ABOUT THE RESEARCH TEAM

Dilys M. Parry, Ph.D. is a board certified Ph.D. medical geneticist. She has been involved in research on familial and hereditary cancers for 27 years. She began studying chordoma families in 1996 and then initiated this study.

Mary Lou McMaster, M.D. is a board certified internist and medical oncologist. She has specialty training in clinical genetics as well. Her main interest is studying cancer-prone families.

Deborah Zametkin, R.N., M.S.N. is a research nurse who has been working with patients with familial and hereditary cancers for 12 years. She has been working with chordoma families since 1996.

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GENETIC EPIDEMIOLOGY BRANCH

DIVISION OF CANCER EPIDEMIOLOGY AND GENETICS

FAMILIAL CHORDOMA

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
National Institutes of Health • **National Cancer Institute**

PURPOSE

The National Cancer Institute (NCI) is studying families with multiple cases of chordoma, a rare bone cancer.

If chordoma runs in your family, NCI invites you and your family members to consider joining the study, to help us learn more about this tumor. The goals of the study are to:

- Find the gene or genes that cause this type of cancer to occur in families;
- Describe the clinical features of familial chordoma;
- Determine whether families prone to chordoma are at increased risk of other types of cancer; and
- Develop better health care choices for families at risk for chordoma.

WHAT IS CHORDOMA?

Chordoma is a tumor that develops from cells found in the base of the skull, the vertebrae, the sacrum or the coccyx (tailbone). These cells are left over from a structure called the "notochord" that is present before birth but usually disappears entirely shortly after birth.

Chordoma is diagnosed most often in people in their 50s but it can occur much earlier or later. It is more common in males than in females. For unknown reasons, it is rare in African Americans. Families in which two or more people have chordoma are very uncommon.

WHO IS ELIGIBLE?

To participate, a family must have one of the following:

- Two or more blood relatives with a history of chordoma, OR
- One blood relative with a history of chordoma and one or more blood relatives with a history of a brain tumor as a child or young adult.

Eligible relatives include people with chordoma, and their parents, siblings and children. Other blood relatives may be invited to participate if they are genetically linked or have another form of cancer.

The spouse of a person with a history of chordoma who is enrolled in the study may also be invited to participate if their children are enrolled in the study.

PARTICIPATING IN THE STUDY

Those who join the study will be asked to:

- Provide a blood sample or cells rinsed from the inside of the mouth for genetic studies which may help us find the gene or genes that cause familial chordoma;
- Provide personal medical and family history information; and
- Give us permission to obtain past medical records for chordoma, other cancers and related illnesses, and a small piece of tumor stored after any cancer surgery.

Some participants will be invited to visit the Clinical Center of the National Institutes of Health (NIH) at no cost to them to participate in detailed clinical, genetic and laboratory studies aimed at finding the gene or genes that cause chordoma in family members.

Treatment is not part of this study. However we will discuss treatment options with participants and provide assistance in establishing care with appropriate physicians as needed. Participants will remain under the care of their primary doctors while taking part in the study.

CONTINUING CONTACT

Participants and family members will be asked to complete a yearly follow-up form to update medical information.

Participants and family members will be able to ask questions and stay in touch with the research team.

The overall findings of the study will be provided to participants when analysis of information from the study is completed.

OTHER DETAILS

Confidentiality. NCI is required by Federal law not to reveal any information that is collected from study participants to anyone other than persons directly involved with the study. No personal identifying information will be released or published.

Costs. All study-related medical expenses and travel costs to the NIH Clinical Center for participants will be paid by NCI.