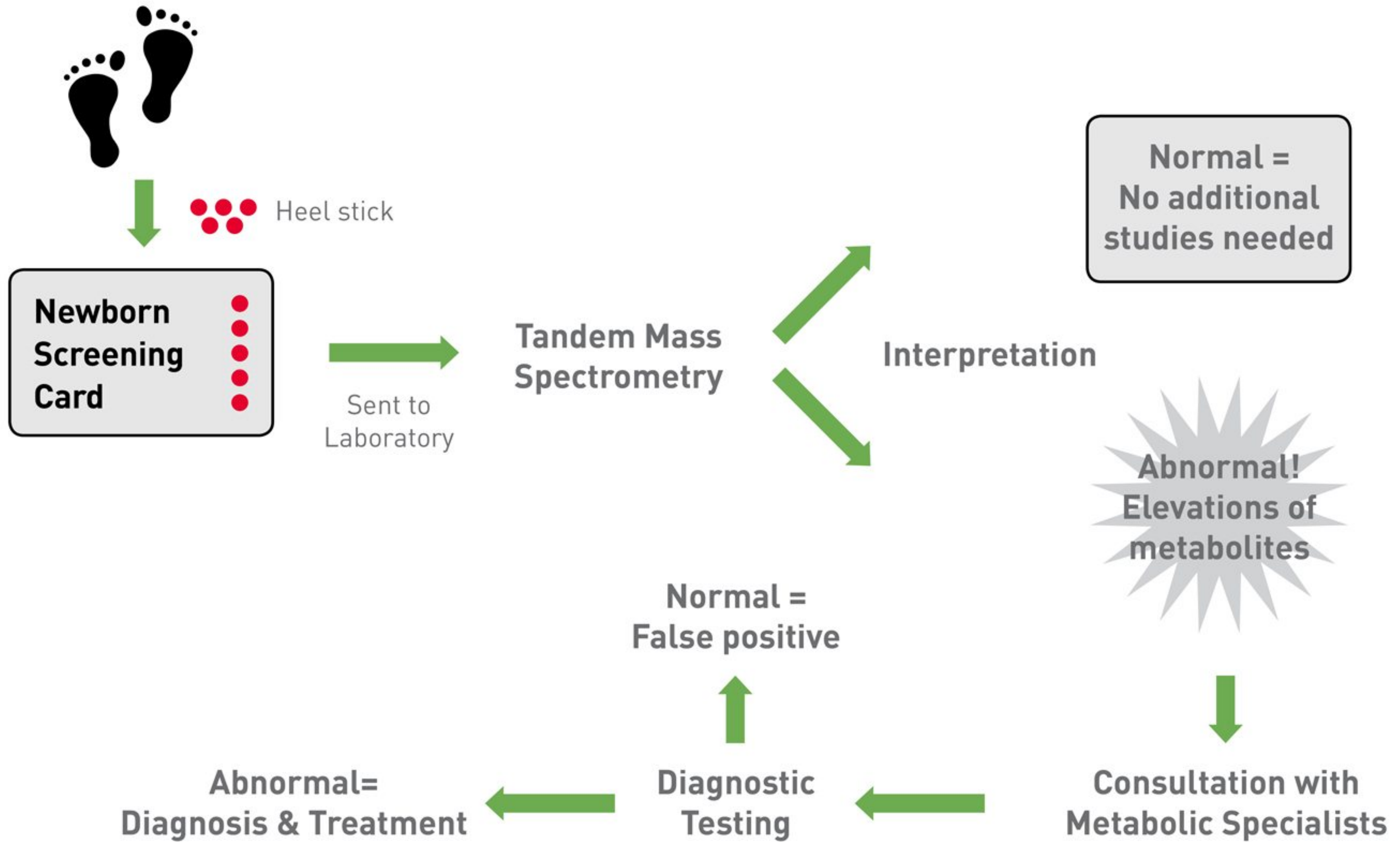


Digestion/Fasting

Different fuels take different times to be digested by the body.

