Gut Microbiome Survey in Individuals with CHARGE Compared to Sibling Controls

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Presenter Information

Emily Chedrawe is a second year medical student at Dalhousie University in Canada. She was first introduced to CHARGE syndrome 3 years ago under the mentorship of Dr. Kim Blake. At the IWK Health Centre clinics, she has the pleasure of meeting individuals with CHARGE and their families from across the Maritimes, where she learns all about care needs from head to toe for individuals with CHARGE. At Dalhousie, she performs research with both zebrafish and people affected with the Chd7 mutation. In zebrafish, she has studied the expression of genes that are important for the formation of nerve cell that helps regulate breathing. In addition, she has done behavioural studies on how zebrafish with the Chd7 mutation respond to anesthesia. Her clinical research involves studying gastrointestinal complications that arise in individuals with CHARGE syndrome, and how those symptoms might be related to the bacteria found in the gut. With her research team, she began studying the gut microbiome in individuals with CHARGE syndrome in Canada, and they hope to expand this research to people from all over North America.

Presentation Abstract

CHARGE syndrome refers to a cluster of characteristics that present from birth and are related to mutations the CHD7 gene. Although gastrointestinal symptoms are not included in the CHARGE mnemonic, dysfunctions in feeding and digestion represent a serious challenge for affected persons. The cause of lower GI tract symptoms is not entirely known, however research in zebrafish with Chd7 mutations show reduction in foregut innervation by the vagus nerve as well as abnormal enteric nerve development needed for intestinal motility. Environmental factors such as diet and the bacterial environment of the gut also contribute to GI function. In this study, we analyzed the gut microbiome in 8 individuals with CHARGE syndrome from Atlantic Canada compared to their sibling who is unaffected by CHARGE. All participants provided a stool sample, a GI symptom severity scale, and a Block food screener. Bacteria was characterized and quantified through sequencing of 16S RNA, and bioinformatic software inferred their metabolic functions. This information was analyzed in conjunction with a nutritional assessment and GI symptom severity to determine if bacterial dysbiosis is associated with constipation and bloating in individuals with CHARGE.