Referral of patients with pleuropulmonary blastoma (PPB) and PPB-associated tumors (cystic nephroma, Sertoli-Leydig cell tumors and other sex cord-stromal tumors)

November 13, 2015

Dear Colleague,

The National Cancer Institute’s Clinical Genetics Branch is conducting a study investigating pleuropulmonary blastoma (PPB) and DICER1-associated tumors, including cystic nephroma, Sertoli-Leydig cell tumor and other sex cord-stromal tumors. This study seeks to characterize PPB and other DICER1-associated tumors and to develop evidence-based management guidelines for cancer prevention and risk-reduction strategies for patients and their family members. I am writing to you to request your assistance in recruiting patients and their families to this important study.

As part of the ongoing study activity, all affected individuals and their family members are asked to complete family history and personal medical history questionnaires. Participants may be asked to provide a sample of blood, saliva or other tissue for clinical and research studies. Some study participants may also be evaluated at the National Institutes of Health (NIH) Clinical Center, with the costs of this evaluation covered by the NIH.

Eligibility criteria for inclusion into the study are as follows:

1. Patients with known mutations in the DICER1 gene,
2. Patients with pleuropulmonary blastoma,
3. Patients with other DICER1-associated tumors, including cystic nephroma, Sertoli-Leydig cell tumor and other sex cord-stromal tumors, embryonal rhabdomyosarcoma, nasal chondromesenchymal hamartoma, ocular medulloepithelioma, Wilms tumor and pineoblastoma, and
4. Relatives of the patients listed above.

This study does not involve treatment modalities, and any ongoing or previous therapeutic treatments do not exclude patients from the study. The study is open to participants of all ages, however, participants or their guardian/parent/proxy must be able to understand and be willing to sign a written informed consent document.
Individuals and family members with any of the diagnoses listed above are encouraged to call the National Cancer Institute at 1-800-518-8474 and speak with our family studies referral nurse. They may also visit the study website at http://ppb.cancer.gov or send an email to the study team at nci.ppb@westat.com to find out more details about the protocol. If you have questions about the study you are welcome to call me at 240-276-7238 or email me at drstewart@mail.nih.gov.

The study pamphlet is attached; you are welcome to distribute it to appropriate patients. Thank you in advance for your assistance.

Sincerely,

Douglas R. Stewart, MD
Investigator
LONGER-TERM COMMITMENT: STAYING IN TOUCH WITH OUR STUDY TEAM

Participants will be asked to complete a periodic follow-up questionnaire in order to update medical and cancer information. We encourage participants to stay in touch with the study team and to share any concerns they might have related to the study, or their diagnosis.

OTHER DETAILS

Confidentiality

Patient privacy is our top priority. NCI will not disclose any information that is collected from study participants to anyone other than persons directly involved in 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 are paid for by the NCI.

Treatment

Although treatment is not a part of this study, treatment options will be discussed with participants and their physician (at the participant’s request). The study will also provide help in establishing care with the appropriate physicians, as needed. Study participants should remain under the care of their regular doctor while participating in the study.

ABOUT THE RESEARCH TEAM

Douglas Stewart, MD (NCI Principal Investigator) is an internist and medical geneticist with a special interest in familial cancer syndromes.

Leslie Doros, MD (Lead Oncologist) is a pediatric oncologist who specializes in the treatment of rare tumors and directs the cancer genetics clinic at Children’s National Medical Center.

Drs. Stewart and Doros also work closely with Ashley Hill, MD, at Children’s National Medical Center, Yoav Messinger, MD, and Kris Ann Schultz, MD, at the International Pleuropulmonary Blastoma Registry, and Louis Dehner, MD, at Washington University in St. Louis.

HOW CAN I JOIN THE STUDY OR LEARN MORE ABOUT IT?

Phone: 1-800-518-8474 to speak with the referral nurse

Email: NCI.PPB@westat.com

Website: Read the study description on the web at http://ppb.cancer.gov

June 2015
WHO IS ELIGIBLE?

People who have been diagnosed with a PPB or a PPB-related tumor (or both), as well as their close relatives, are invited to join the study. We welcome people who are newly diagnosed, as well as surviving relatives from families whose affected loved one has passed away.

PPB-related tumors and conditions include:

- Cystic nephroma
- Sertoli-Leydig cell and other sex-cord stromal tumors
- Familial multi-nodular goiter
- Embryonal rhabdomyosarcoma
- Nasal chondromesenchymal hamartoma
- Ocular medulloepithelioma
- Wilms tumor
- Pineoblastoma

WHAT IS INVOLVED IN PARTICIPATING IN THIS STUDY?

Those who join the study will be asked to:

- Complete questionnaires about the family’s overall medical history. Individuals affected with PPB and/or related conditions will be asked to provide more detailed individual medical information.
- Provide permission for the study team to obtain medical records and possibly a tumor sample.
- Give a blood or saliva specimen for genetic testing, if needed.

Participants will receive:

- Education and counseling on what is known about changes in the DICER1 gene and cancer risk, and an opportunity to learn their genetic testing results, if desired.

SOME FAMILIES WILL BE INVITED TO COME TO NIH FOR FURTHER EVALUATION

The study team may invite some individuals and family members to come to the NIH Clinical Center (Bethesda, MD) for a three-day detailed evaluation. The evaluation is free and travel expenses (transportation, meals, lodging) are paid for by the study. The visit to the NIH includes:

- Detailed evaluation of personal medical history along with family history of cancer
- Thorough physical examination
- Chest CT scan and other imaging tests, as necessary, based on the individual participant and protocol
- Consultations with other medical specialists, as appropriate
- Review of findings from clinical tests and cancer screening evaluations, along with a summary report of the visit