

Introductory Speech for Roberta A. Pagon*

Kurt Hirschhorn



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It is a great pleasure to introduce Dr. Roberta Pagon, known to one and all as “Bonnie,” as the recipient of the 2006 ASHG Award for Excellence in Human Genetics Education. She is a highly respected clinical geneticist and holds the position of professor of Pediatrics, Medicine, and Ophthalmology at the University of Washington, where she also serves as attending physician in the Children’s Hospital in Seattle. Today, however, we are recognizing her for her work as the founder, director, and guiding light of the GeneTests Web site. I can remember when GeneTests was launched as “Helix, a National Directory of DNA Diagnostic Laboratories.” It was a much-needed information resource, a “Yellow Pages of testing labs,” as Dr. Pagon puts it, for genetics physicians and counselors faced with the frustrating task of finding who in the world was willing and able to perform molecular genetic testing for rare genetic diseases. Back then, positional cloning was already in full swing, and the list of genes responsible for Men-

delian disorders was growing by the week. It remains an interesting irony, remarkable then as now, that funding sources are willing to support basic research but often do so little to make sure the results of the research are actually usable to improve our patients’ health and well-being. GeneTests was an exception. It was meant to be a practical resource and was wisely supported over many years by the National Library of Medicine, with additional help recently from National Human Genome Research Institute and National Cancer Institute. The Clinic Directory that followed was an equally useful site, because, with most rare disorders, patients and their families live all over the world and cannot all be taken care of in the same genetics clinic. Knowing where to refer to colleagues with expertise in particular disorders is essential.

However, as useful as these directories are, the major educational impact of the GeneTests program is Dr. Pagon’s brainchild, GeneReviews, a collection of expertly authored and frequently updated reviews of genetic disorders that is available to one and all freely on the Internet. With reviews of over 360 disorders and a new review being added each week, GeneReviews has become a major educational resource for geneticists worldwide. One very appealing aspect of GeneReviews for me is that the authors are widely distributed geographically and constitute a worldwide community of geneticists who contribute their knowledge, without any remuneration, for the sake of their colleagues and their patients. I do not know of any clinical geneticist, genetics resident, medical student, genetics counselor, or counseling student who does not often make GeneReviews his or her first stop, to learn (or relearn) about various disorders to prepare for clinic patients and consults. GeneReviews serves a central role in genetics education, and we are grateful to Dr. Pagon’s vision in nurturing and expanding the Web site and bringing it to the prominent place it holds today. The ASHG recognizes Dr. Pagon’s contribution to all our educations and hereby presents her the 2006 ASHG Award for Excellence in Human Genetics Education.

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* Previously presented at the annual meeting of The American Society of Human Genetics, in New Orleans, on October 13, 2006.

Am. J. Hum. Genet. 2007;81:657–657. © 2007 by The American Society of Human Genetics. All rights reserved. 0002-9297/2007/8104-0006\$15.00
DOI: 10.1086/521406