

Succinic semialdehyde dehydrogenase (SSADH) deficiency is a rare inborn error of metabolism that is inherited as an autosomal recessive trait. In individuals with the disorder, deficient activity of the SSADH enzyme disrupts the metabolism of gamma-aminobutyric acid (GABA). GABA is a natural chemical known as a "neurotransmitter" that serves to inhibit the electrical activities of nerve cells (inhibitory neurotransmitter). SSADH deficiency leads to abnormal accumulation of the compound succinic semialdehyde, which is reduced or converted to 4-hydroxybutyric acid, also known as GHB (gamma-hydroxybutyric acid). GHB is a natural compound that has a wide range of effects within the nervous system. The "hallmark" laboratory finding associated with SSADH deficiency is elevated levels of GHB in the urine (i.e., 4-hydroxybutyric or gamma-hydroxybutyric aciduria), the liquid portion of the blood (plasma), and the fluid that flows through the brain and spinal canal (cerebrospinal fluid [CSF]).