Carnosinemia is a very rare inherited metabolic disorder characterized by developmental delays and seizures. Symptoms can begin during infancy and may include drowsiness, seizures that may be accompanied by involuntary jerking muscle movements of the arms, legs, or head (myoclonic seizures), and intellectual disability. The symptoms of carnosinemia include extreme drowsiness and seizures that can occur in children under the age of one year. Slow growth, low muscle tone, motor delays, and delayed intellectual development also occur in children with this disorder. Seizures may be accompanied by myoclonic seizures. By approximately 2 years of age, affected children show variable degrees of intellectual deficit leading to intellectual disabilities and developmental regression. Some affected children also have muscle weakness (congenital myopathy). Electroencephalogram (EEG), a test that detects electrical activity in the brain, may be abnormal. A few patients reported with this condition have few or no symptoms. Carnosinemia is a very rare disorder that affects males and females in equal numbers. Approximately 30 individuals with carnosinemia have been reported in the medical literature world-wide. The diagnosis of carnosinemia may be made by testing the levels of amino acids in blood and/or urine, which reveals abnormally high levels of carnosine and anserine in the serum and urine. Very specialized testing of the blood will detect very low activity of the enzyme carnosinase in the blood. Diagnosis is based on amino acid analysis of serum and/or urine after exclusion of meat from the diet.