(7) Biochemical Aspect of Porphyria

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The classification of porphyrias has depended chiefly on the site where the increased porphyrin concentration was found.

In a biopsy specimen of the bone marrow obtained from a patient with porphyria congenita, activities of δ -aminolevulinic acid (ALA) synthetase, ALA dehydrase and porphyrin formation from ALA were elevated to approximately twice the control. No increase of activities of ALA synthetase and ALA dehydrase was observed in the patient's liver, though moderately increased porphyrin concentrations were found in it.

On the contrary, in the liver of a patient with acute intermittent porphyria, ALA synthetase activity was increased markedly and ALA dehydrase activity was increased to twice the control, while those in the bone marrow were almost unchanged.

The administration of 3, 5-dicarbethoxy-1, 4-dihydrocollidine (DDC) and griseofulvin increased the circulating erythrocyte and plasma protoporphyrin concentration in mice activities of δ -aminolevulinic acid (ALA) synthetase, heme synthetase in the spleen, and formation of porphyrins from ALA in erythrocyte were unchanged. After an intraperitoneal injection of protoporphyrin, protoporphyrin concentration in the circulating erythrocyte increased. The overproduction of protoporphyrin in the liver induced by the porphyria-inducing chemicals caused a rise in the concentration of plasma protoporphyrin, which apparently penetrated into the red blood cells of mice, resulting in an increased erythrocyte protoporphyrin concentration.

The classification of porphyrias should be based on the site where abnormal enzymatic processes actually occur.