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PXE International was founded in 1995 to promote research and support individuals affected by pseudoxanthoma elasticum (PXE). We work on behalf of individuals and their families to improve quality of life through advancing research, educating clinicians and supporting individuals. We are the prime force in conducting basic and clinical research and providing financial support for applied translational research, product development, and treatment development for PXE. Two important, ongoing PXE International research projects are identifying new mutations that cause PXE and understanding if there is a connection between mutations and symptoms that an individual experiences. You can contribute to these research efforts by sharing with us the signs of PXE that you experience, and by releasing the results of your PXE genetic test to us.

To participate in this research, please complete the PXE International Clinical Data Form (on the next page) with your doctor to report the PXE symptoms you have. Read the consent statements at the bottom of the Clinical Data Form, and ask any questions you have about the research and the consent. If you consent to both statements, PXE International will receive the GeneDx Mutation Analysis Laboratory Report and the Clinical Data Form with your identification. We will enter your information into the PXE International Registry and BioBank, a secure and private repository of biological and clinical data of people with PXE. Registration is free, and you will receive our monthly eNewsletter, email announcements, doctor referrals, and connections with the greater PXE community.

If you agree to release your Mutation Analysis Laboratory Report and Clinical Data Form to us, please complete these easy steps:

1. Fill out the next page, including the bottom portion that contains the consent.
2. Photocopy the Clinical Data Form or scan and print it.
3. Give one physical copy to your doctor so s/he can include it with the genetic testing order forms s/he sends to GeneDx.
4. Send the Clinical Data Form to PXE International either via email (amoore@pxe.org) or mail to the address found at the top of this letter.

If you have any questions, please contact us at any time.

Sincerely,

A handwritten signature in black ink, appearing to read 'Sharon Terry'.

Sharon Terry, CEO

Fill out this form with your doctor and return one copy with the test order to GeneDx and one copy to PXE International.

Clinical Data Form for Genetic Testing Pseudoxanthoma Elasticum (PXE)

Patient name: _____ Today's date: ____/____/____
month day year

Submitting physician: _____

Physician contact information: _____

Date of birth: ____/____/____
month day year

Sex: [] Male [] Female

Weight: lbs _____ (or kg: _____)

Height: ft & in: _____ (or cm: _____)

Ethnicity: [] Caucasian [] Black/African American [] American Indian [] Asian
[] Hispanic [] Hawaiian/Pacific Islander [] Mixed [] Other

SKIN

Normal skin: [] Yes [] No Skin biopsy results (select one):
Bumps/papules: [] Yes [] No [] Positive [] Negative
Plaques/coalesced papules: [] Yes [] No [] Inconclusive [] Not performed
Lax/loose/folded: [] Yes [] No

EYES

Peau d'orange: [] Yes [] No Angioid streaks: [] Yes [] No
Retinal bleeding/scarring: [] Yes [] No Vision loss: [] Yes [] No

CARDIOVASCULAR

Weak or absent pulses: [] Yes [] No Chest pain: [] Yes [] No
Intermittent claudication: [] Yes [] No Abnormal EKG: [] Yes [] No
Vascular surgery: [] Yes [] No Abnormal stress test: [] Yes [] No
Heart attack: [] Yes [] No

GASTROINTESTINAL

Gastrointestinal bleeding: [] Yes [] No
Gastrointestinal bleeding diagnosed as related to PXE: [] Yes [] No

FAMILY HISTORY

Affected sibling: [] Yes [] No How diagnosed: _____
Affected parent: [] Yes [] No How diagnosed: _____
Affected grandparent: [] Yes [] No How diagnosed: _____
Affected cousin: [] Yes [] No How diagnosed: _____
Other affected relative: _____ How diagnosed: _____
Other family information: _____

CONSENT

Please read each statement below. If you agree, check the box and initial each statement.

- [] I consent to release my Clinical Data Form to PXE International. _____ (Initial)
[] I consent to release my Mutation Analysis Laboratory Report to PXE International. _____ (Initial)

Testing recommendations: All individuals with characteristic yellow papules and/or lax redundant skin in the flexural areas (neck, axilla, antecubital or popliteal fossa, groin). All individuals with retinal angioid streaks. Individuals with siblings with confirmed PXE.