"Gastrointestinal and feeding difficulties in CHARGE syndrome: A review from head-to-toe"
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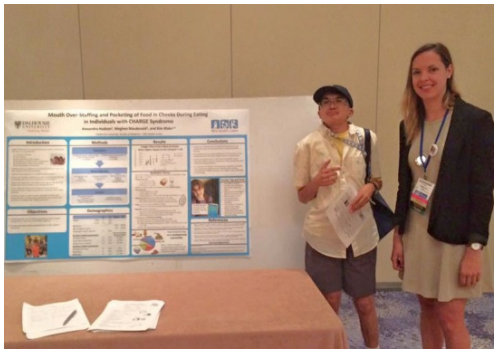
AUTHORS AND THEIR CONNECTION TO THE CHARGE SYNDROME FOUNDATION:

Kim Blake, MD, is a professor of Pediatrics at Dalhousie University Medical School and a General Pediatrician at the IWK Health Center in Halifax, Nova Scotia, Canada, where she runs the multidisciplinary Atlantic Canadian CHARGE Syndrome Clinic. Kim is an international expert in CHARGE syndrome and has been researching gastrointestinal issues and other aspects of CHARGE syndrome (CS) for over 30 years. She has presented at countless CHARGE conferences around the world and has mentored many of the emerging experts on CS. Kim has published extensively on CS and has received multiple clinical research grants from the CHARGE Syndrome Foundation. She started the family support group in the UK and has been involved with the CHARGE Syndrome Foundation from the beginning. Over the years, Kim has received multiple awards and recognition for her work on CHARGE.



Alexandra Hudson, MD, is a recent graduate of Dalhousie University Medical School in Halifax, Nova Scotia, Canada. She has been conducting research in feeding/gastrointestinal dysfunction in CS under the supervision of Dr. Kim Blake for the past three years. Alex was a Sandra Davenport Fellow at the 2015 International CS Conference (see her reflections on that experience **here**), where she also presented a poster on the initial findings of the feeding research. She continued this research after the 2015 conference and presents the findings here.

SUMMARY OF THE PAPER:

***Abstract*:** CHARGE syndrome (CS) is an autosomal dominant genetic condition that is primarily diagnosed based on clinical features, with genetic testing available for confirmation. The CHARGE mnemonic stands for some of the common characteristics: coloboma, heart defects, atresia/stenosis of the choanae, retardation of growth/development, genitourinary anomalies, and ear abnormalities. However, many of the common clinical features are not captured by this mnemonic, including cranial nerve dysfunction, considered by some to be one of the major diagnostic criteria. Over 90% of individuals with CS experience feeding and gastrointestinal dysfunction, which carries great morbidity and mortality. The aim of this review is to examine the nature of gastrointestinal (GI) symptoms and feeding difficulties in CS, focusing on their underlying pathology, associated investigations, and available treatment options. We also provide information on available tools (for parents, clinicians, and researchers) that are important additions to the lifelong healthcare management of every individual

with CS. We review how cranial nerve dysfunction is one of the most important characteristics underlying the pervasive GI and feeding dysfunction, and discuss the need for future research on gut innervation and motility in this genetic disorder.

Additional summary: Nearly every individual with CS will have GI or feeding issues at some point in his/her life. The authors begin by pointing out how nearly all of the clinical features of CS contribute to the GI and feeding issues, including everything from facial clefts and cranial nerve anomalies to lack of sense of smell and compulsive behaviors (Figure 1). They point out that GI and feeding issues contribute significantly to extended hospitalizations, the need for medications and social issues. The authors summarize information from their own research and experience as well as review 71 articles in the medical literature. Table 1 summarizes the clinical features of CS and their potential effects on the GI system. Table 2 summarizes the recognized GI issues in CS and the currently available treatments. Table 3 lists recommendations for assessment and management of feeding and GI issues in CS.

In the text, the authors again review structural, clinical and behavioral features of CS and their potential effects on GI/feeding issues, and go on to discuss how many of these issues interact with one another. These descriptions cover far more than what is simply listed in the Tables. Some issues have an obvious effect on feeding (e.g. tracheo-esophageal fistula) while others are far more subtle but could be equally as important (e.g. poor breathing due to choanal atresia or aspiration). The authors not only describe many scenarios, but suggest techniques that may be helpful. There are gems of useful advice sprinkled throughout this paper.

This article is an excellent example of how every tiny piece of CS is related to every other piece. Rather than considering features in isolation, every aspect must be considered in relation to the whole child. For example, excessive secretions can interfere with not only feeding, but also with speech and even with social situations. Recurrent pneumonias may be the result of aspiration of the excessive secretions. They conclude by stating “GI and feeding difficulties are pervasive and ongoing issues in CHARGE syndrome, carrying great morbidity and mortality” and making recommendations for future research.

WHAT DOES THIS MEAN TO FAMILY/PERSON WITH CHARGE?

This is a paper that will help you understand a lot about you or your child. Not simply the feeding and GI issues (and they are not “simple”), but a lot about how each aspect of CS affects every other aspect of CS. It includes descriptions (what the heck is “late dumping syndrome”?) as well as potentially helpful management - what strategies (or which types of therapists) others have found helpful.

SHOULD I READ IT? SHOULD ONE OF MY DOCTORS READ IT?

Yes, and yes. This is a paper every family should print out, read carefully and refer to often. Although it does contain necessary medical terms and jargon, it is very clearly written and will be understandable to anyone with the medical knowledge quickly attained by any parent of a child with CS. Go through it and highlight the areas that are relevant to your child. Make extra copies to take to doctors. The information in this paper can be helpful to the general pediatrician, gastroenterologist, neurologist, pulmonologist, dietician, speech therapist, and maybe others. Even if they only look at the abstract and the tables, it will be helpful to them. There are references to several additional papers which also would be useful to have (CHARGE Health Checklist, CHARGE Non-Vocal Pain Assessment scale, etc.)

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