

ANMC Congenital Sucrase-Isomaltase Deficiency (CSID) Diagnosis and Treatment Guidelines

Updated 8/19/20

Background Information ¹	Symptoms ¹	Diagnosis ¹ ²
<ul style="list-style-type: none"> It is estimated that 3-10% of Alaska Native people have CSID, but exact prevalence is unknown There is a mutation present in people from Arctic regions that results in a complete lack of enzyme activity Higher prevalence of CSID is seen along the western/northern coasts of the state, especially Yup'ik and Inupiaq people CSID is an autosomal recessive (homozygous) disorder, but heterozygote carriers may also be symptomatic ANTHC has a CSID Program to provide education and support for patients, families, and providers* Click here to access the Pocket Guide to Alaska Native Pediatric Diagnoses 	<ul style="list-style-type: none"> Osmotic diarrhea Abdominal pain & distention Malnutrition Failure to thrive <p style="text-align: center;"><i>Symptoms vary between affected individuals depending on the amount of sucrose and/or starch consumed and other unknown factors</i></p> <p style="text-align: center;"><u>Other Considerations</u></p> <p>Can onset of symptoms be traced back to introduction of table foods? Breast and most formula fed infants are asymptomatic</p> <p>Is there a family history of intolerance or “allergy” to sugar?</p> <p>Are symptoms worse when eating store bought foods versus Alaska Native traditional foods?</p>	<ul style="list-style-type: none"> Genetic screen (blood) through Fulgent Genetics <ul style="list-style-type: none"> Click here for test requisition and here for consent form In the Test Requested box write “SI gene c.273_274delAG p.Gly92Leufs*8” Cost: \$200 2-3 week turnaround for results Consider full sequencing of SI gene if known mutation test is negative and symptoms still indicate possible CSID Notify CSID Program Coordinator with positive results

Nutrition	Infant Formulas	Medication Management ³	Prescribing Sucraid®
<ul style="list-style-type: none"> Avoid and/or limit sucrose (table sugar) and starch The Alaska Native traditional diet is excellent for managing symptoms of CSID since it is naturally low in sucrose and starch Refer patient to a Registered Dietitian <ul style="list-style-type: none"> If no RD available, refer to CSID Program Coordinator CSID education materials for patients/families are available. Contact the CSID Program Coordinator for copies. 	<ul style="list-style-type: none"> Breast milk and standard lactose based formulas, such as Similac® Advance® or Enfamil® Infant, are typically tolerated Most other formulas contain sucrose and/or starch and are generally not tolerated 	<ul style="list-style-type: none"> Sacrosidase (Sucraid®): enzyme replacement for sucrose <ul style="list-style-type: none"> Taken with each meal and snack that contain sucrose Dosage is weight dependent Requires refrigeration Most private insurance covers Sucraid® For Medicaid or IHS recipients, refer patient to ANTHC’s Tribally Sponsored Health Insurance Program (TSHIP). See TSHIP section 	<ul style="list-style-type: none"> If prescribing for patients located in the Anchorage Service Unit click here for instructions If prescribing for patients in the Utqiagvik, Kotzebue, and Nome Service Unit check for Sucraid® availability with the local pharmacy

***For questions or assistance, contact ANTHC’s CSID Outreach and Education Coordinator at 729-3628**

REFERENCES: 1. Marcadier, J., Boland, M., Scott, C., Issa, K., Wu, Z., McIntyre, A., Hegele, R., Geraghty, M. and Lines, M. (2014). Congenital sucrase–isomaltase deficiency: identification of a common Inuit founder mutation. Canadian Medical Association Journal, 187(2), pp.102-107. 2. Treem, W. (2012). Clinical Aspects and Treatment of Congenital Sucrase-Isomaltase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 55, pp.S7-S13. 3. Sucraid for Healthcare Providers. (2018). Sucraid® - Information for Healthcare Providers. [online] Available at: <https://www.sucraid.net/hcp/> [Accessed 10 Sep. 2018].

Tribally Sponsored Health Insurance Program (TSHIP)

- ANTHC partners with THOs to sponsor eligible individuals for health insurance coverage
- Covers the cost of Sucraid®
- 2-6 week period between enrollment and effective coverage
- Does not provide retroactive coverage
- Refer patient to TSHIP by calling 729-2935. A Health Benefits Specialist will work with the patient for eligibility verification and enrollment