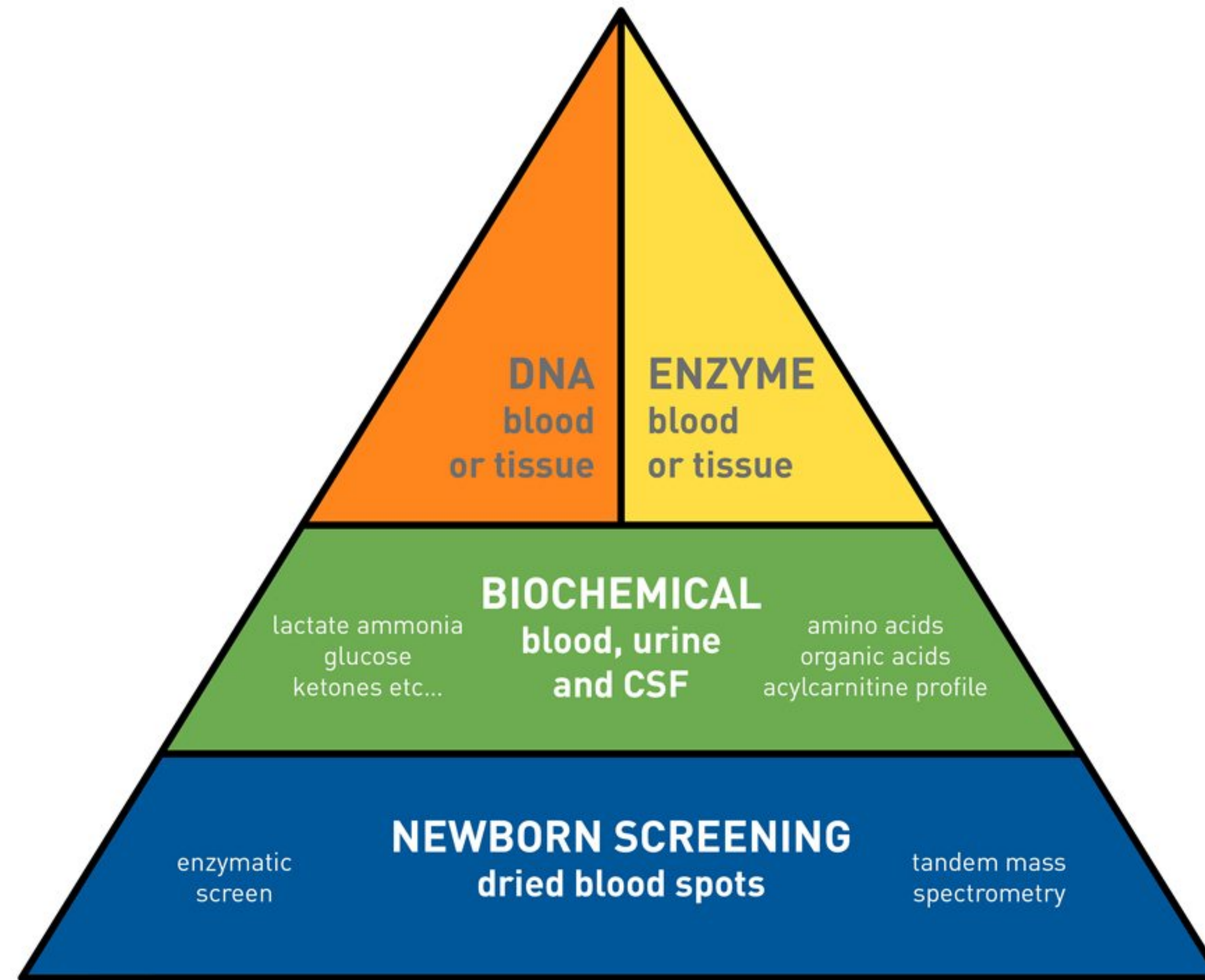


Newborn Screening



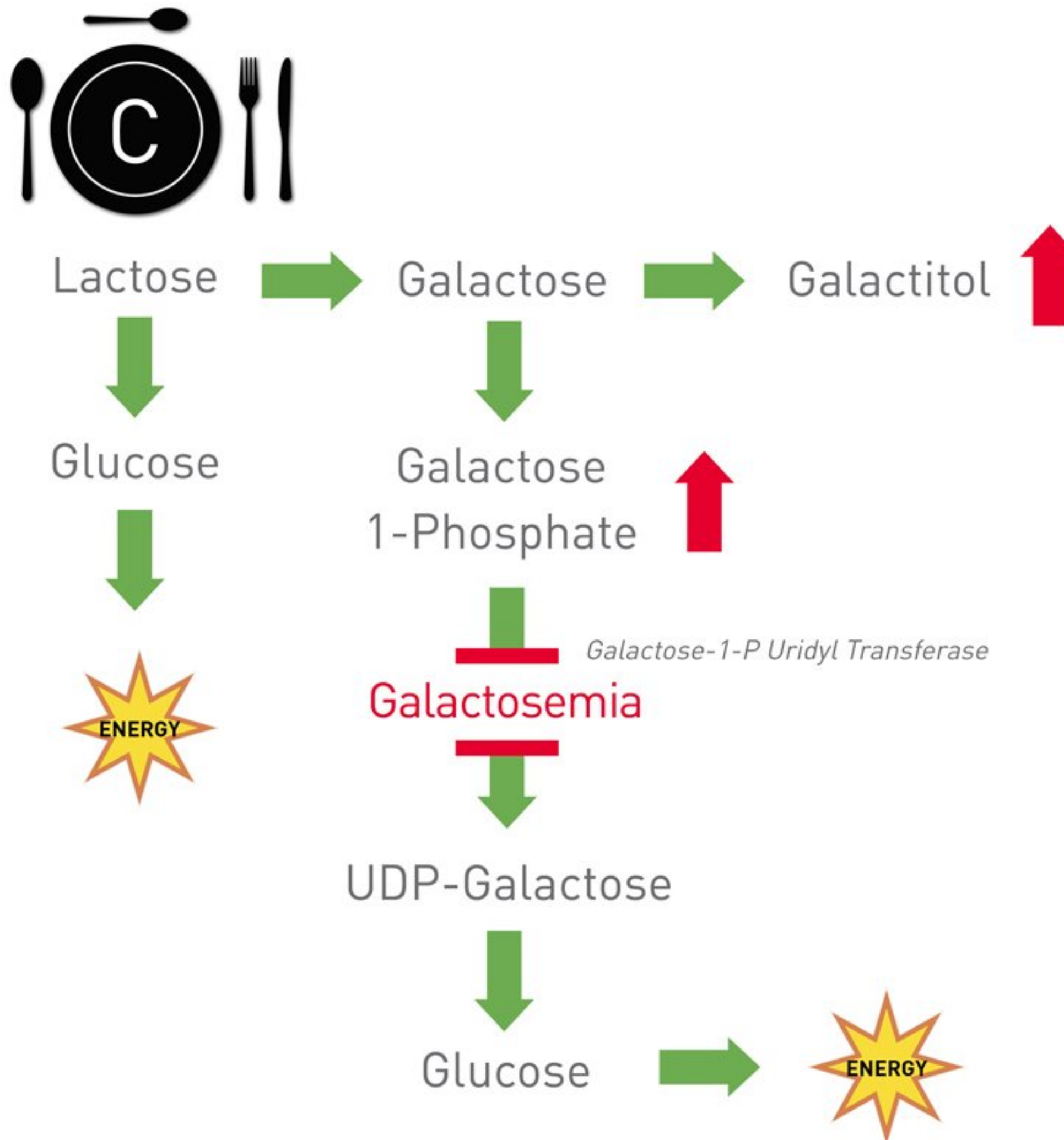
Diagnostic Process

Diagnosis and Treatment



Galactosemia

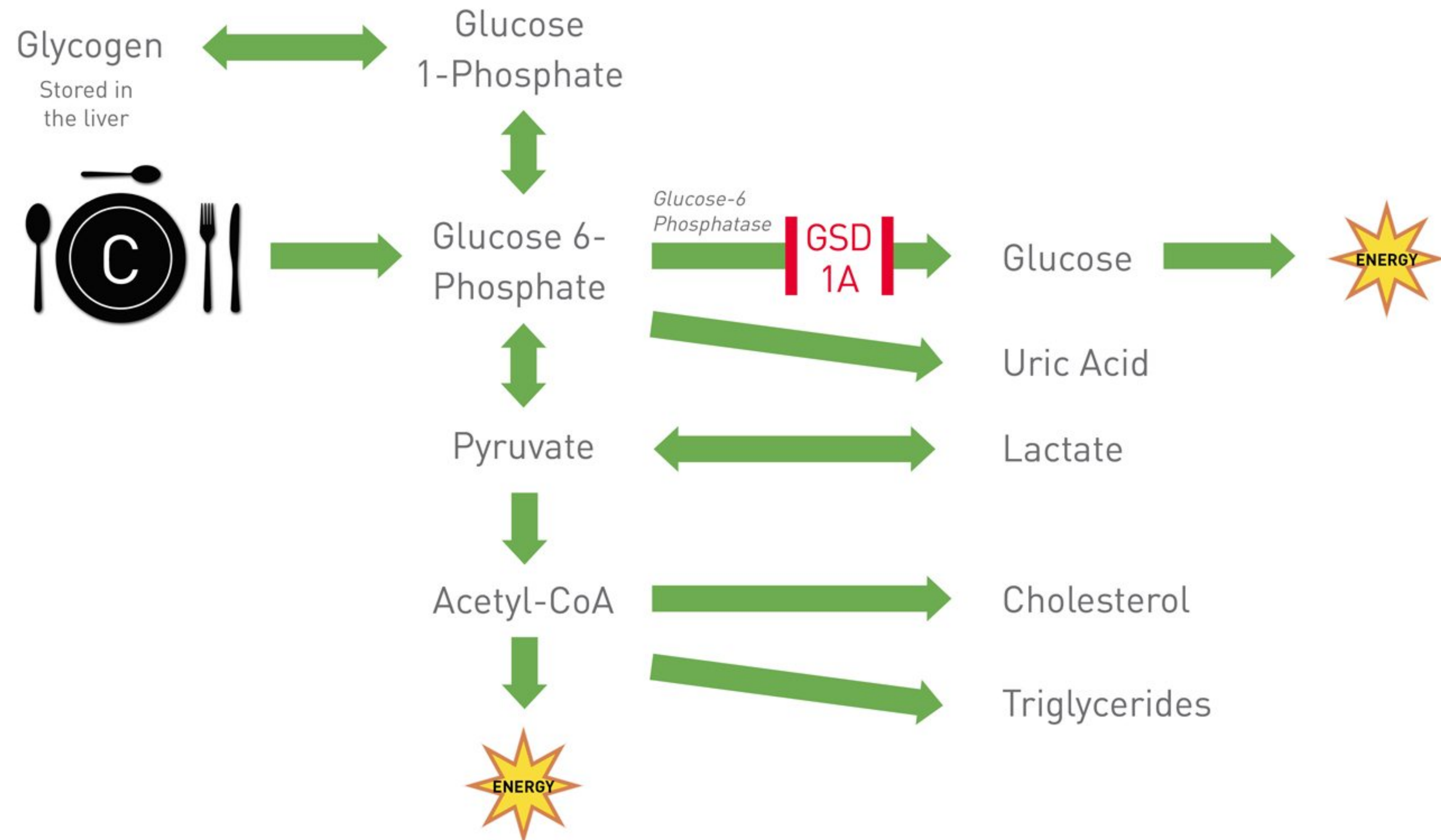
*Occurring mainly
in the liver...*



Glycogen Storage Disease

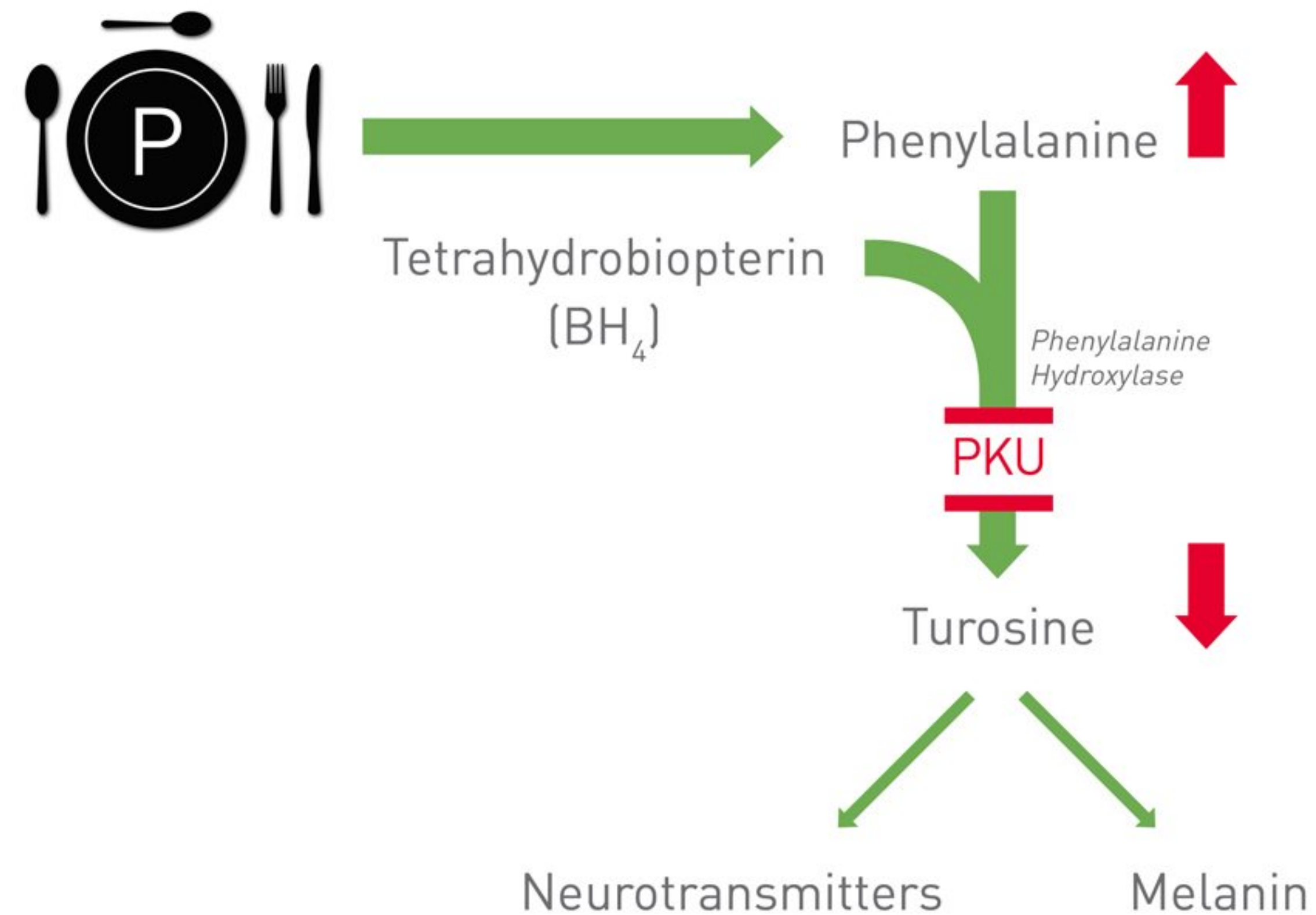
Occurring mainly
in the liver...

Type 1A (GSD 1A)



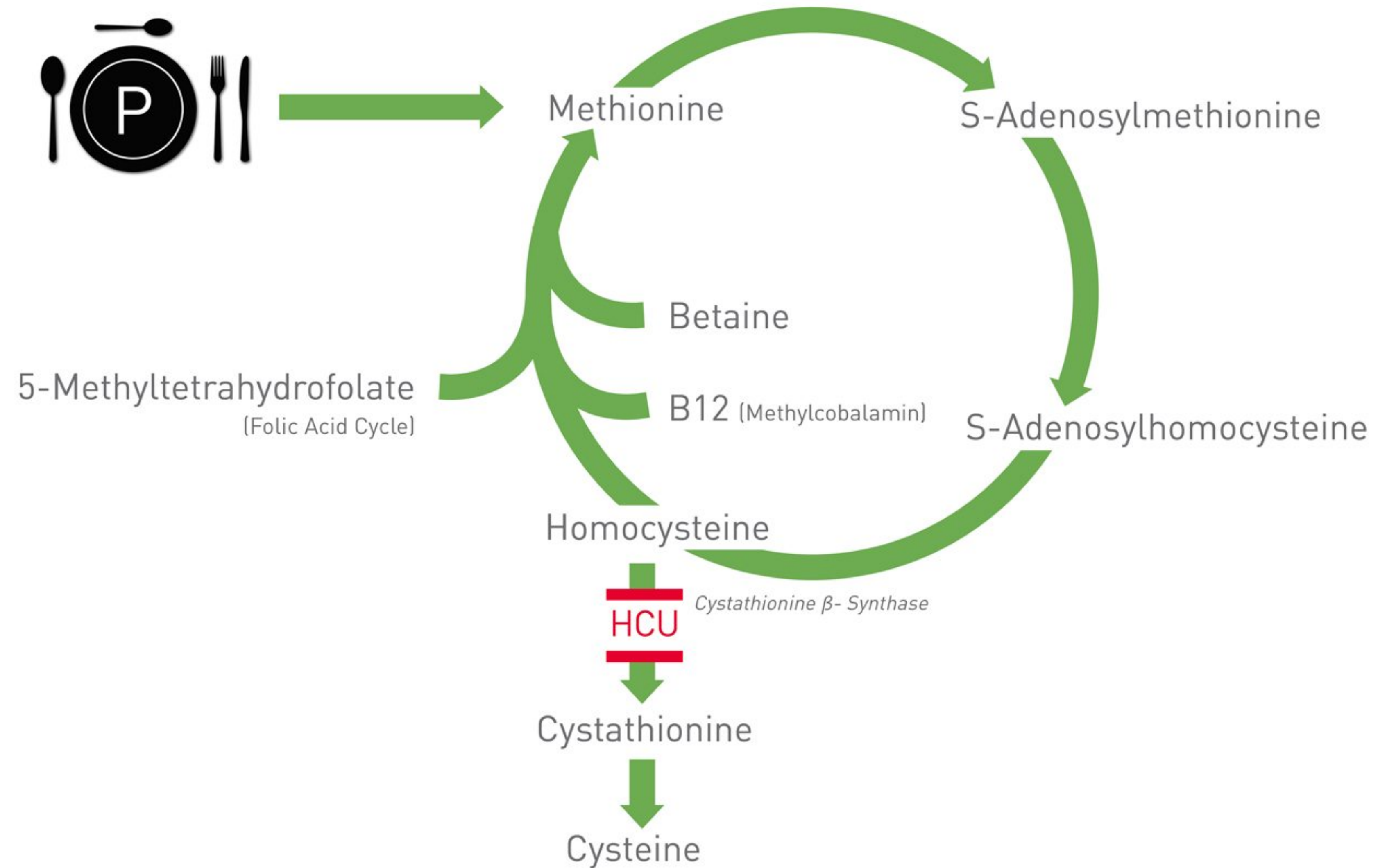
Phenylketonuria (PKU)

*Occurring mainly
in the liver...*



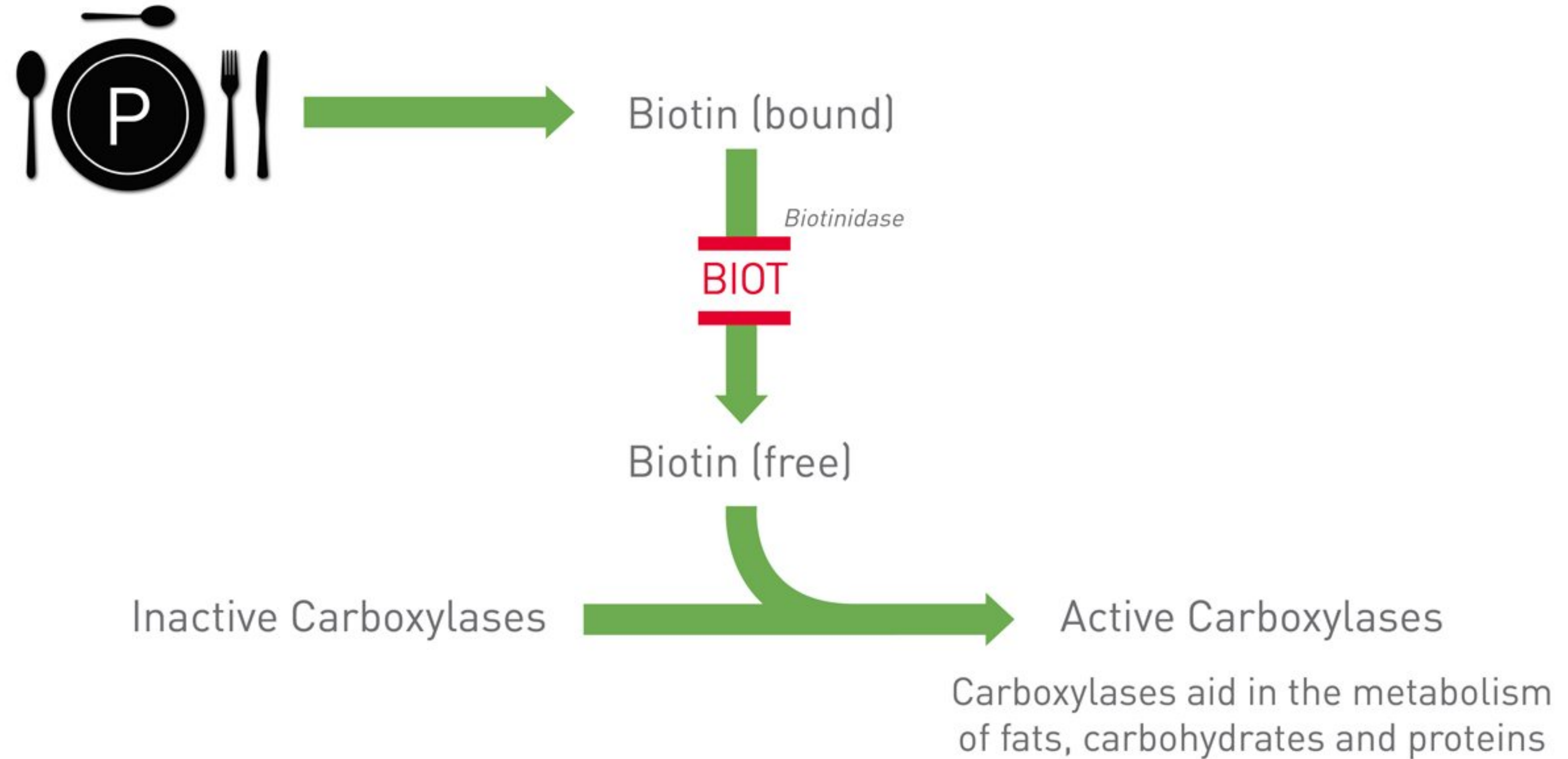
Homocystinuria (HCU)

