PROTOCOL

Research Title:

Medical Issues in adults with CHARGE Syndrome.

Principal Investigator:

Liandrah Gapare, PhD.
Medical Student,
Dalhousie University

Co-Investigator:
Laith Alhadeed, MSc.
Medical Student
Dalhousie University

Supervising Investigator:

Kim Blake, MD
General Pediatrics, Department of Pediatrics
IWK Health Center and Dalhousie University

Translator / Community Partner: Claudia Junghans
1. president CHARGE Syndrom e.V. / Germany

1. Background and Rationale

CHARGE syndrome is a complex genetic condition associated with multisystem involvement beginning in infancy. While pediatric manifestations are well characterized, there is limited structured data regarding how specific medical conditions evolve, stabilize, or deteriorate in adulthood. Adults with CHARGE syndrome are living longer due to advances in pediatric medical care; however, limited information exists regarding the long-term health concerns, healthcare experiences, and monitoring needs of adults with CHARGE syndrome. Most published literature focuses on children, leaving a significant gap in knowledge regarding adult health outcomes. This study seeks to characterize the spectrum of medical conditions affecting adults with CHARGE syndrome and identify unmet healthcare needs to inform the development of an adult care checklist.

2. Research Question

What are the current healthcare experiences, medical concerns, and perceived gaps in care among adults living with CHARGE syndrome?

3. Study Objectives

Primary Objective:

To characterize the spectrum and frequency of medical conditions affecting adults with CHARGE syndrome to inform development of a structured, prioritized clinical checklist to guide adult care.

Secondary Objectives:

1. To identify perceived gaps in adult healthcare services.
2. To identify areas requiring routine adult surveillance and monitoring.
3. To explore patterns between adult medical issues and CHARGE syndrome features.
4. To inform the development of a practical clinical checklist for adult care.

4. Study Design

This is an investigator-initiated, international, cross-sectional online survey study. Approximately 100 participants will be recruited, including approximately 5 participants from Nova Scotia and approximately 95 participants from outside Nova Scotia. Participants will be recruited through CHARGE syndrome organizations, conferences, and

community networks. Adults (18 years or older) with CHARGE syndrome who are able to provide informed consent will complete the survey themselves. For adults who are unable to provide informed consent independently, the survey may be completed by a parent or legally authorized substitute decision-maker following provision of informed consent. The study is not a multicentre research project. Data will be collected anonymously through a secure online questionnaire. No interventions, clinical testing, or changes to participants' medical care will occur.

5. Study Population

The study population will consist of approximately 100 adults (18 years or older) with a clinical or genetic diagnosis of CHARGE syndrome. Approximately 5 participants are expected to be recruited locally in Nova Scotia, with the remaining participants recruited internationally through CHARGE syndrome organizations and community networks. Participants may complete the survey themselves if they have the capacity to provide informed consent. If an adult lacks decision-making capacity, the survey may be completed by a parent or legally authorized substitute decision-maker on their behalf..

6. Recruitment

Participants will be recruited in Nova Scotia and internationally through CHARGE syndrome organizations, international CHARGE syndrome conferences, invitation emails, and online CHARGE syndrome community networks. Recruitment is anticipated to include participants from Canada, the United States, Germany, Australia, New Zealand, and other countries where eligible individuals become aware of the study through these recruitment methods. Participants are not excluded based on country of residence. No hospital records, clinic lists, registries, or medical records will be used to identify or recruit participants. Participation is entirely voluntary.

The Supervising PI will also send out an email to participate to patients with CHARGE syndrome who she is in touch with that are within her circle of care. The letter will come from her but there will be contact information for the PI and a QR code they can go to for the consent.

7. Consent Process

Informed consent will primarily be obtained electronically. Potential participants will review the study information and consent form before accessing the survey. The

Principal Investigator will be available by email, telephone, or virtual meeting to answer participant questions. Formal consent discussions will occur only when requested by participants or when additional support is required to facilitate informed consent.

For participants who lack capacity to provide informed consent independently, consent will be obtained from a legally authorized substitute decision-maker. A separate assent form will not be used. For adults who lack full capacity to provide informed consent, consent will be obtained from a legally authorized substitute decision-maker. Whenever possible, the participant will be involved in the discussion and their willingness to participate will be confirmed verbally or behaviorally before participation proceeds.

A separate written assent form will not be used. When appropriate, the participant's willingness to participate will be confirmed before the survey is completed, and any indication that the participant does not wish to participate will be respected.

8. Risks and Safeguards

This study involves minimal risk. Participants may experience mild emotional discomfort when reflecting on their health experiences or answering questions about medical conditions. Participants may skip any question they do not wish to answer, pause the survey, or withdraw from the survey at any time before submitting their responses. Because no directly identifying information will be collected, the risk of a breach of confidentiality is low. Results will be reported only in aggregate form.

9. Benefits

There is no direct medical benefit to participants. However, findings will inform development of a prioritized clinical checklist to improve structured adult care for individuals with CHARGE syndrome.

10. Privacy and Confidentiality

No directly identifying personal information or personal health information will be collected as part of this study. Participants will complete an anonymous online questionnaire that includes demographic information, health experiences, and questions relating to CHARGE syndrome.

Survey responses will be collected through a secure online survey platform and stored on password-protected institutional systems accessible only to authorized members of the research team. Results will be analyzed and reported in aggregate form. Every reasonable

effort will be made to protect participant confidentiality. Although CHARGE syndrome is a rare condition and there is a small possibility of indirect identification through combinations of demographic information, no names, contact information, medical record numbers, or other direct identifiers will be collected.

Study data will be retained for 5 years following study completion and will then be securely destroyed in accordance with institutional policies.

11. Data Analysis Plan

Descriptive statistics will be used to calculate frequencies and proportions of medical conditions across organ systems. Findings will inform checklist development and prioritization of adult care needs.

12. Knowledge Translation

Study findings will be presented at scientific meetings, CHARGE syndrome conferences, and published in peer-reviewed journals. A summary of the findings may also be shared with the CHARGE syndrome community. All results will be reported in aggregate form, and no participant will be identified in any publication, presentation, or report.

13. Funding

No funding.

14. Ethical Considerations

The study involves a potentially vulnerable population. Safeguards include accessible study materials, electronic informed consent, availability of the research team to answer participant questions, the use of legally authorized substitute decision-makers when appropriate, voluntary participation, anonymous data collection, secure data storage, and reporting of findings only in aggregate form.