1. **Title:** ‘Experiences with feeding and swallowing, as it relates to mouth over-stuffing and pocketing of food in cheeks, during meals in children with CHARGE syndrome’, Alexandra Hudson, path 1, Dr. Kim Blake, Division of General Pediatrics, Department of Pediatrics, I.W.K. Health Centre.

2. **Lay Summary:**
Children with CHARGE syndrome commonly experience feeding issues throughout their lifetime, which may be associated with their congenital structural anomalies, motor impairment, and/or oral sensory impairment. Most of these children have to be on tube feeding at the beginning of their lives, and the transition to oral feeding can be difficult. Parents have reported mouth over-stuffing or pocketing of food in their child’s cheeks while eating. This behavior has been reported to lead to consequences such as choking, and in the most severe case, even death. This study aims to describe the mouth over-stuffing phenomenon by interviewing caregivers of children who have experienced this issue in the past or present. Caregivers will complete a short questionnaire regarding the impact of their child’s feeding and swallowing issues and a questionnaire regarding demographics and the characteristics of their child’s CHARGE syndrome. Following the questionnaire, the caregiver will complete a 45 minute interview with the student researcher, Alexandra Hudson, where they will be asked questions about their child’s eating habits, mouth over-stuffing, and any therapy or solutions they have used for their child’s feeding issues.

3. **Objectives:**
Research Question: What is the feeding and swallowing experience, as it relates to mouth over-stuffing and pocketing of food in cheeks, for a child with CHARGE syndrome and what are the resulting feeding and digestive issues they face throughout their lifetime?

4. **Significance:**
This study will describe a specific feeding issue reported in the CHARGE population that has not been previously well described in the literature. It is important to identify the feeding issue, any associated consequences, and potential therapy and solutions that will reduce the mouth over-stuffing during the eating process of individuals with CHARGE syndrome.

5. **Background:**
CHARGE syndrome is caused by a genetic defect and encompasses a cluster of symptoms: coloboma of the eye, heart defects, atresia of the choanae, retardation of growth and/or development, genital and/or urinary abnormalities, and ear abnormalities/deafness. Children with CHARGE syndrome commonly experience issues with feeding, including swallowing and controlling the amount of food they put in their mouth. These issues may be associated with congenital structural anomalies, motor impairment, and/or oral sensory impairment. In children with CHARGE syndrome, the introduction of functional oral feeding is often delayed and there may be long-term feeding complications as a result. Furthermore, many children with CHARGE
syndrome experience issues with over-stuffing their mouth or pocketing food in their cheeks during feeding, which can lead to aspiration, and in the most severe cases, death\textsuperscript{4,6}. Identifying potential solutions to reduce mouth over-stuffing may help decrease such consequences\textsuperscript{3,5}.

This study will investigate the prevalence and nature of feeding difficulties and swallowing issues in patients with CHARGE syndrome and attempt to illustrate the natural history of the feeding and swallowing issues of patients with CHARGE syndrome through time. This research study will fill a gap in the literature and provide a deeper understanding of the mouth over-stuffing phenomenon during feeding in children with CHARGE syndrome. Parents have expressed that swallowing and feeding issues is an area of CHARGE syndrome that they would like to see further research.

6. Methodologies:
The intended participants for this study are the parents/guardians of individuals with CHARGE syndrome. International participants will be recruited through the CHARGE Syndrome Association (Germany), the CHARGE Syndrome Association of Australasia (New Zealand and Australia). Canadian participants will be recruited through a custodian (outside any patient's circle of care) in Halifax. The members of the local CHARGE syndrome population have previously agreed to being contacted for interest in future CHARGE syndrome related projects. An email with an invitation to participate will be distributed. The 'Invitation to Participate' document will also be placed on the CHARGE Syndrome Facebook page. Interested families will be asked to contact the student researcher, Alexandra Hudson for more information. Participants who are interested will be asked to participate in a phone interview consisting of pre-set questions.

In addition, Dr. Blake and Alexandra Hudson will be travelling to both Chicago for conferences (in 2015) regarding CHARGE syndrome and will be available to discuss this study with families and provide assistance for completing paper versions of the questionnaires and in-person interviews during the conference.

Questionnaires provided to caregivers of patients with CHARGE syndrome will include:
1. Demographic and CHARGE characteristics
2. Feeding/Swallowing Impact on Children's Caregivers

The interview will be asking open-ended questions regarding medications, surgeries, and experience with the following categories.
1. Feeding
2. Mouth over-stuffing and pocketing of food in cheeks during feeding
3. Chewing and swallowing during feeding
4. Choking Incidents
5. Speech-language pathologist and physiotherapist experiences
6. Neurological

Each questionnaire is brief and we predict that the total time to complete the questionnaires is 30 minutes. We predict that the total time to complete the additional
open-ended questions is approximately 45 minutes depending on the level of detail parents provide.

Many of the parents of children with CHARGE syndrome are very familiar with the medical terminology regarding CHARGE syndrome and characteristic features. However, if a parent/guardian is unfamiliar with any of the terminology used in the study they are encouraged to contact the student researcher, Alexandra Hudson.

Some members of the CHARGE syndrome community have identified themselves as being very interested in participating in and staying active with CHARGE related research. A select number of these individuals will be asked to pilot the study. They will asked to provide feedback on the length of the questionnaires, overall quality of the questions, and terminology. Additional feedback on the questionnaire will be sought from speech language pathologists and occupational therapists. This feedback will be incorporated (if appropriate) into the study before we disperse the questionnaires to the larger population.

The quantitative data from the feeding/swallowing impact survey will be analyzed using basic statistics, including descriptive statistics (means, standard deviations) and frequencies. The qualitative data will give us a richer understanding of the challenges individuals with CHARGE syndrome face on a general basis over time in regards to eating and swallowing issues. The narrative responses will be reviewed in the context of the individual patient and on a macroscopic level of CHARGE syndrome for recurrent themes. The information gathered from the CHARGE characteristic portion of the demographic questionnaire will help us to potentially identify any genotype/phenotype correlations.

7. **Roles of student:**
   - Find a suitable questionnaire to eating and swallowing difficulties in a pediatric population
   - Develop the interview questions based on current literature and feedback from pediatricians, speech language pathologists, and occupational therapists
   - Write and submit documents for I.W.K. REB
   - Recruit participants
   - Perform and transcribe interviews with participants
   - Analyze questionnaire responses and interview answers
   - Maintain an up-to-date analysis of questionnaires and interview answers
   - Submit an abstract to present a poster at the 2015 CHARGE conference in Chicago
   - Prepare a poster for the 2015 CHARGE conference in Chicago
   - Prepare a manuscript to submit to a journal

8. **Research Activities Timeline**
   March 2015
      - Complete REB approval
   April-May 2015
- Pilot the study with a few participants
- Ask for feedback on questionnaire from clinicians.

June-August 2015
- Recruit participants
- Complete interviews
- Analyze questionnaire responses and interview transcripts
- Attend and present a poster at the CHARGE conference in Chicago July 28-Aug 2 2015
- Begin writing manuscript

9. **Ethical Approval:**
   I.W.K. REB approval #1018719
   March 9th 2015

10. **References:**