*Occurring mainly
in the liver...*



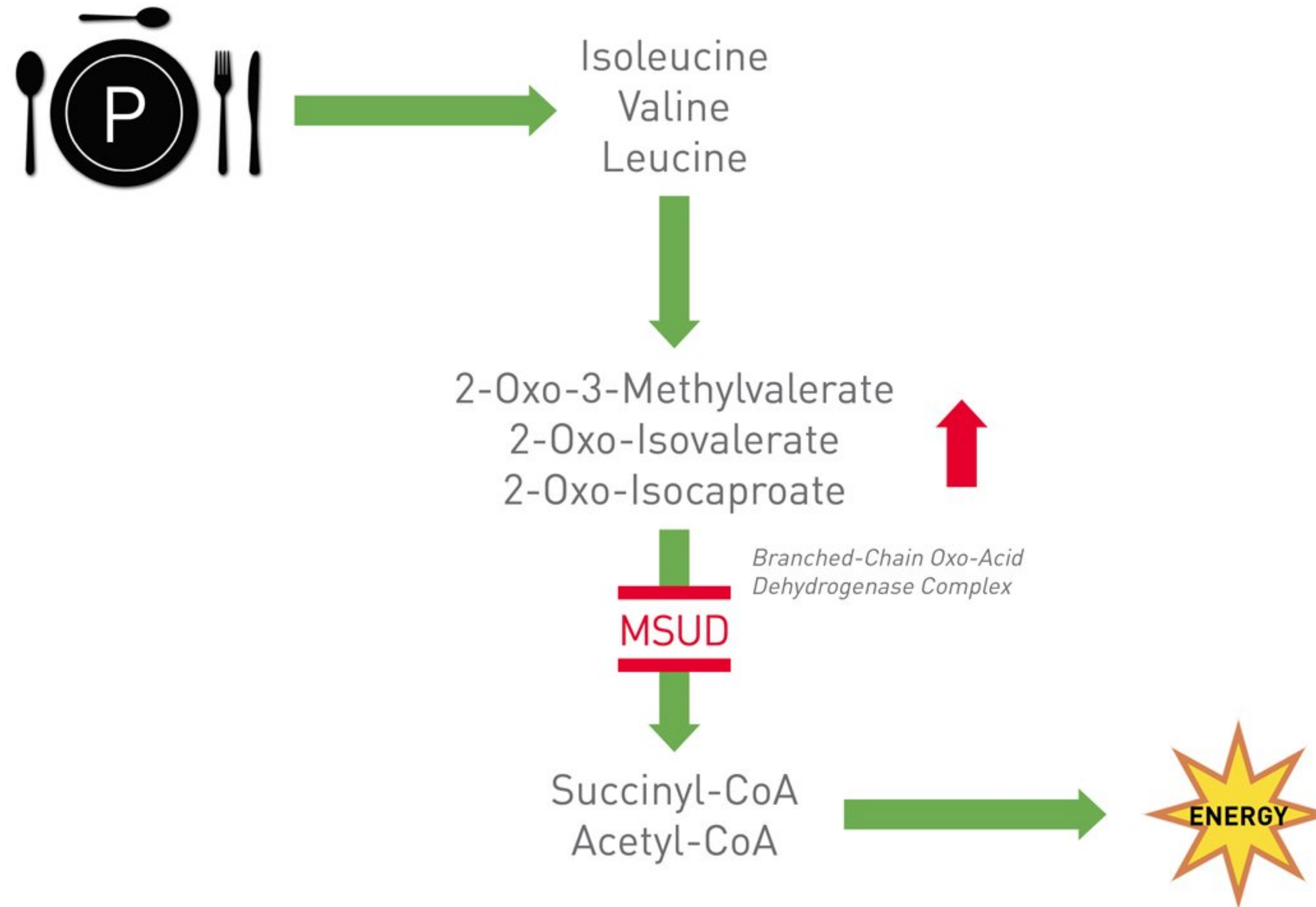
Biotinidase Deficiency (BIOT)

Occurring across tissues including liver and brain...



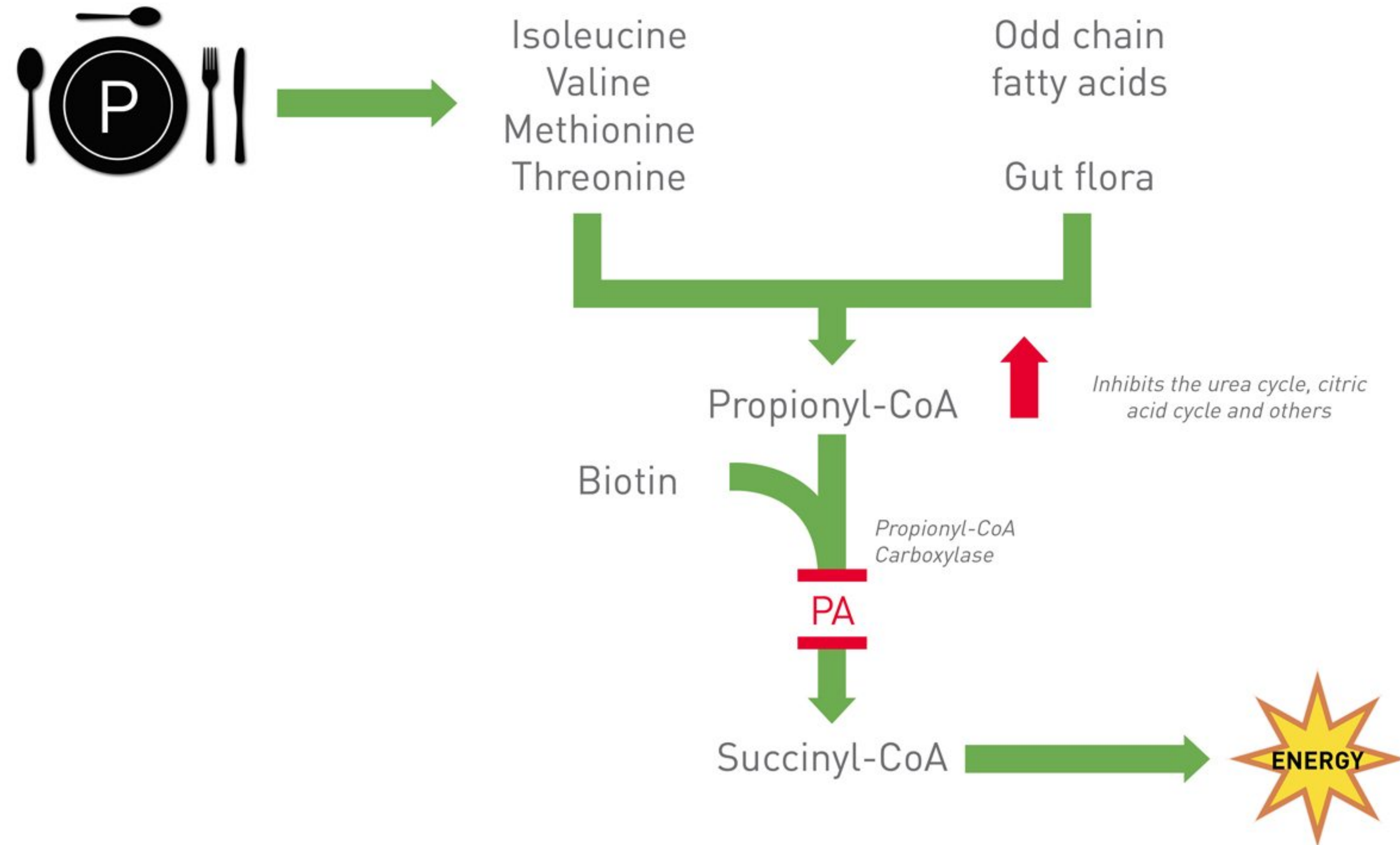
Maple Syrup Urine Disease (MSUD)

*Occurring mainly
in the liver...*



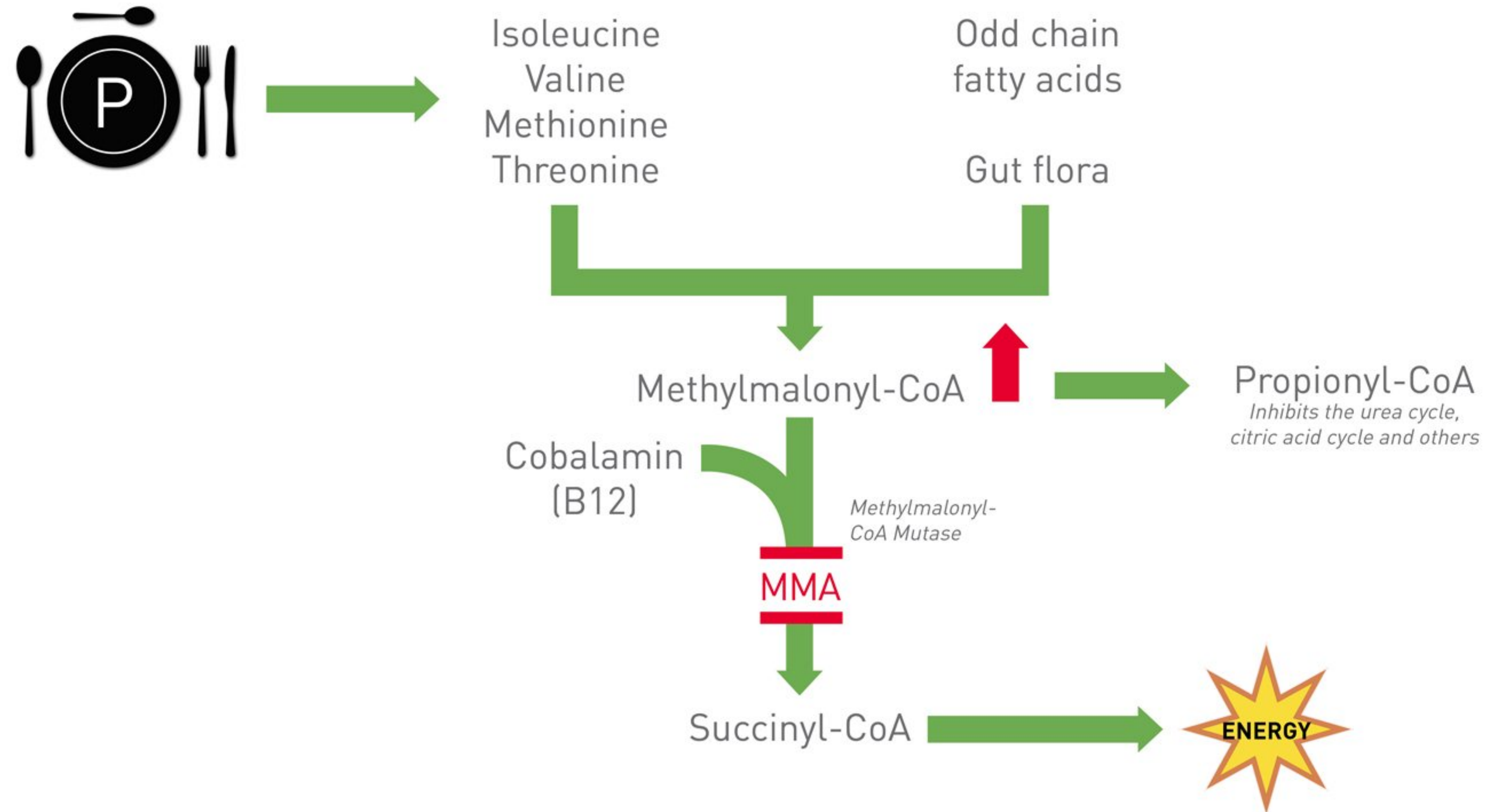
Propionic Acidemia (PA)

*Occurring across tissues
including liver...*

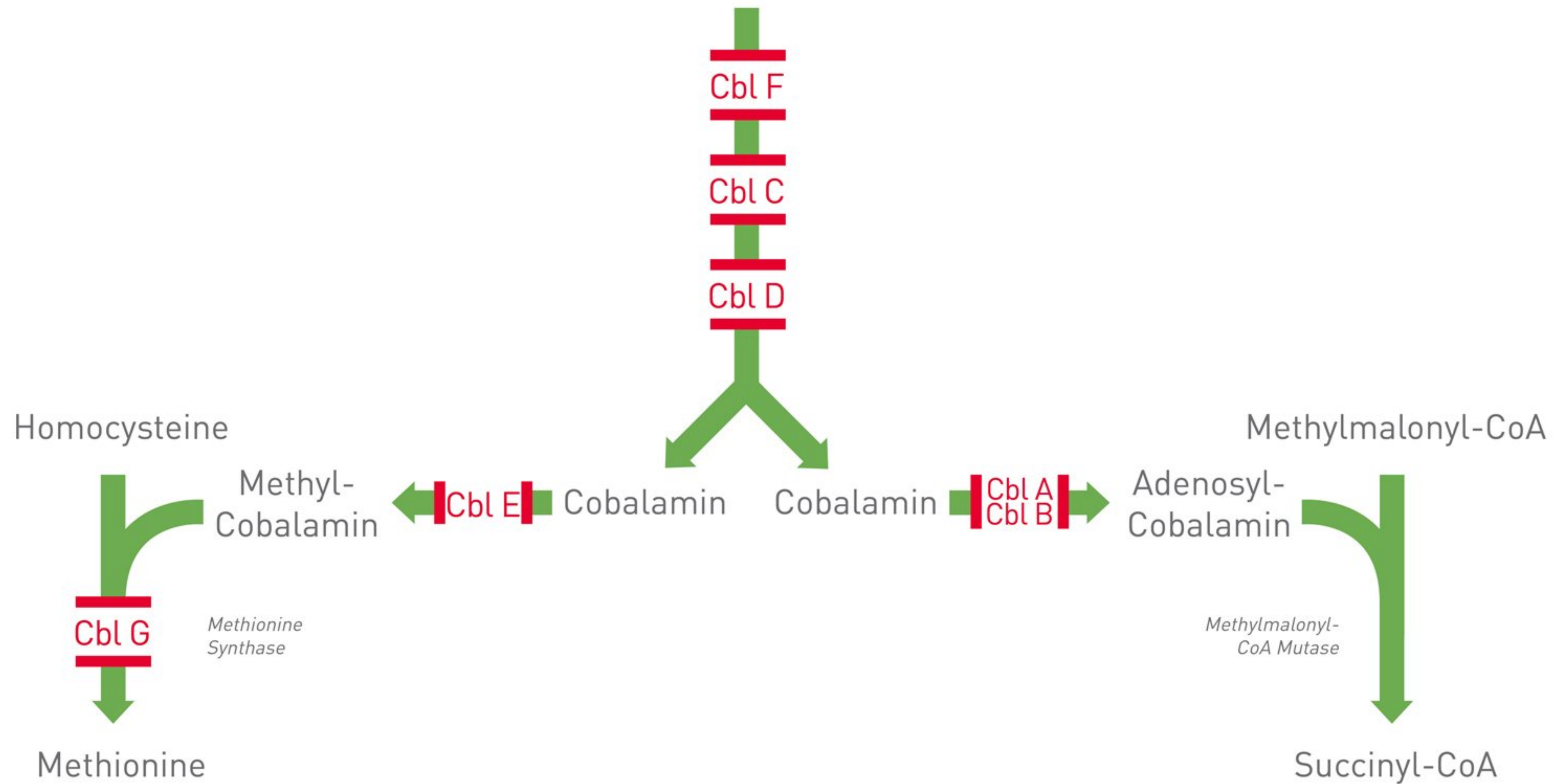


Methylmalonic Acidemia (MMA)

Occurring across tissues including liver...

