

Valinemia is a very rare metabolic disorder. It is characterized by elevated levels of the amino acid valine in the blood and urine caused by a deficiency of the enzyme valine transaminase. This enzyme is needed in the breakdown (metabolism) of valine. Infants with valinemia usually have a lack of appetite, vomit frequently, and fail to thrive. Low muscle tone (hypotonia) and hyperactivity also occur. Valinemia is usually present at birth. Symptoms in the newborn period include protein intolerance, metabolic acidosis, frequent vomiting, failure to thrive, and coma. The condition may become life-threatening. The levels of the amino acid valine in the blood and urine are elevated. Abnormally low muscle tone, excessive drowsiness, and/or hyperactivity can also occur. Valinemia is a rare disorder, occurring once in about 250,000 live births in the United States. It is present in affected infants at birth. Diagnosis depends on the precise laboratory identification and measurement of metabolic products that accumulate in the blood and urine. Sophisticated chromatographic equipment is required to identify these metabolites.