What can my family do to help discover the causes of CM I?

Research aimed at discovering the causes of CM I will only be successful because of the participation of families with individuals who are diagnosed with CM I.

Your family may be able to participate in this research. Please contact us to obtain more information about participating in the study.

Does my family qualify for the study?

A family is eligible for the study if at least two individuals related by blood have been diagnosed with CM I (confirmed by MRI) and would both like to participate. At the current time we are not able to enroll families in which the only diagnosed members are a parent and a child.

If your family does not qualify but you would still like to contribute to the research, financial donations are accepted and are extremely appreciated.

How can my family enroll in the study or get more information?

The study coordinator can provide you or your clinician with more information about the CM I study and answer any questions you may have.

Please contact:

1-877-825-1694 (toll-free)

Email: chiari@chg.duhs.duke.edu

or

CM I Study Coordinator
Duke University Medical Center
Box 3445
Durham, NC 27710

For more information about CM I and the CM I study, check the Duke Center for Human Genetics website: http://www.chg.duke.edu.

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DUKE center for HUMAN GENETICS

The Genetics of Chiari Type I Malformation (CM I) with or without Syringomyelia

A study aimed at discovering the causes of CM I

INFORMATION FOR FAMILIES

Duke Center for Human Genetics
Duke University Medical Center
IRB# Pro00011231

Why should my family consider participating in this research?

Families often wonder what caused a member to have Chiari Type I Malformation (CM I) with or without Syringomyelia. By participating in this study, families can contribute to research aimed at identifying the causes of CM I. The initial study goal is to find the genetic (inherited) factors related to CM I. The long-term goal is to find out how these genetic factors cause or contribute to CM I, with the hope that this knowledge will lead to improved diagnosis and more effective treatments.

Several lines of evidence point to a genetic contribution to CM I. This evidence includes families with multiple individuals all diagnosed with CM I. Observations of identical twins (genetically the same) have shown that if one twin is diagnosed with CM I, the other twin often also has CM I. Finally, CM I is known to co-occur with a number of different genetic syndromes.

Because the causes of CM I are likely to be complex, the data from many families will need to be compared to uncover the specific genes involved. Once blood samples are received, DNA (the genetic material) is removed and stored. This will allow us to compare the DNA sequence from those individuals who have CM I with those individuals who do not have CM I.

What will my family be asked to do?

- Complete a telephone interview and online questionnaire to collect detailed family, medical, and environmental risk factor history.
- Allow our research staff to review medical records and MRIs.
- Provide blood samples from the family members diagnosed with CM I and other family members (such as siblings and parents), regardless of whether or not they have CM I.

Facts about participation

- Participation is voluntary.
- There is no cost to participate.
- Travel to Duke is not required.
- Participants are reimbursed for blood drawing fees.
- All information collected is confidential.
- Participation will not affect your health care.

Will my family receive genetic results?

Samples and information given to the study are for research purposes only. Individual or family results are not available. Each participant will receive information on the overall study results and the study's progress through periodic newsletters.

The CM I Research Team

The CM I research team combines expertise in genetics, neurosurgery and neuroradiology at Duke University Medical Center (DUMC). This experienced team of researchers and clinicians includes:

- Allison Ashley-Koch, Ph.D. and Simon Gregory, Ph.D., Principal Investigators
- David Enterline, M.D., Neuradiologist
- Herbert Fuchs, M.D., Pediatric Neurosurgeon
- Study Coordinator: This is the person who will work with your family through the enrollment process and answer any questions you might have about the study.