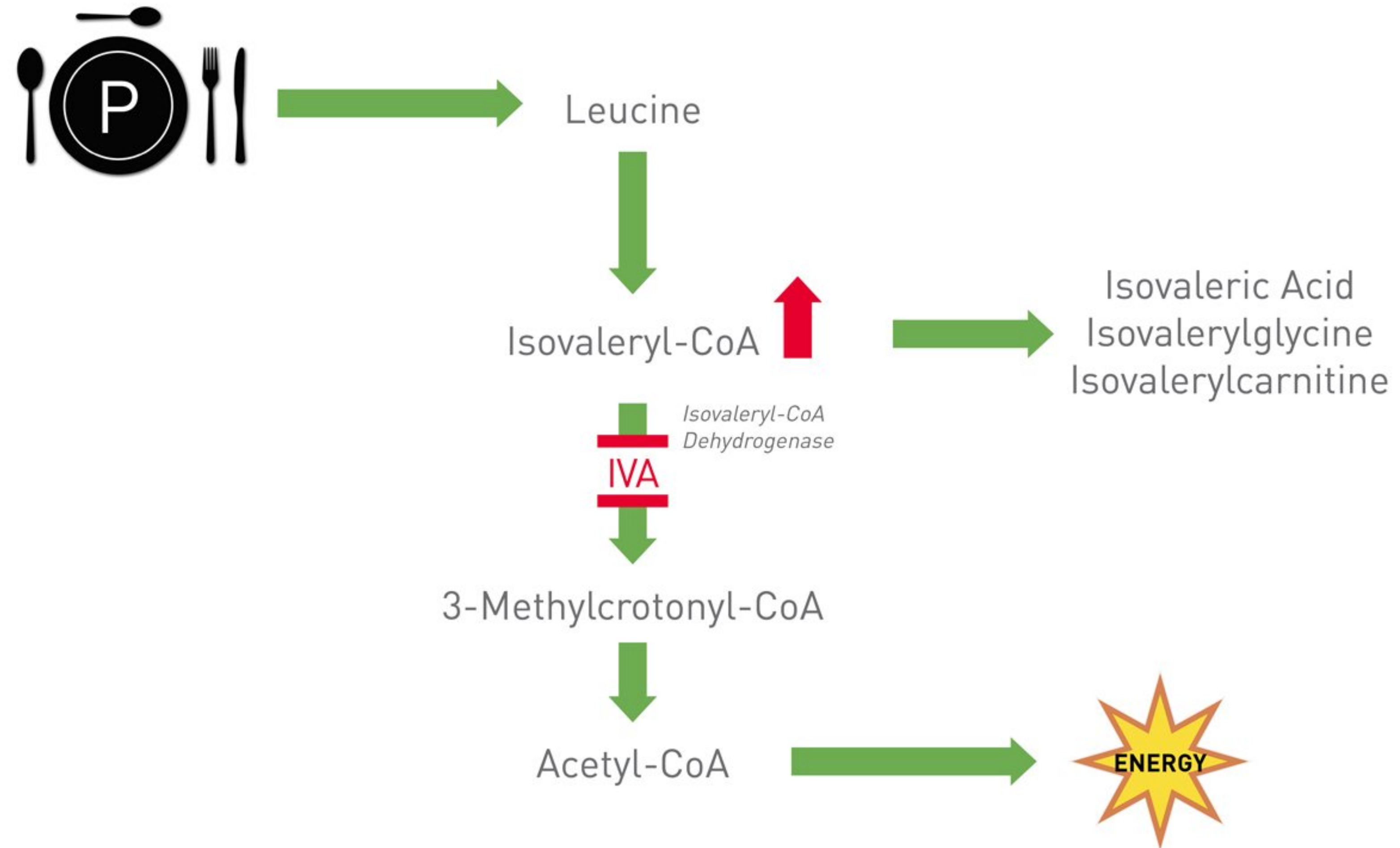


Cobalamin (B12) Processing



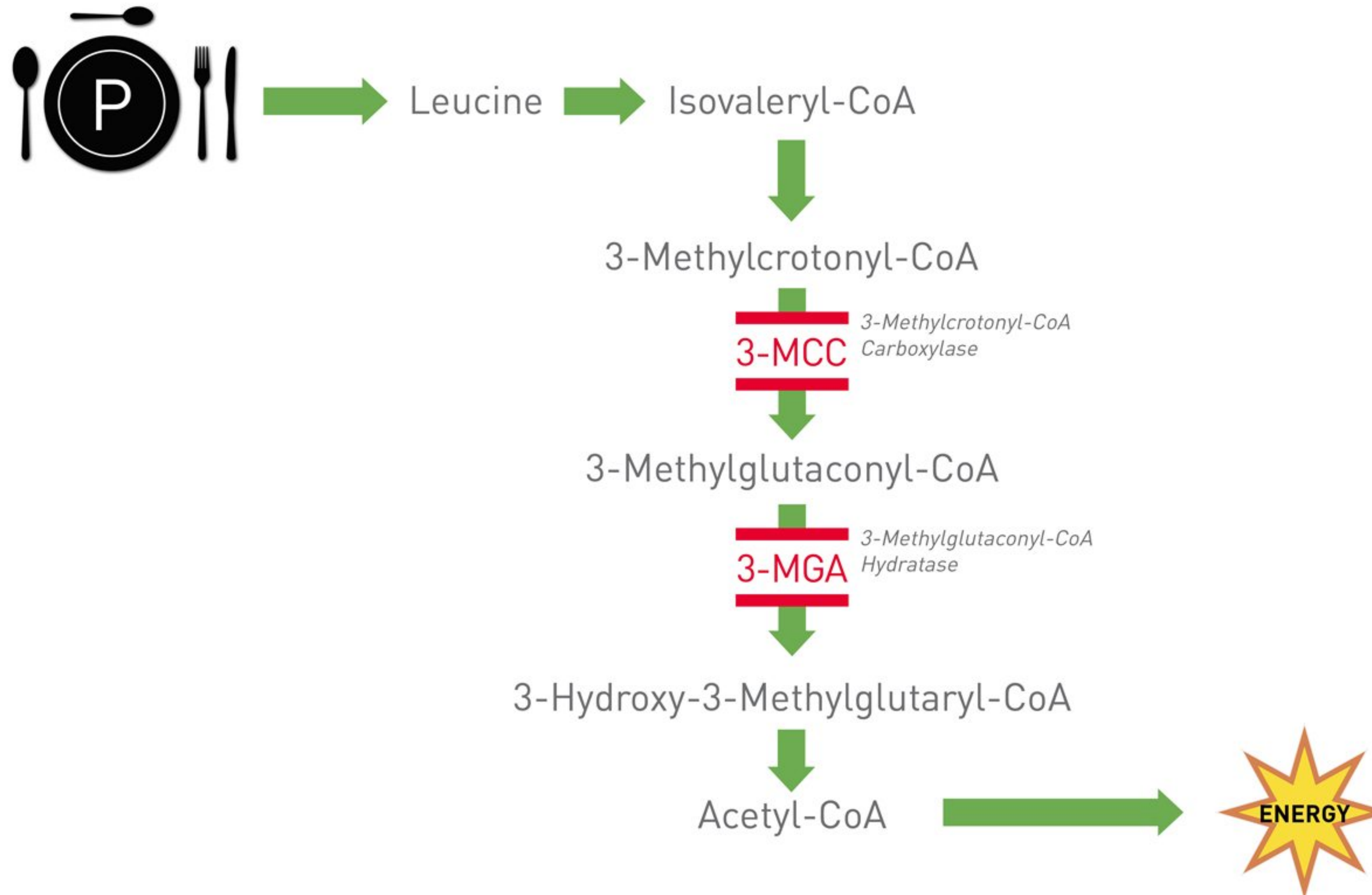
Isovaleric Acidemia (IVA)

Occurring across tissues including liver and kidney...



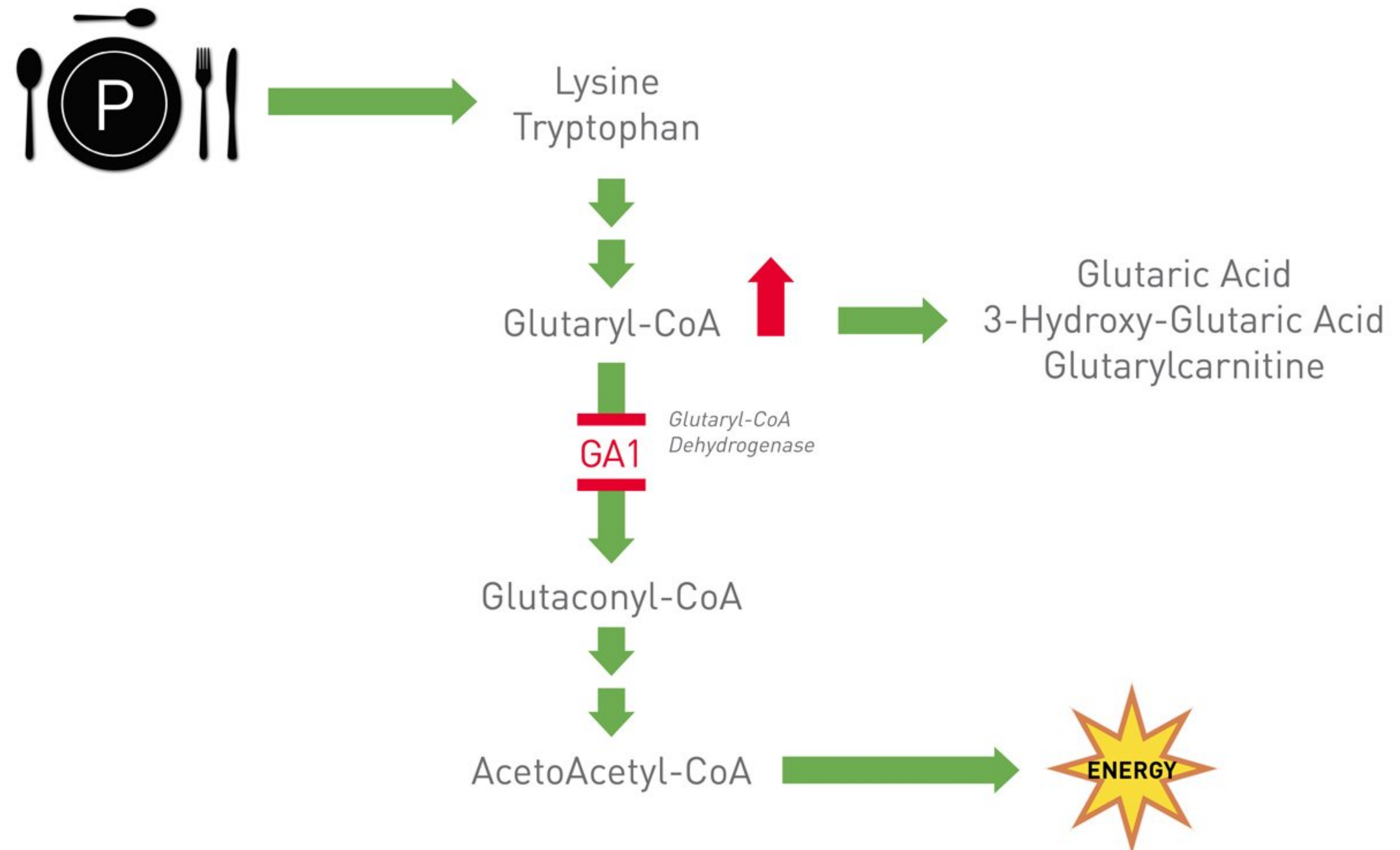
Leucine Metabolism (3-MCC, 3-MGA)

*Occurring across tissues
including liver and kidney...*



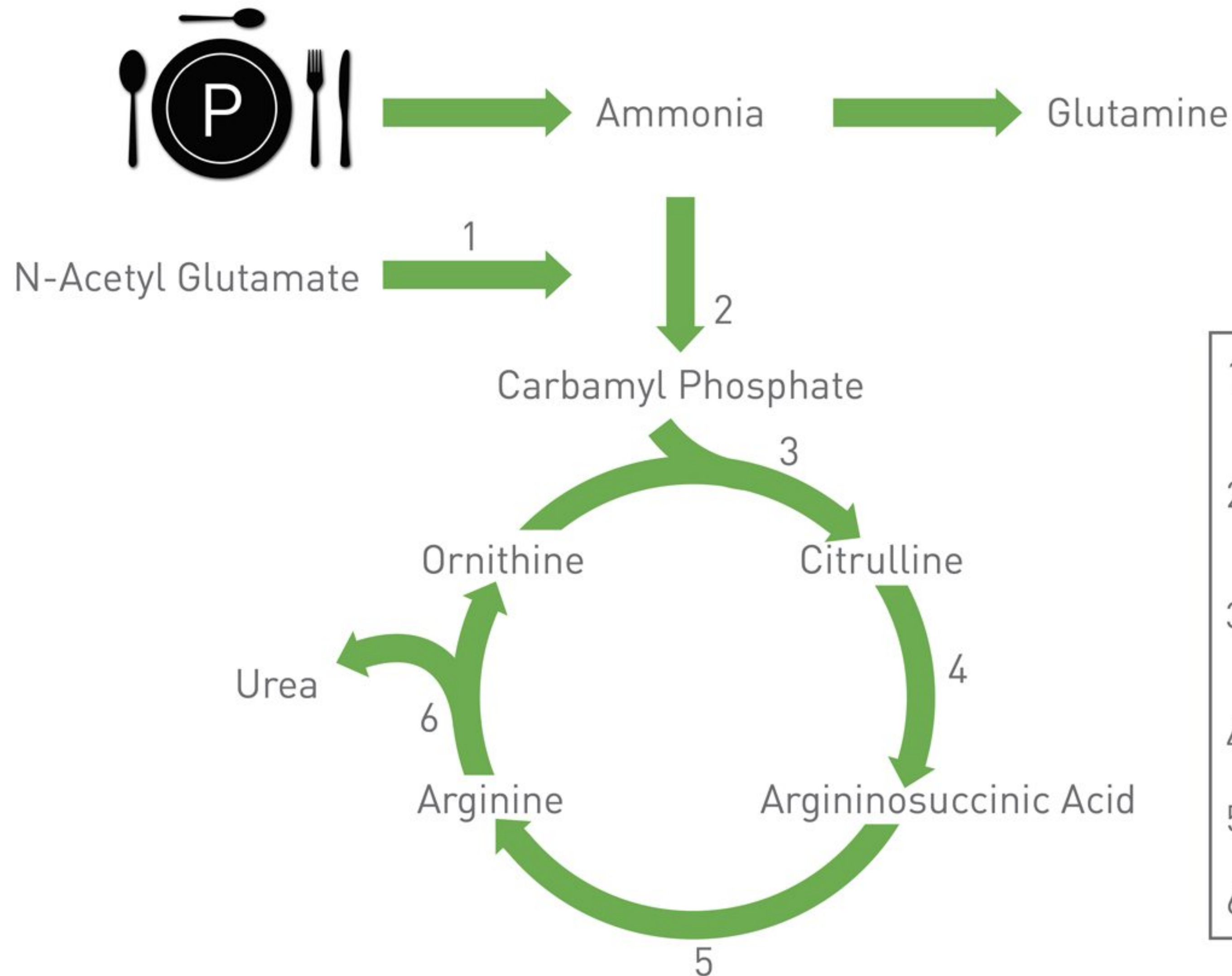
Glutaric Acidemia, Type 1 (GA1)

