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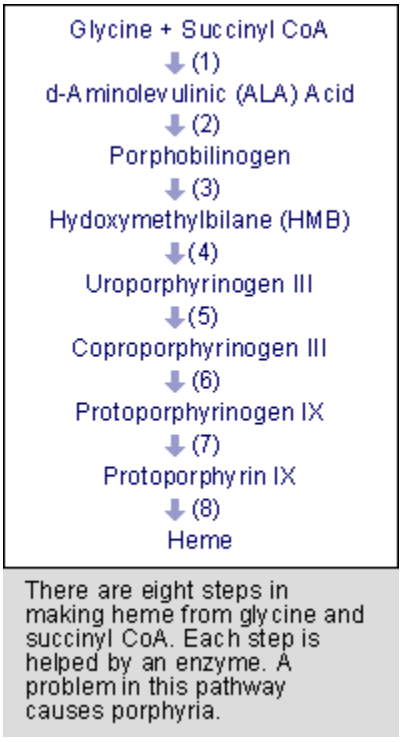
Porphyria



There are eight steps in making heme from glycine and succinyl CoA. Each step is helped by an enzyme. A problem in this pathway causes porphyria.

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Porphyria is a diverse group of diseases in which production of heme is disrupted. Porphyria is derived from the Greek word "porphyra", which means purple. When heme production is faulty, porphyrins are overproduced and lend a reddish-purple color to urine.

Heme is composed of porphyrin, a large circular molecule made from four rings linked together with an iron atom at its center. Heme is the oxygen-binding part of hemoglobin, giving red blood cells their color. It is also a component of several vital enzymes in the liver including the group known as cytochrome P450. This enzyme family is important in converting potentially harmful substances such as drugs to inactive products destined for excretion.

Heme synthesis takes place in several steps, each of which requires a specific enzyme of which there are 8 in total. The genes that encode these enzymes are located on different chromosomes, and mutations of these genes can be inherited in either an autosomal dominant or autosomal recessive fashion, depending on the gene concerned. Affected individuals are unable to complete heme synthesis, and intermediate products, porphyrin or its precursors, accumulate.

Environmental triggers are important in many attacks of porphyria. Example triggers include certain medications, fasting, or hormonal changes. Genetic carriers who avoid a triggering exposure may never experience symptoms.

The cutaneous porphyrias cause sun sensitivity, with blistering typically on the face, back of the hands, and other sun-exposed areas. The most common of these is porphyria cutanea tarda (PCT). Triggering factors are alcohol use, estrogen, iron, and liver disease, particularly hepatitis C.

The acute porphyrias typically cause abdominal pain and nausea. Some patients have personality changes and seizures at the outset. With time the illness can involve weakness in many different muscles.

The cutaneous and acute forms are treated differently. Cure of these genetic diseases awaits the results of ongoing research on the safest and most effective means of gene transfer or correction.

Step in Pathway	Enzyme	Disease caused by enzyme deficiency
1	ALA synthase	
2	ALA dehydratase	ALAD porphyria
3	HMB synthase	Acute intermittant porphyria
4	Uroporphyrinogen synthase (UROS)	Congenital erythropoietic porphyria
5	Uroporphyrinogen decarboxylase (UROD)	Porphyria cutanea tarda
6	Coproporphyrinogen oxidase	Hereditary copropophyria
7	Protoporphyrinogen oxidase	Variegate pophyria
8	Ferrochelatase	Erythropoietic protoporphyria

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