

Adenylosuccinate lyase deficiency (ASLD) is a rare, inherited metabolic disorder due to a lack of the enzyme adenylosuccinate lyase (ASL). The defect is characterized by the appearance of two unusual chemicals, succinylaminoimidazole carboxamide riboside (SAICA riboside) and succinyladenosine, in cerebrospinal fluid, in urine and, to a much smaller extent, in plasma. These compounds, which are never found in healthy individuals, are formed from the two natural compounds acted upon by the enzyme. The symptoms and the physical findings associated with ASLD vary greatly from case to case. As a rule, patients with ASLD present with a mix of neurological symptoms that usually will include some of the following: psychomotor retardation, autistic features, epilepsy, axial hypotonia with peripheral hypertonia, muscle wasting, and secondary feeding problems. Although abnormal physical features (dysmorphism) are not common, when they do occur they may include severe growth failure, small head circumference, brachycephaly, flat occiput, prominent metopic suture, intermittent divergent strabismus, small nose with anteverted nostrils, long and smooth philtrum, thin upper lip, and low set ears.