*Occurring mainly in the
liver and brain...*



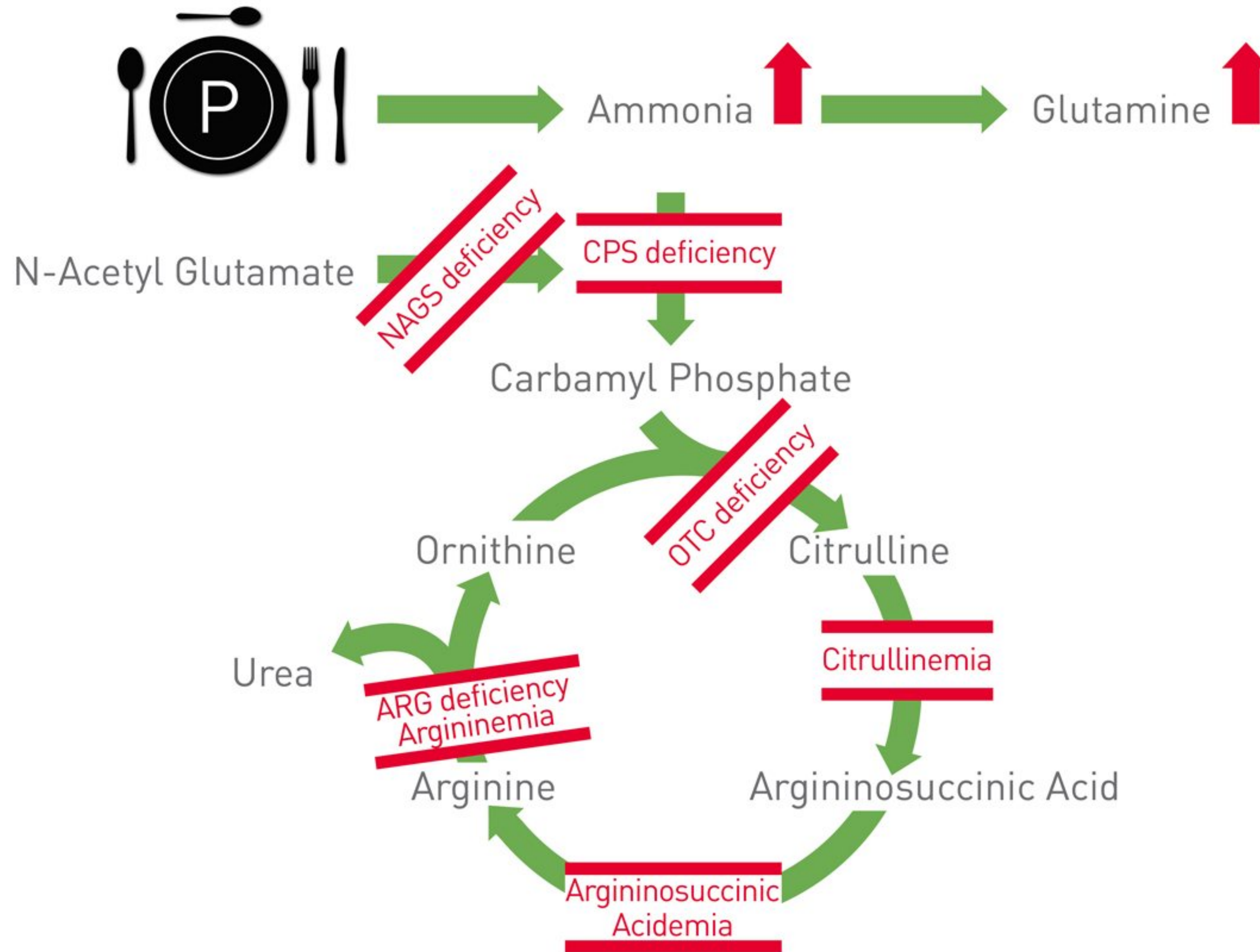
The Urea Cycle

*Occurring mainly
in the liver...*



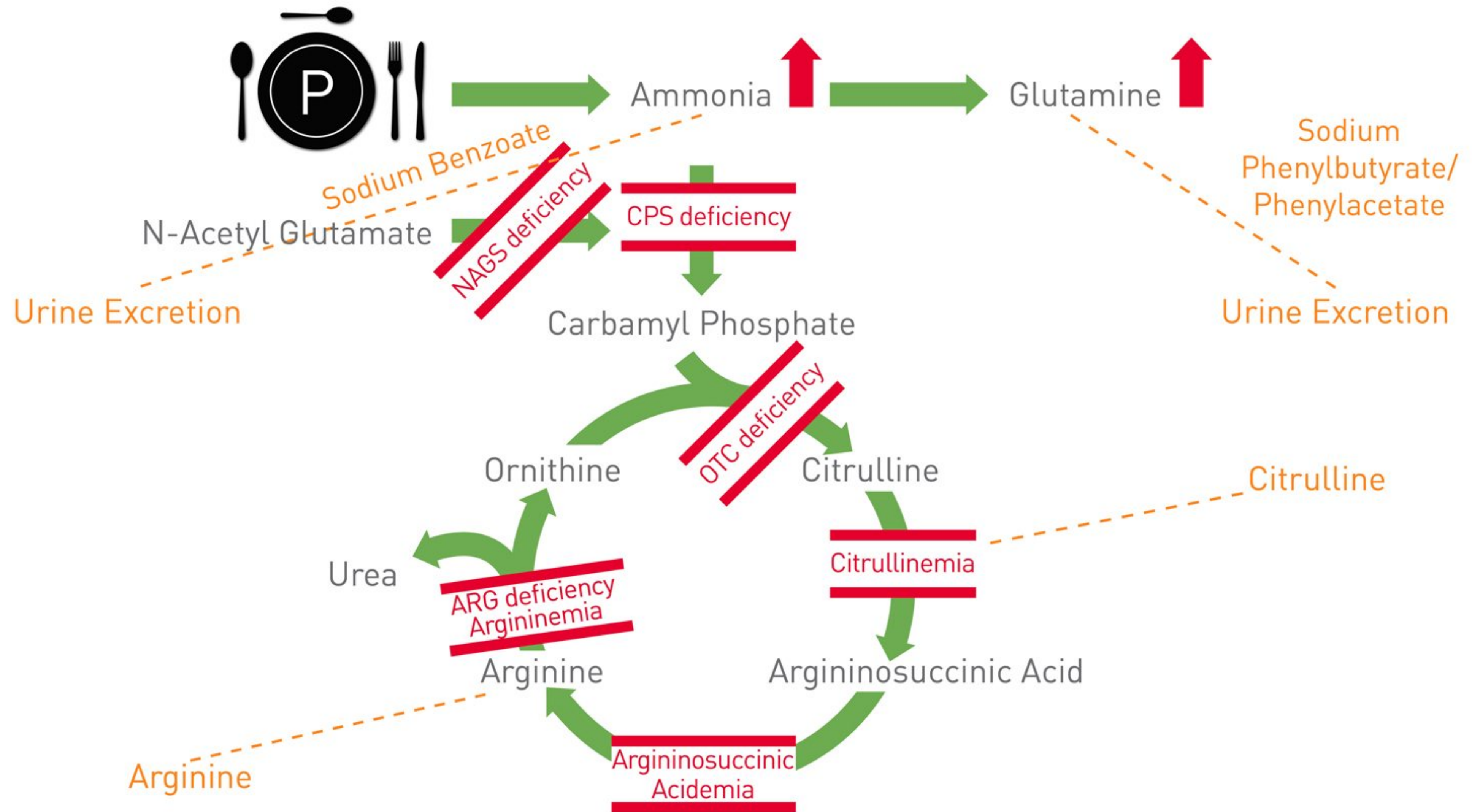
The Urea Cycle

Occurring mainly
in the liver...



