

Congenital lactic acidosis affects males and females in equal numbers. The exact incidence of congenital lactic acidosis is unknown. One estimate places the incidence at 250-300 live births per 1,000 per year in the United States. However, it is likely that many cases go undiagnosed or misdiagnosed, making it difficult to determine the true frequency of congenital lactic acidosis in the general population. A diagnosis of congenital lactic acidosis is made based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests. Blood and cerebrospinal fluid tests can reveal certain findings associated with congenital lactic acidosis such as elevated levels of lactate. An enzyme deficiency may be diagnosed by tests conducted in white blood cells or in skin or muscle cells obtained by biopsy.