Pearson Syndrome

National Cancer Institute

Source

National Cancer Institute. Pearson Syndrome. NCI Thesaurus. Code C115326.

The most severe syndrome in the spectrum of single, large-scale mitochondrial DNA (mtDNA) deletions (SLSMDs), usually presenting shortly after birth with sideroblastic anemia. The condition is often associated with exocrine pancreas insufficiency and multi-system dysfunction including diabetes mellitus, cortisol deficiency, hypothyroidism, hypoparathyroidism, and growth hormone deficiency. Commonly associated clinical findings include the following: failure to thrive, hypotonia, ptosis, ophthalmoparesis, and renal disease.