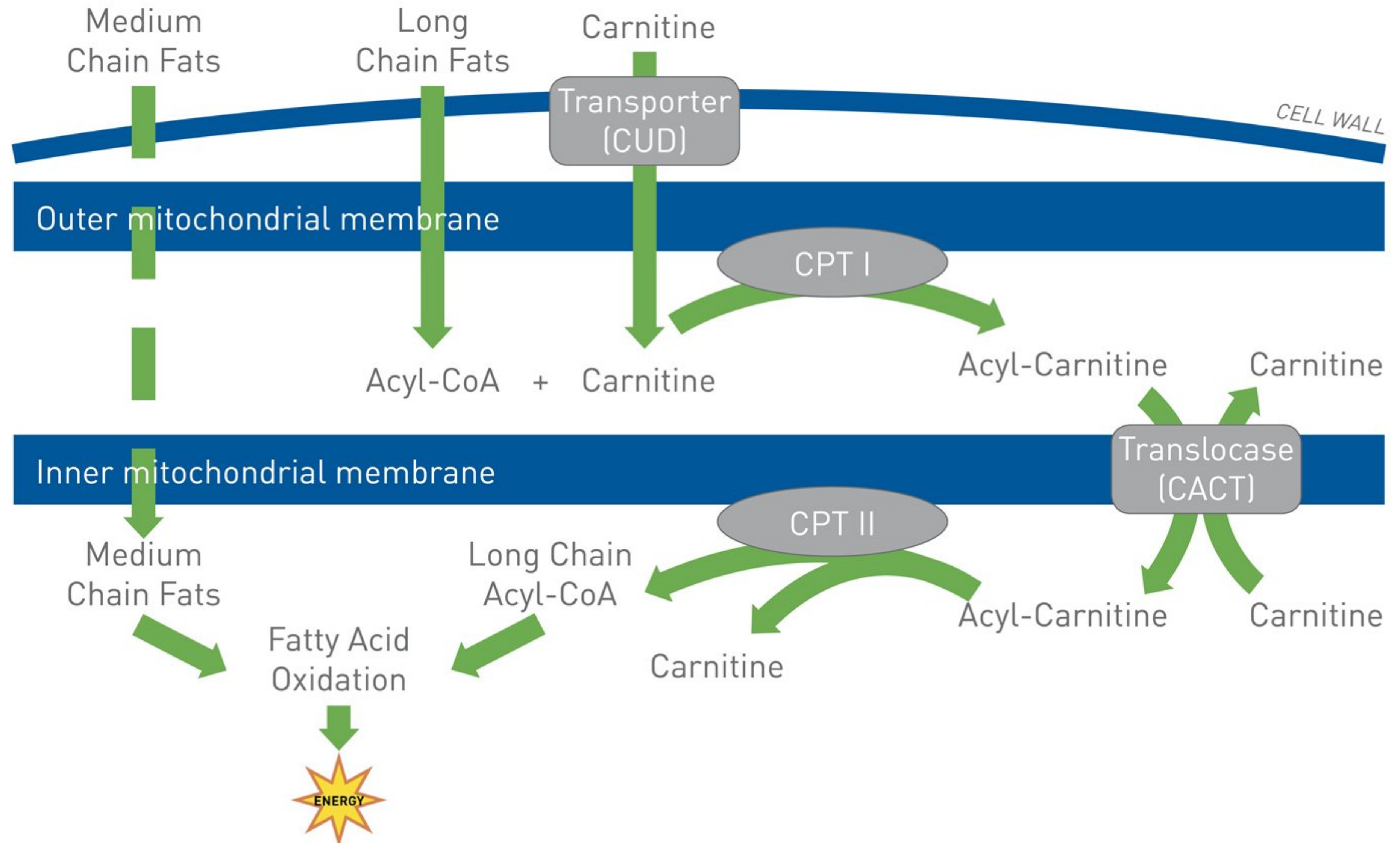
The Urea Cycle

*Occurring mainly
in the liver...*



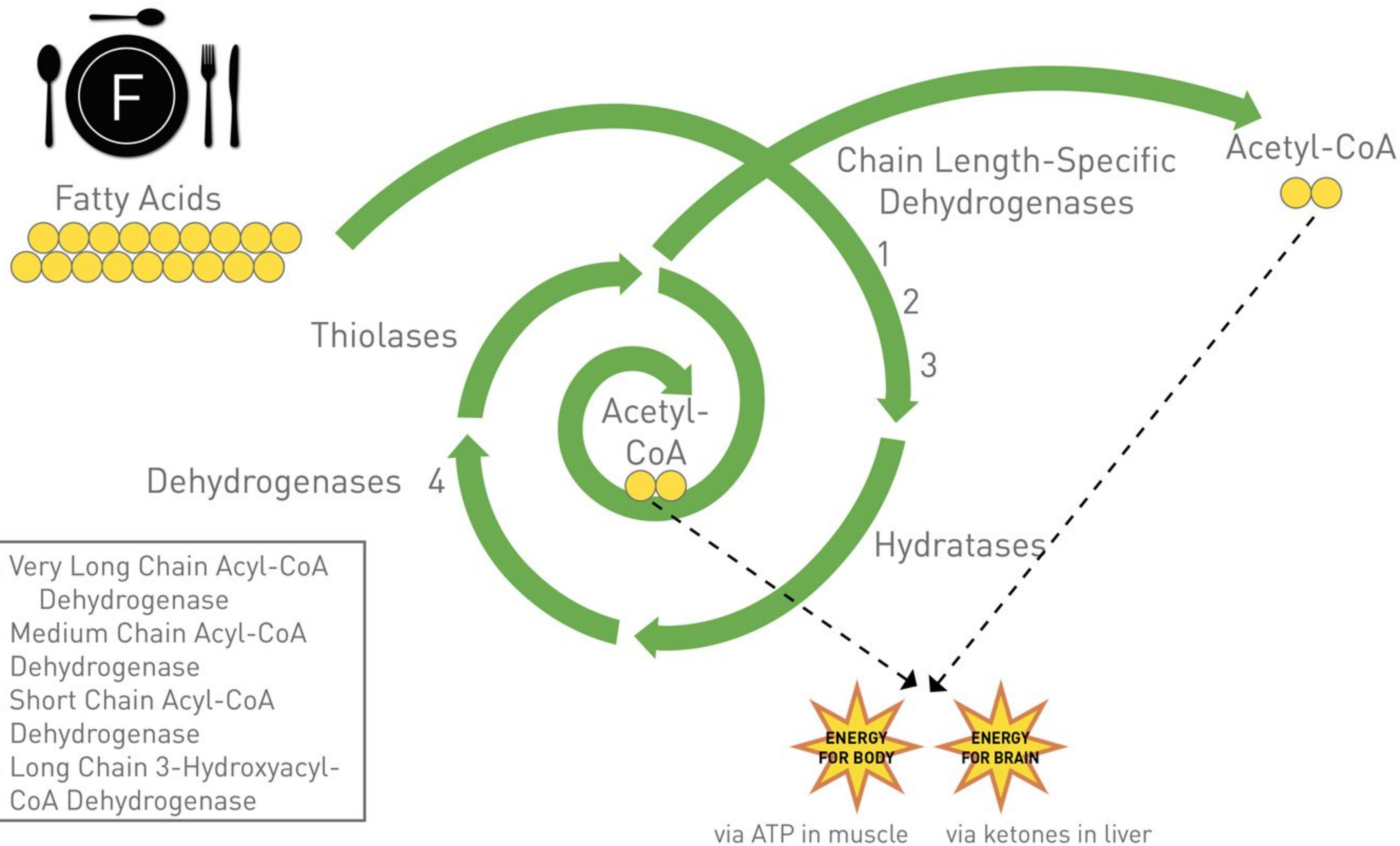


The Carnitine Cycle



Fatty Acid Oxidation

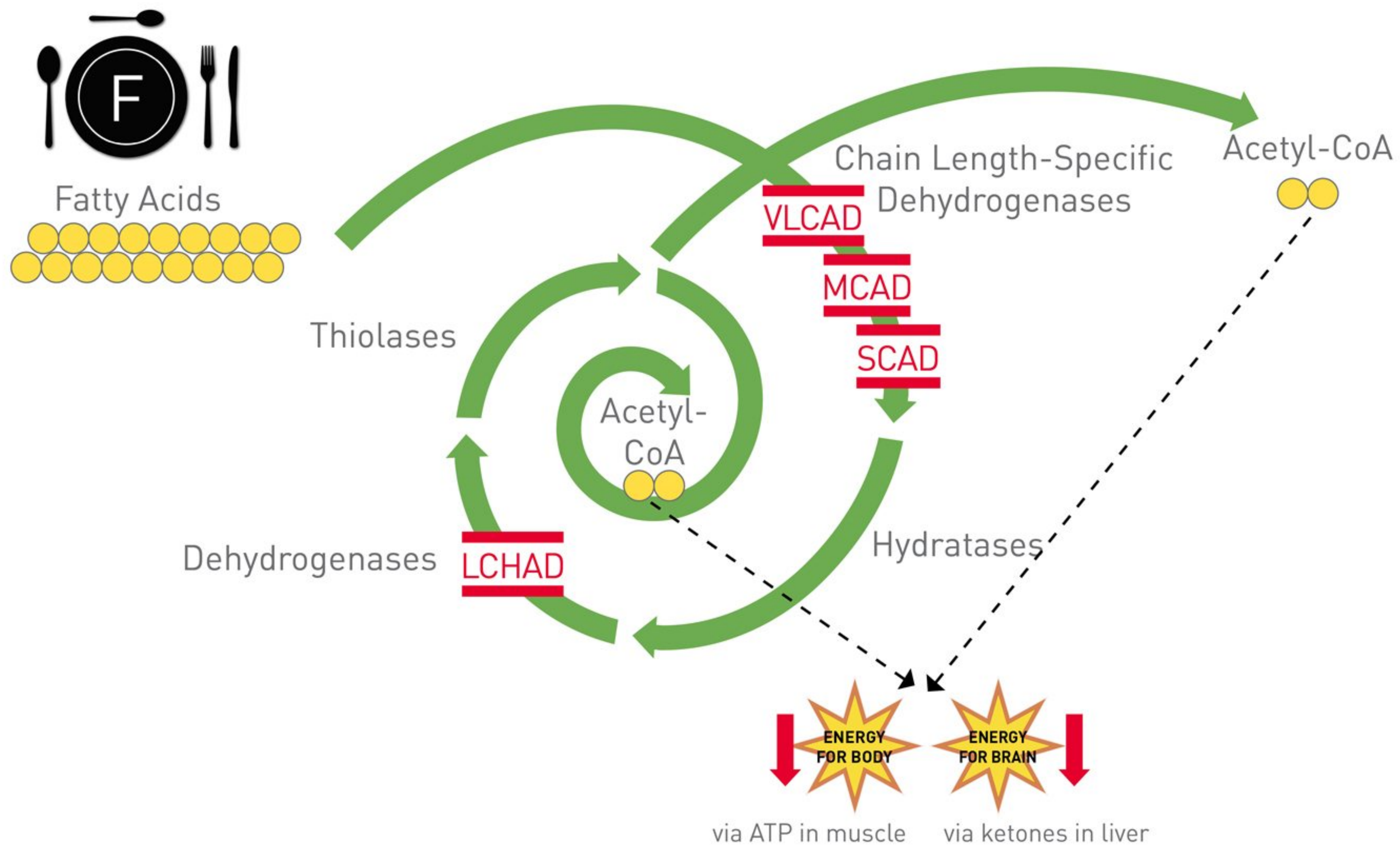
Occurring mainly
in the heart, muscle,
and liver...



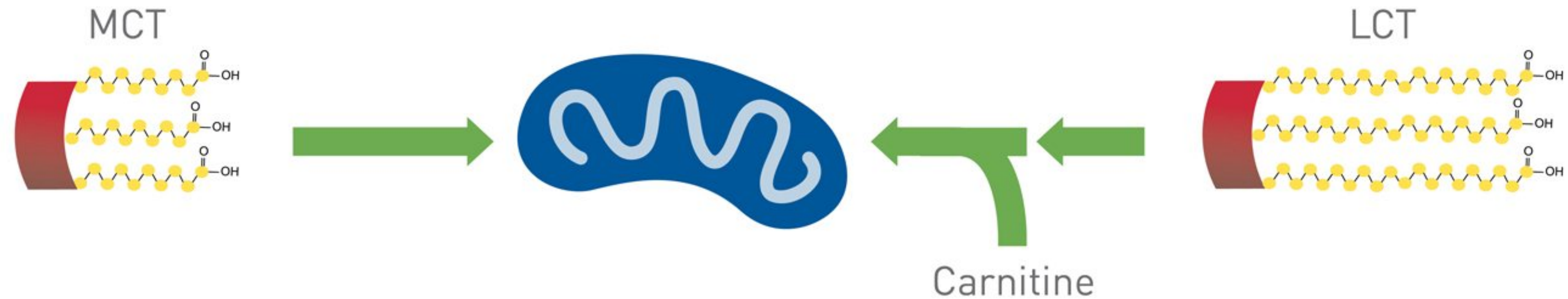
1. Very Long Chain Acyl-CoA Dehydrogenase
2. Medium Chain Acyl-CoA Dehydrogenase
3. Short Chain Acyl-CoA Dehydrogenase
4. Long Chain 3-Hydroxyacyl-CoA Dehydrogenase

Fatty Acid Oxidation

*Occurring mainly
in the heart, muscle,
and liver...*



Medium Chain Triglycerides (MCT) in Fatty Acid Oxidation Disorders



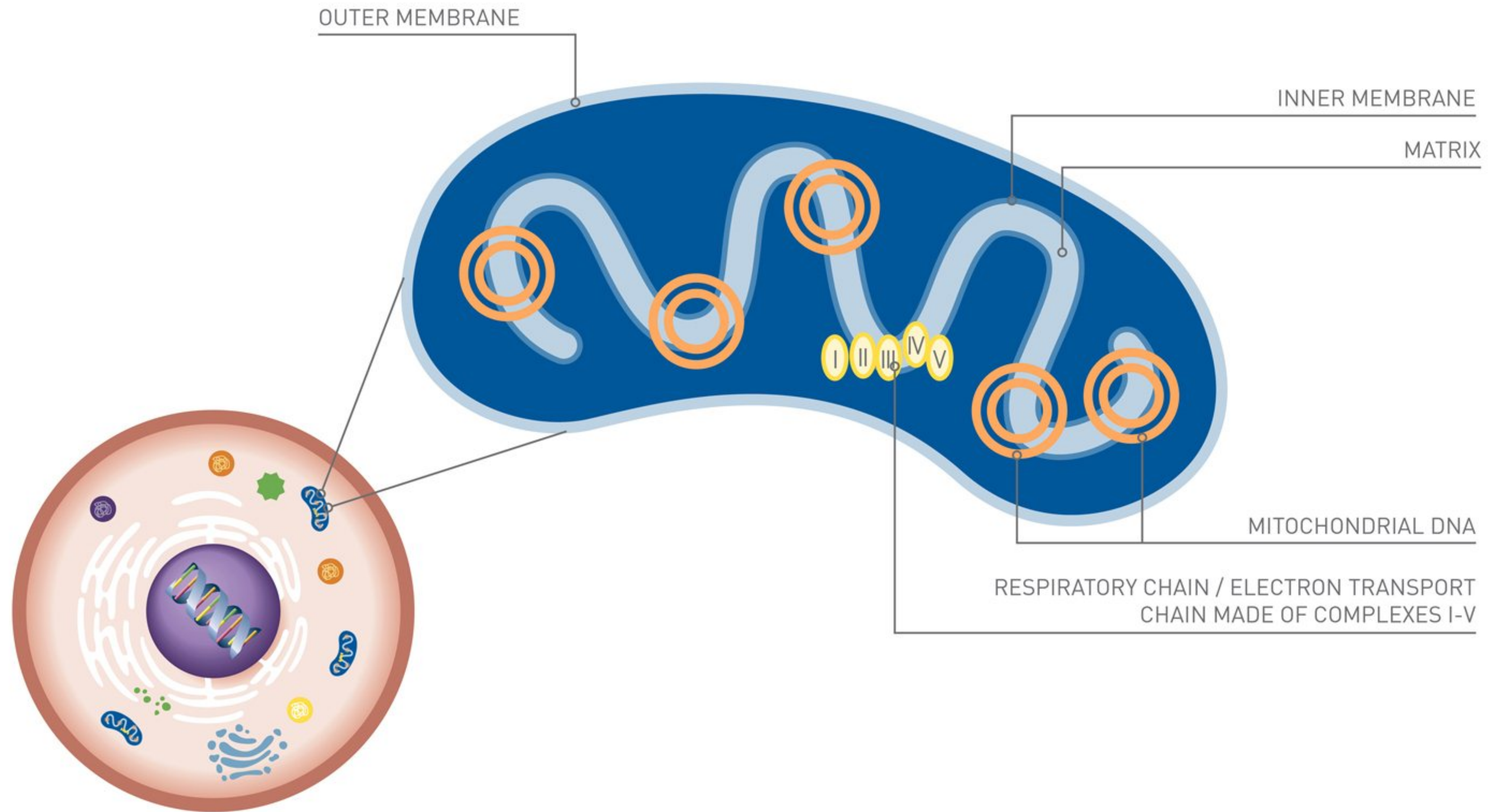
Medium Chain Triglycerides (MCTs):

MCTs are fats of medium carbon chain length (6-12 carbons). MCTs are used to make energy through fatty acid oxidation inside the mitochondria. MCTs may be given in formula or as a supplement. They are found naturally in coconut and palm oils and are also made by our bodies by breaking down long chain fats.

Long chain fats enter the mitochondria with the help of carnitine, which acts like a shuttle. In some disorders of the carnitine cycle, a block prevents long chain fats from entering the mitochondria. MCTs do not require carnitine to be transported into the mitochondria. Therefore, MCTs can be used for making energy through fatty acid oxidation despite a problem with the carnitine shuttle.

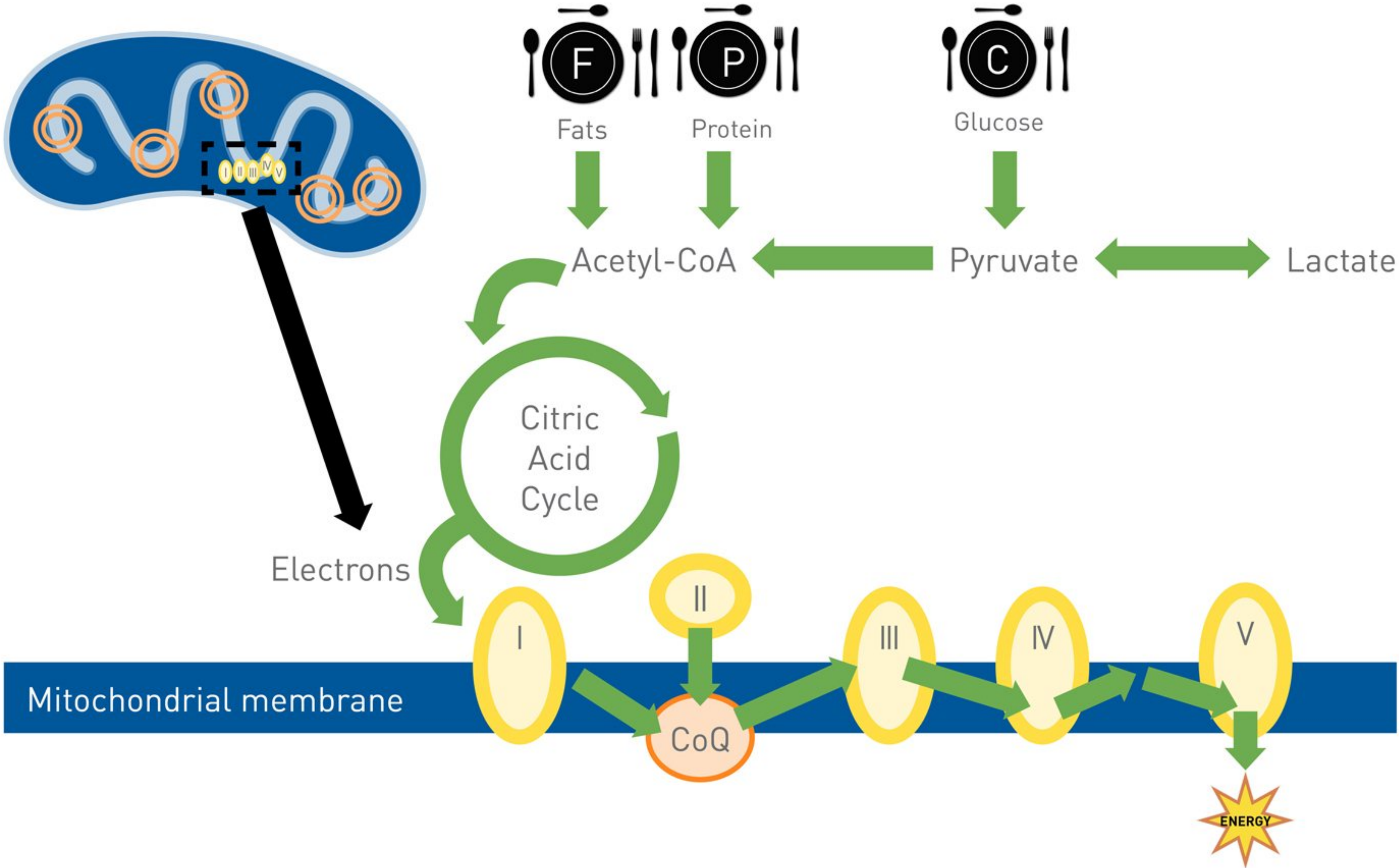
In disorders of long chain fat metabolism, the fats enter the mitochondria but then cannot be broken down into smaller chain lengths and used to make energy. MCTs bypass this first step of the breakdown process, allowing fatty acid oxidation to proceed.

