News from our Geneticist

by

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The Wellcome Trust have recently agreed to support me in a study of the family impact of ectodermal dysplasia. When I was travelling around the country talking to families, examining affected boys with the X-linked type of ED and taking blood samples for genetic studies, I obviously heard a lot about the practicalities of living with ED and the emotional impact of the condition on affected boys and various female relatives. I was surprised but delighted when the Wellcome Trust agreed to support me in a study looking primarily at these issues rather than just at the clinical and medical issues connected with ED. I will therefore be contacting individuals and families whom I already know around the country to see if they would let me visit and interview them. I would also be very willing to hear from other families who might be interested in taking part in this study even if I do not already know them. If individuals or families have written down something about their own experiences then it could be very helpful to draw upon those accounts as well.

Finally, I think that X-linked ED families around Britain might be interested to know that our diagnostic laboratory has been working hard on some of the DNA samples gathered from families around the UK and they have now managed to identify a large number of mutations in XHED gene in these families. While this has so far been carried out as a research project, it should be possible to use this information in the future to identify women carrying the XHED in families with an affected male - where the women wish to know about this. The best way to access this type of testing is through the families' local clinical genetics unit who could make contact with our laboratory as and when appropriate.

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