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Pyruvate Dehydrogenase Deficiency

National Cancer Institute

Source

National Cancer Institute. *Pyruvate Dehydrogenase Deficiency*. NCI Thesaurus. Code C103968.

A genetic disorder caused by mutations in the genes that are responsible for production of protein components of the pyruvate dehydrogenase complex. It may present with lactic acidosis. Signs and symptoms include developmental delays, seizures and hypotonia.