

NY

INFORMED CONSENT FOR GENETIC TESTING: SHOX-DNA-DX TEST

Physician: Please ask your patient to read and complete this consent form for genetic testing. Esoterix Endocrinology will perform genetic testing only when this form has been completed.

I have counseled Mr/Ms _____ regarding genetic testing and have obtained his/her informed content for each of the genetic tests requested.

Signature of Physician	Date
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PATIENT INFORMATION REGARDING THE SHOX-DNA-DX TEST

Description of the disease

Short stature is defined as being below the 5th percentile for height when compared to your peer group.

Description of the test

DNA is purified from your blood sample. Certain SHOX-specific DNA is amplified and examined in a way that automatically identifies "single or multiple DNA components or DNA sequences" that contain unexpected changes. If changes are seen they are then verified by examining that DNA in a component by component manner using a technique known as DNA sequencing.

Principle of the test

The SHOX-DNA-DX test evaluates DNA taken from your blood sample to determine whether your SHOX "DNA" has unexpected changes. These unexpected changes may include polymorphisms [DNA changes that are not known to affect stature] and various mutations that cause short stature as determined by studies published in peer-reviewed medical journals.

Meaning of a positive test result

A positive SHOX-DNA-DX test identifies an unexpected change or changes in your DNA that has been or have been proved by published medical studies to be associated with or cause short stature. Such a result usually ends the diagnostic evaluation of this disorder.

Meaning of a negative test result

A negative SHOX-DNA-DX test did not identify any unexpected change in your DNA that has been proved by published medical studies to cause short stature. In most cases such a negative test will have shown that your DNA is exactly as expected within the tested regions. In some cases an unexpected change has been found but has not been previously proved by published medical studies to cause short stature. Some of these changes may be a harmless change in SHOX DNA or may later be proved as causes of short stature.

Confidentiality and distribution of your test result

SHOX-DNA-DX test results are confidential. Esoterix Endocrinology will release SHOX-DNA-DX test results only to my physician.

Genetic counseling, further testing, additional physician consults

As a patient you may seek the advice of a professional genetic counselor prior to signing this form. If my test results are positive I may seek advice, including the request for more testing, from my physician and or a genetic counselor to learn how my test result relates to short-stature. I may contact Esoterix Endocrinology at (800) 444-9111 for further information on the SHOX-DNA-DX test

Specimen retention

Esoterix Endocrinology may retain specimens for up to 1 year for the purpose of repeat testing, research, or quality control.

Patient Signature

I have received counseling from my physician regarding the SHOX-DNA-DX test and have read all the information provided above.

Signature of Patient	Date
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