Congenital sucrase-isomaltase deficiency (CSID) is a rare inherited metabolic disorder characterized by the deficiency or absence of the enzymes sucrase and isomaltase. This enzyme complex (sucrase-isomaltase) assists in the breakdown of a certain sugars (i.e., sucrose) and certain products of starch digestion (dextrins). The sucrase-isomaltase enzyme complex is normally found within the tiny, finger-like projections (microvilli or brush border) lining the small intestine. When this enzyme complex is deficient, nutrients based on ingested sucrose and starch cannot be absorbed properly from the gut.

Symptoms of this disorder become evident soon after sucrose or starches, as found in modified milk formulas with sucrose or polycose, are ingested by an affected infant. Breast-fed infants or those on lactose-only formula manifest no symptoms until such time as sucrose (found in fruit juices, solid foods, and/or some medications) is introduced into the diet. Symptoms are variable among affected individuals but usually include watery diarrhea, abdominal swelling (distension) and/or discomfort, among others. Intolerance to starch often disappears within the first few years of life and the symptoms of sucrose intolerance usually improve as the affected child ages. CSID is inherited as an autosomal recessive genetic trait.

CSID is a metabolic disorder that affects males and females in equal numbers and is diagnosed in approximately 0.2 percent of North Americans. This disorder has a higher frequency among Greenland and Canadian Inuit populations, with a reported incidence of approximately 10 percent. In addition, researchers report a higher degree of homozygosity among these more frequently affected groups than among other groups. Also, as a diagnosis may be easily missed, especially in those individuals with mild symptoms, many researchers suspect there may be a higher incidence of this disorder than is actually reported in the medical literature.