

PANTOTHENATE KINASE-2 DEFICIENCY

Marc E. Tischler, PhD;
University of Arizona

PANTOTHENATE KINASE-2 DEFICIENCY

- coenzyme A is:
 - ✓ synthesized from the vitamin pantothenic acid (Fig. 1)
 - ✓ essential for storage of fatty acids in fat cells
 - ✓ essential for the complete oxidation of both fatty acids and glucose to provide usable energy in all cells of the body that require oxygen
- insufficient energy production particularly affects:
 - ✓ brain causing a severe neurological disorder
 - ✓ muscle function accounting for symptoms such as:
 - writhing hand and finger movements
 - inability to relax muscles
 - speech abnormalities
 - chewing and swallowing difficulty

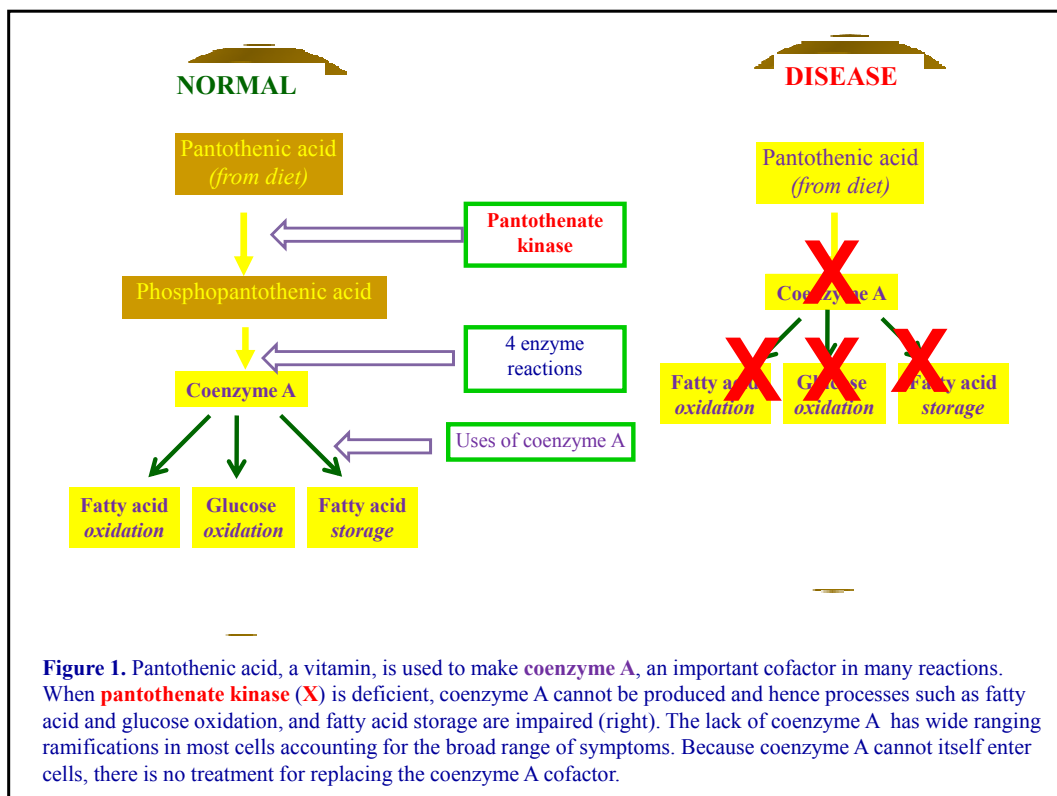


Figure 1. Pantothenic acid, a vitamin, is used to make **coenzyme A**, an important cofactor in many reactions. When **pantothenate kinase (X)** is deficient, coenzyme A cannot be produced and hence processes such as fatty acid and glucose oxidation, and fatty acid storage are impaired (right). The lack of coenzyme A has wide ranging ramifications in most cells accounting for the broad range of symptoms. Because coenzyme A cannot itself enter cells, there is no treatment for replacing the coenzyme A cofactor.