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

Updated 4/11/2022

Background Information[1]

- 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
- Click here to access the Pocket Guide to Alaska Native Pediatric Diagnoses

Symptoms[1]

- **Osmotic diarrhea**
- **Abdominal pain & distention**
- **Malnutrition**
- **Failure to thrive**

Symptoms vary between affected individuals depending on the amount of sucrose and/or starch consumed and other unknown factors

Other Considerations

Can onset of symptoms be traced back to introduction of table foods? Breast and most formula fed infants are asymptomatic

Is there a family history of intolerance or "allergy" to sugar?

Are symptoms worse when eating store bought foods versus Alaska Native traditional foods?

Diagnosis[1] [2]

- Genetic screen (blood) through Fulgent Genetics
  - 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
- Cost: $895 for full sequencing of SI gene. Click here for test requisition

Nutrition

- 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
  - If no RD available, refer to CSID Program
- CSID education materials for patients/families are available. Contact the CSID Program for copies.

Infant Formulas

- 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

Medication Management[3]

Sacrosidase (Sucraid®): enzyme replacement for sucrase

- 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

Prescribing Sucraid®

- If prescribing for patients located in the Anchorage, Bethel, Dillingham, and Southeast Service Unit click here for instructions
- If prescribing for patients located in the Utqiagvik, Kotzebue, and Nome Service Unit check for Sucraid® availability with the local pharmacy

For questions or assistance, contact ANTHC's CSID Program at 729-3628

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

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].