They have found the Gene

by

Dr. Angus Clarke, Senior Lecturer in Medical Genetics
University of Wales College of Medicine, Cardiff

The August 1996 issue of the journal Nature Genetics contained a report on the isolation of the gene for X-linked hypohidrotic ectodermal dysplasia (XHED). It is required for the proper formation of hair, teeth, sweat glands and other ectodermal structures. Every X chromosome carries a copy of this gene, men having one copy and women having two. If the copy of the gene present in a male has changed in some way, so that it fails to function properly, then he will be affected by XHED. This condition is much the commonest type of ectodermal dysplasia, and so the discovery of the gene is a major step forward in understanding the whole group of ED conditions.

The team who reported this discovery is an international group of scientists and clinicians who have been working on XHED for some years. The role played by the British element in the team - Dr. Nick Thomas and myself in Cardiff - was to identify changes in the gene in a number of samples from British families with XHED. This helped to prove that the gene actually was the XHED gene itself rather than he next gene along on the chromosome. We have also played a part, over the years, in narrowing down the site of the XHED gene on the long arm of the X chromosome. All this, of course, has only been possible because of the willingness of so many ED families to be included in the genetic research - thank you all very much indeed. I do not know whether these advances will ever lead to better treatment for people with ED, but it is certainly helping us to understand the condition.

I am hopeful that the laboratory scientists interested in the development of skin and other tissues will be able to make real progress over the next few years in understanding XHED and how it is related to the other types of ED. In order to help take this work forward, I would like to organise another study of individuals and families with all types of ED. I will be looking for funds to carry out this work, which will involve a researcher visiting people at home to ask about the condition in them or their family, and to examine them and (perhaps) take a blood sample as well. This study could help the laboratory scientists in their work on genes involved in skin and tooth development, and would help to clarify the distinctions between the different types of ED. When we know about the funding of this research I will let the ED Support Group know.

Thank you for all your help in the past. If you have questions about ED, particularly about the recent advances in XHED, you could contact your local clinical genetics unit or you could contact me directly.

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