Mitochondria



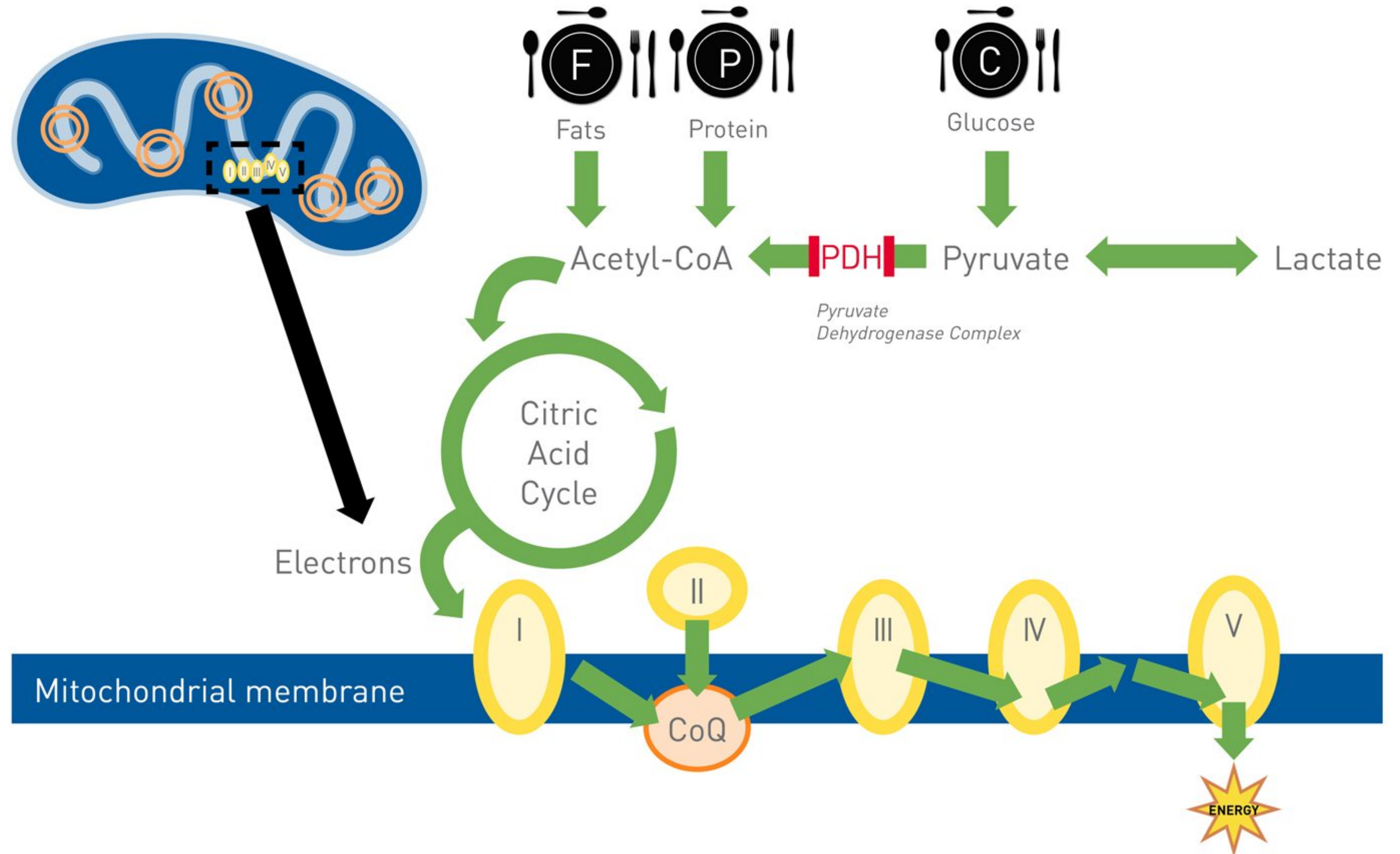
Electron Transport Chain

*Occurring across
most tissues...*

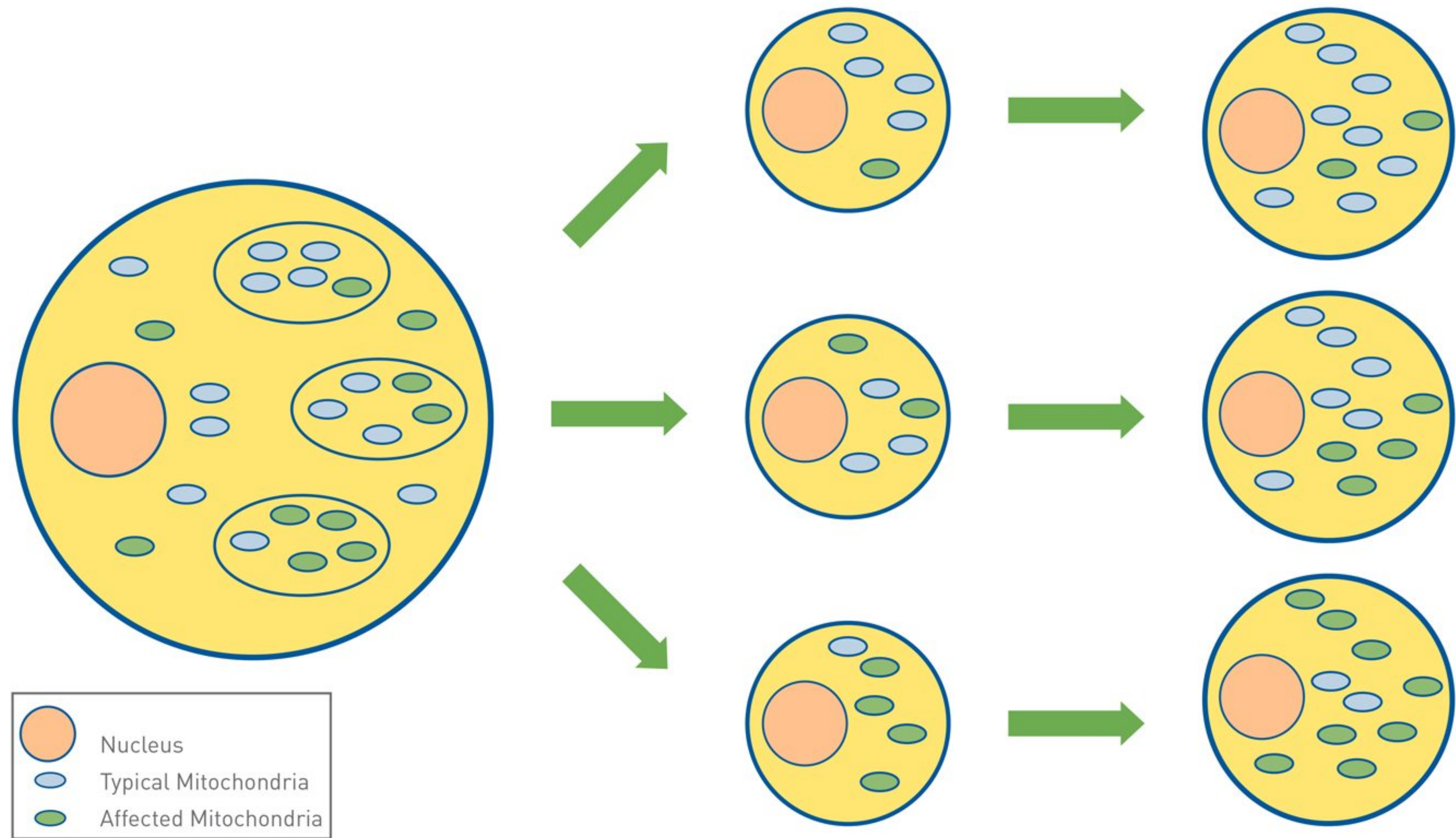


Pyruvate Dehydrogenase

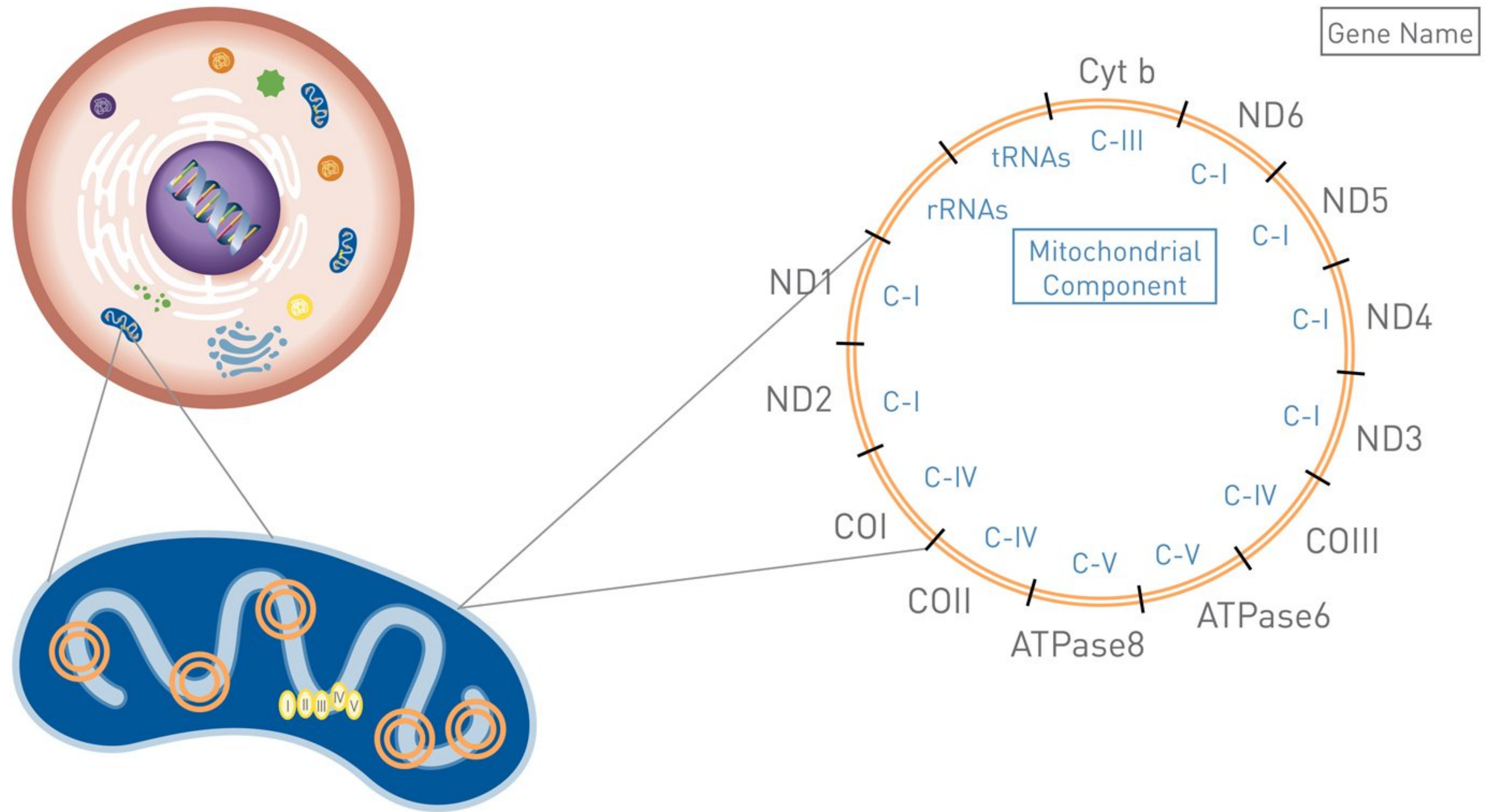
Occurring across
most tissues...



