



Patient Information Letter

Study Title: Incidence and Prevalence of CHARGE syndrome in Canada

Investigator: Kim Blake, MB, MRCP, FRCP(C), Paediatrician, Department of Pediatrics, IWK Health Centre, Halifax, Nova Scotia

Sponsor(s): N/A

Introduction

As you may know, CHARGE syndrome is currently diagnosed by observation of an individual's clinical features (e.g., hearing problems, vision problems and/or heart/breathing problems, etc.). It is a rare but highly varied condition. That is, affected individuals have a range of clinical features that can be mild to severe. Recently, a group of researchers identified defects in a human gene that they believe can cause CHARGE syndrome (Vissers *et al.*, 2004, *Nature Genetics* 36:955). One area of research we are interested in involves testing individuals with CHARGE syndrome for the presence of these gene defects. Currently, this additional proposed research study is undergoing the ethical approval process (Project #3164) with the IWK Research Ethics Board.

We would like to invite you, an individual with CHARGE syndrome, to participate in our research. It is important that you understand the purpose of this study, how it may affect you, the risks and benefits of participating, and what you will be asked to do before you decide if you want to participate. This **Patient Information Letter** is designed to help you make this decision. Participating in this study is entirely voluntary (it is your choice), so you do not have to take part. Questions are encouraged. If you have any questions that this letter does not answer, either Dr. Kim Blake (1-902-470-6499) or a member of her research team, will be happy to give you more information.

If you agree to participate in this study, we would ask you to complete the **Patient Contact Form** and to give the **CHARGE Syndrome Reporting Form** to your doctor or other health professional to be completed. The information on the **CHARGE Syndrome Reporting Form** will provide us with details about your clinical features of CHARGE syndrome. By supplying your contact information on the **Patient Contact Form**, we will be able to contact you directly to discuss and describe future research into CHARGE syndrome.

Purpose of the Study

One purpose of this study is to collect information about the incidence of CHARGE syndrome in Canada and about the clinical features of individuals with CHARGE syndrome. An additional purpose of this study is to identify and make contact with those individuals with CHARGE syndrome who agree to be contacted, in order to discuss and describe future research studies.

Study Design

Dr. Kim Blake is well known to many Canadian families affected by CHARGE syndrome and their health professionals because of her research and her involvement in the support group, CHARGE Syndrome Canada. As such, it is anticipated that individuals with CHARGE syndrome will contact Dr. Blake directly or through their own doctor or health professional about becoming involved in future research studies.

After reading the **Patient Information Letter**, you may agree to participate by completing the **Patient Contact Form**. You will then kindly ask your doctor or health professional to complete the **CHARGE Syndrome Reporting Form**. All forms can be returned to Dr. Kim Blake at the IWK Health Centre in Halifax, Nova Scotia. The IWK Health Centre is the only centre involved in this research project.

If you agree to be contacted by Dr. Kim Blake or a member of her research team, this contact (usually by phone) would take approximately 15 minutes to discuss and describe future research.

Potential Harms

There are potential harms that may arise from your participation in this study. You may be inconvenienced when contacted by Dr. Blake or a member of her research team. You may become distressed when you are contacted by Dr. Blake or a member of her research team to describe and discuss future research into CHARGE syndrome. As well, there may be potential harms arising from your participation that are unforeseen at this time.

Potential Benefits

Although there will be no direct benefit to you from your participation, it is hoped that this study will add to the current knowledge of CHARGE syndrome. Adding to our understanding of CHARGE syndrome may indirectly benefit affected individuals and families in the future. This study may also prompt new research into other aspects of CHARGE syndrome.

Alternatives to the Study

Before you decide to participate in this study, you should know that you are not obligated to take part. Participation in this study is entirely your choice. Your decision whether you take part or not will have no effect on your present or future care at any hospital. You may decide that it is best for you not to participate.

Withdrawal from Participation

You may withdraw from participation in this study at any point. For example, you may initially supply your contact information on the **Patient Contact Form** and offer to be contacted, but prior to being contacted by us, you may decide you do not want to be contacted. Even if you supply your contact information on the **Patient Contact Form** and are contacted by Dr. Kim Blake or a member of her research team, you may decide not to participate or may later decide you do not want to be contacted further by us.

Withdrawal from this study would mean that any personal data you have given us during the course of this study (i.e., **Patient Contact Form, CHARGE Syndrome Reporting Form** and other information) would be removed from our files and destroyed; your data would not be used in any analyses. Withdrawing from this study will not affect your present or future health care in any way.

The confidential data supplied by to us for this study by participants and doctors or other health professionals will be kept at the IWK Health Centre for 25 years under the supervision of Dr. Kim Blake.

Costs and Reimbursement

If you agree to be contacted by Dr. Kim Blake or a member of her research team, we have some limited funds to pay your postage or telephone long distance calls. We unfortunately cannot pay costs for your time or inconvenience.

Confidentiality

Any information about you will be kept private. There will be no names used when the study results are published. If the results of any study are published in the medical literature, they will not contain any information that could identify you as an individual.

The research team will have access to a file on your clinical details of CHARGE syndrome. In addition, the research records may be shown to the Research Office of the IWK Health Centre if they review the ethical aspects of our research. Study records will be stored in a locked area and will be kept for a minimum of 25 years under the supervision of Dr. Kim Blake, as required by the IWK Health Centre Research Ethics Board.

Research Rights

By supplying your contact information on the attached **Patient Contact Form**, you have shown that you have understood this information about the proposed research study. Agreeing to be contacted by Dr. Kim Blake or a member of her research team does not mean that you give up your rights.

By supplying your contact information on the attached **Patient Contact Form**, you are not giving up any of your rights, and you are not changing the responsibilities of the investigators, or of other people involved.

If you have any questions at any time during or after the study about these rights or about research in general, and you would like an independent opinion, you may contact the Research Office at the IWK Health Centre at (902) 470-8765, Monday to Friday between 9 a.m. and 5 p.m. (Atlantic Time).

Contact Person

If you have any questions or concerns following your participation in this study, you may contact Dr. Kim Blake (paediatrician) at (902) 470-6499.



Patient Contact Form
CHARGE research study group

I understand that Dr. Kim Blake is working with a research group involved in learning more about CHARGE syndrome.

I agree to be contacted by Dr. Blake and/or her research team members to learn more about specific research projects involving CHARGE syndrome. I understand that my participation is entirely voluntary and I can withdraw at any time.

To allow Dr. Blake and/or her research team to contact me, I have provided the following information which I will return to Dr. Blake via fax at (902) 470-7216, e-mail at kblake@dal.ca or mail to the following address:

Dr. Kim Blake
IWK Health Centre
5850/5980 University Avenue, P.O. Box 9700
Halifax, Nova Scotia B3K 6R8

Name of individual with CHARGE syndrome: _____

DOB of individual with CHARGE syndrome: _____

Contact phone number(s): _____

Contact e-mail (if applicable): _____

I understand that I can use the information listed below to contact either Dr. Blake or the CHARGE parent support group without any obligation on my part.

Dr. Kim Blake
Tel.: 902-470-6499 Fax: 902-470-7216
E-mail: kblake@dal.ca

CHARGE Parent Support Group and CHARGE Syndrome Canada
Community Mail Box, Suite 28
20 Haven Ave
Devon, Alberta
T9G 2B9
Phone: (780) 987-4649
E-mail **info@chargesyndrome.ca**

Enclosed is a data collection form (CHARGE Syndrome Reporting Form) for your doctor(s) to complete.