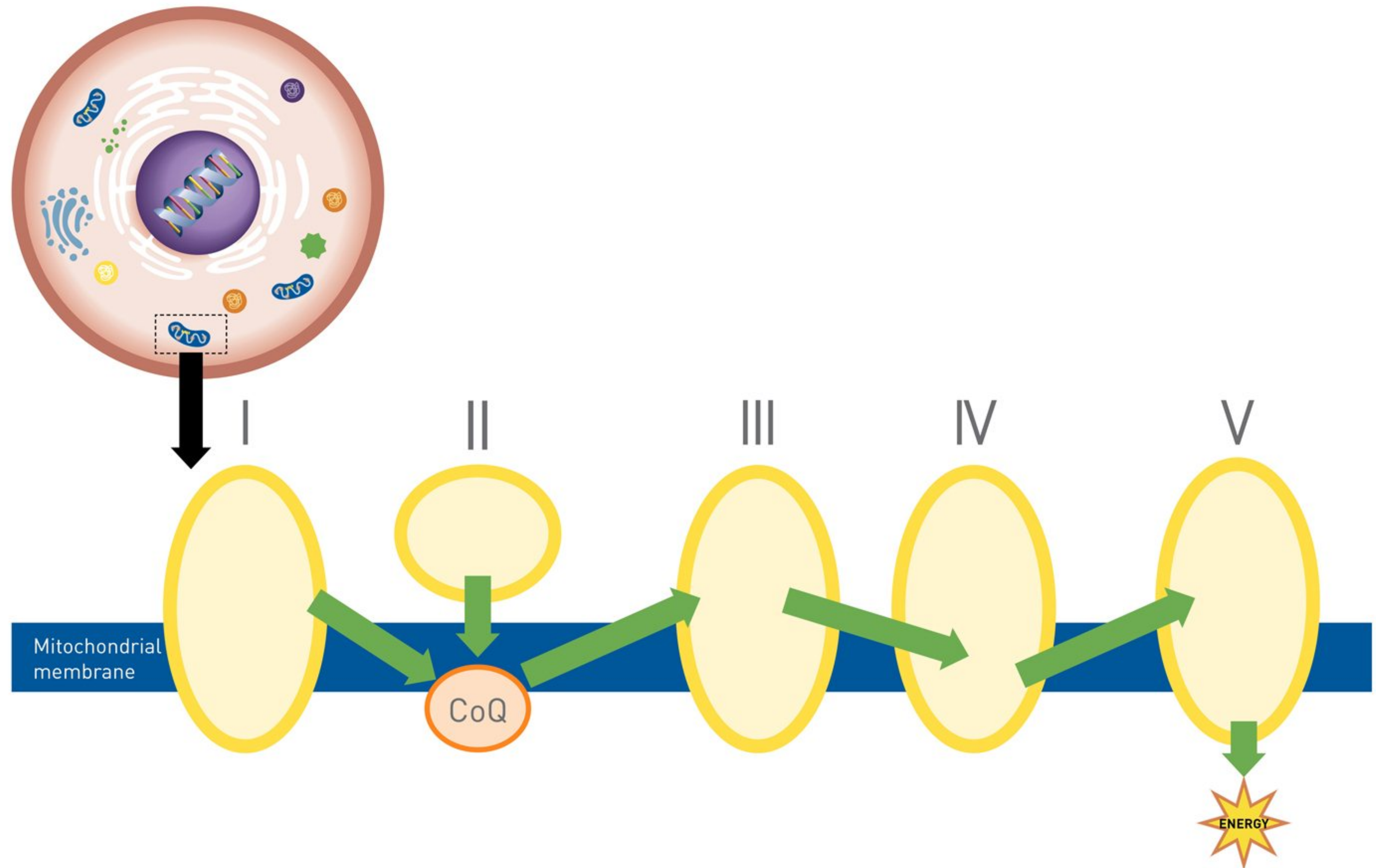
Mitochondrial Heteroplasmy



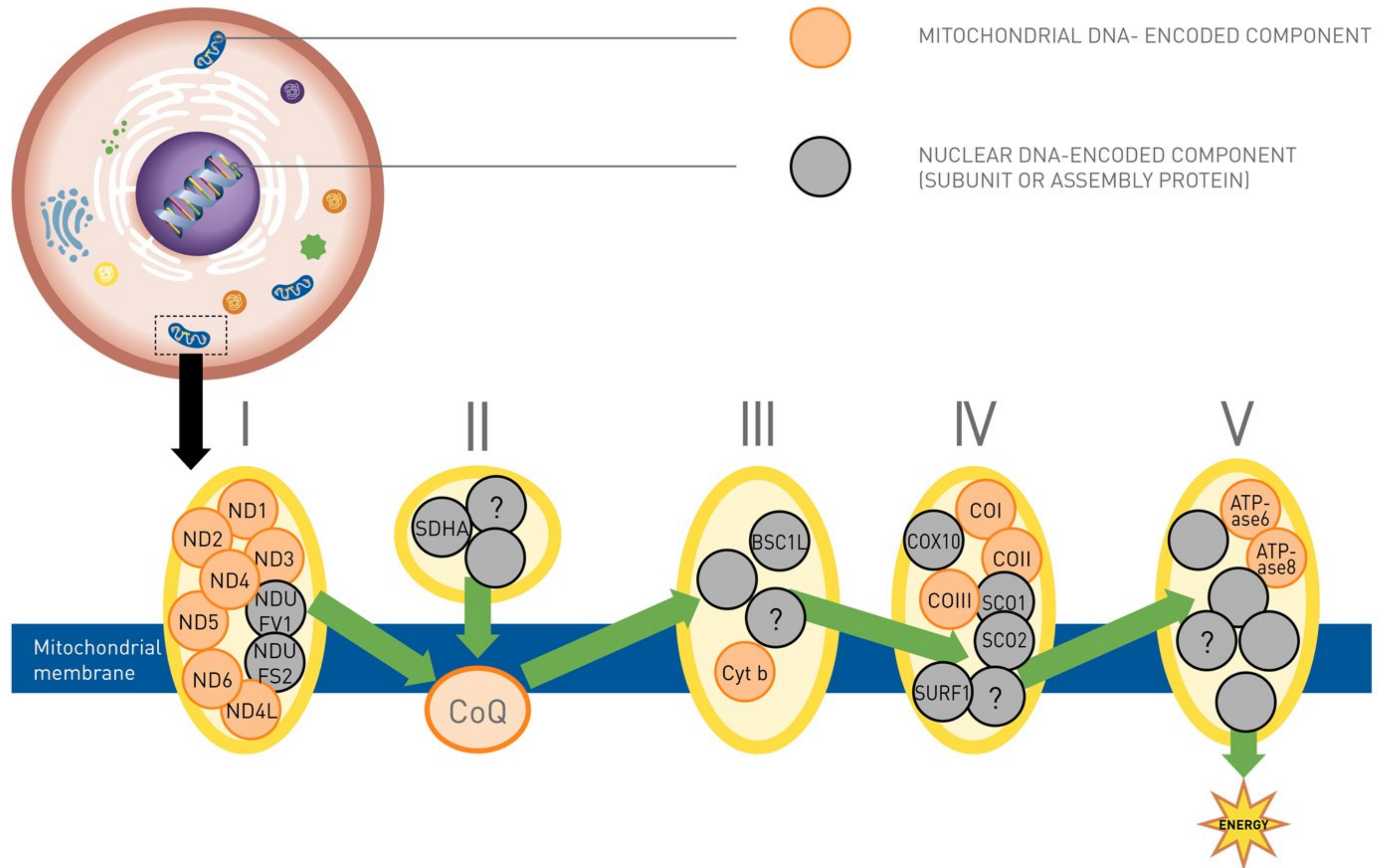
Mitochondrial DNA



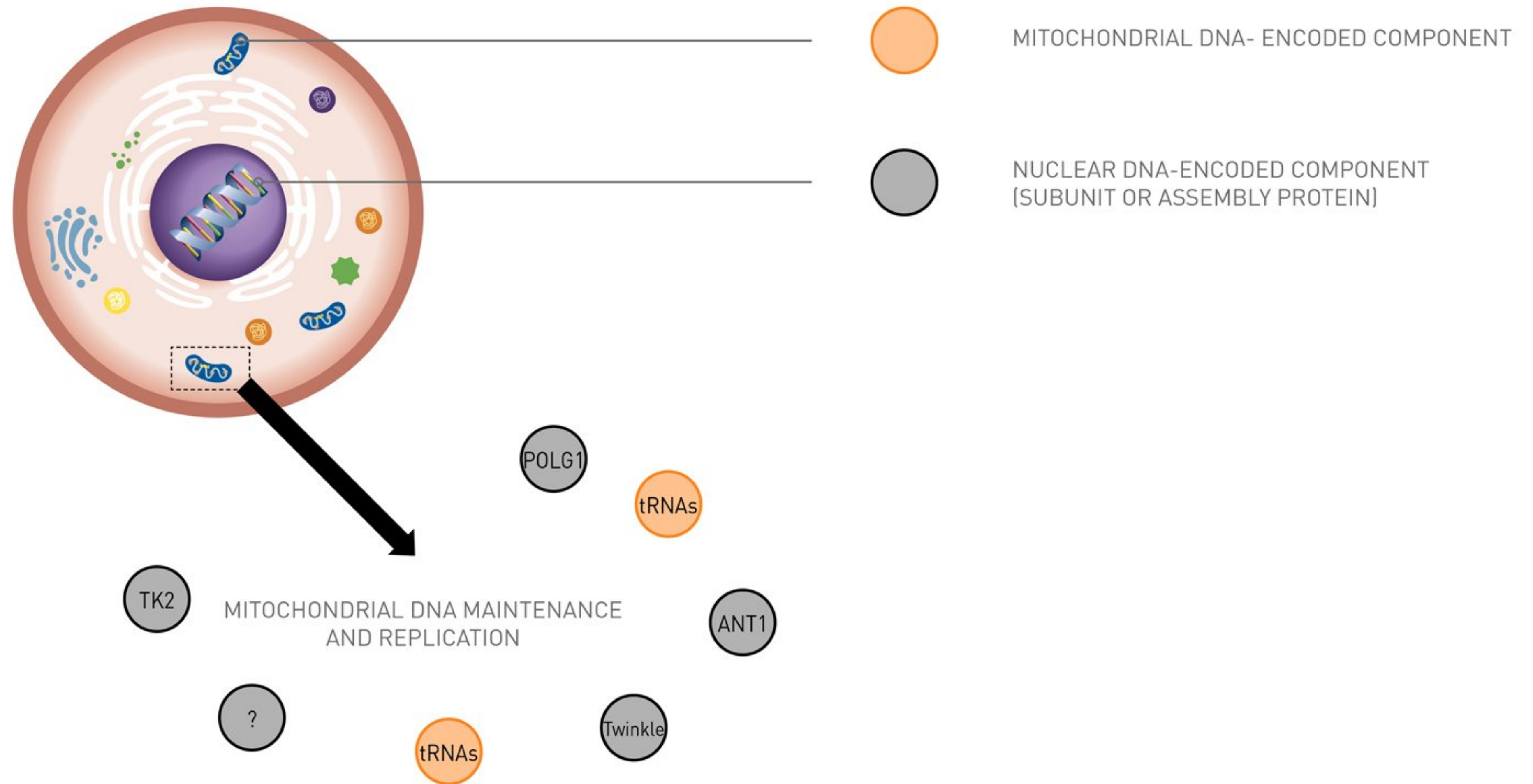
Nuclear and Mitochondrial DNA



Nuclear and Mitochondrial DNA

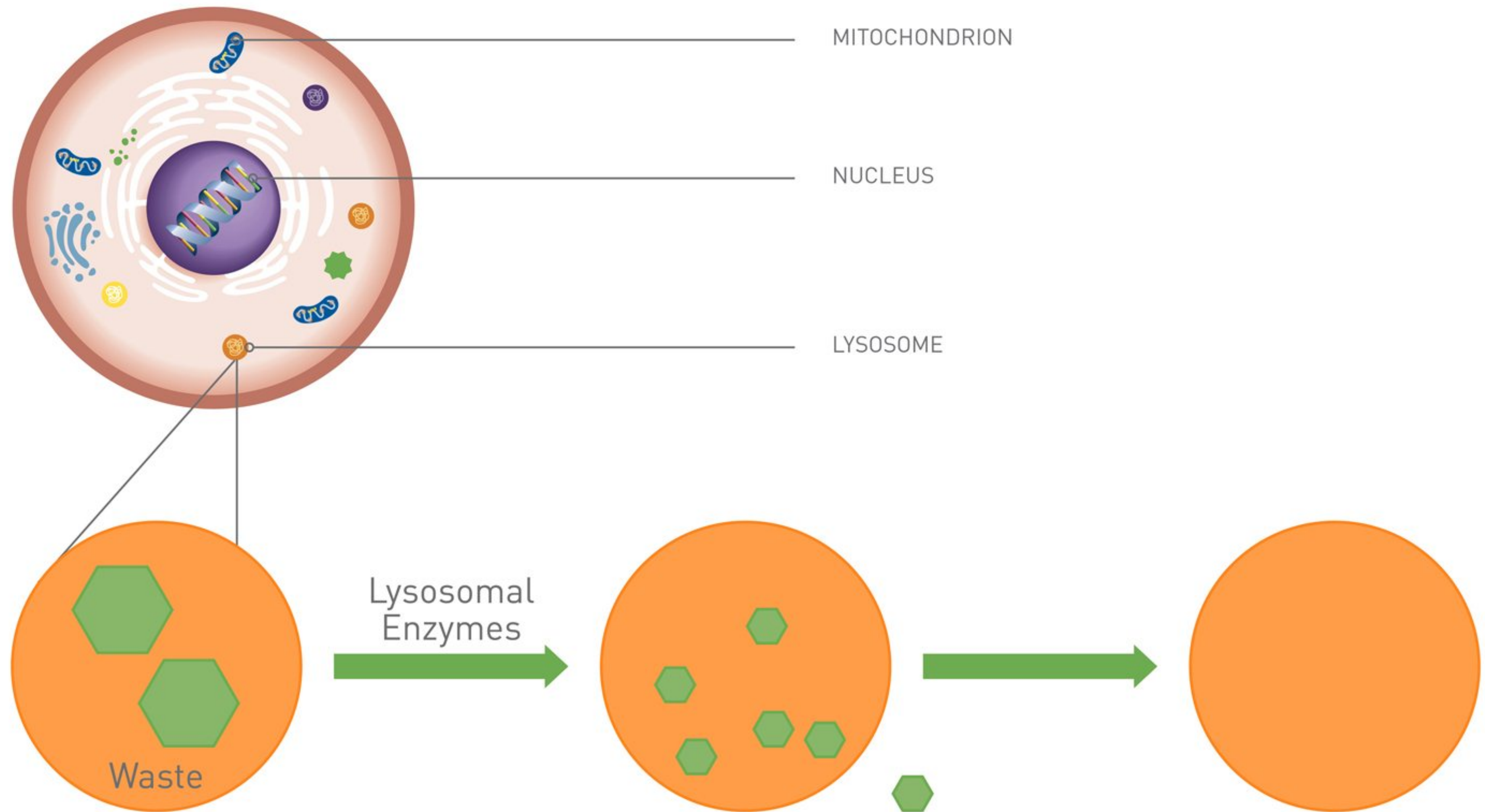


Nuclear and Mitochondrial DNA



Lysosomal Storage

*Occurring across
most tissues...*



Lysosomal Storage Disorder

*Occurring
across
most tissues...*

