2004 ASHG AWARD FOR EXCELLENCE IN HUMAN GENETICS EDUCATION
And the Band Played On…*

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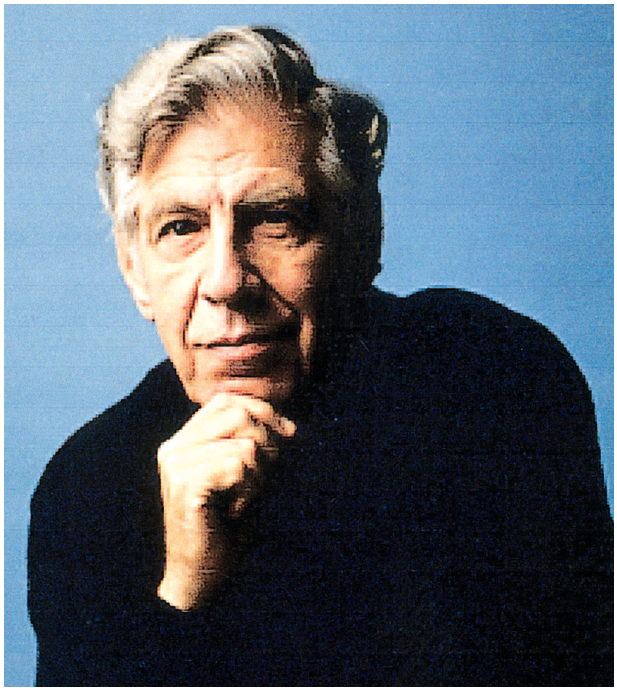

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When I received a phone call apprising me that I was the recipient of this august prize, I was, frankly, more than somewhat surprised. To date, I have written over 620 articles, 60 chapters, and have coauthored or edited 20 books, some of which have had Russian, Spanish, and Japanese translations—but the truth is that I am *not* a facile writer. I've always envied an author who puts pen to paper and out comes a nice, clean manuscript. I truly agonize over a sentence, and my way is *definitely not* the way textbooks should be written. In human genetics circles, my best-known text, *Syndromes of the Head and Neck,* was first published in 1964, with Jens Pindborg from Copenhagen, undoubtedly then the world's best-known and revered oral pathologist. That was after Woody Guthrie was in the thralldom of Huntington Disease and soon after Marilyn breathed/sang "Happy Birthday, Mr. President" and about the time the Kingston Trio was in its heyday—just after Martin Luther King's "I have a dream" speech, Kennedy said "Ich bin ein Berliner," and Cassius Clay fought Sonny Liston; everyone was saving Green Stamps. Our second edition appeared in 1976, with Mike Cohen added, and, for the third edition, Mike and I asked Steve Levin to join us. The text was totally rewritten and greatly expanded, finally seeing light in 1990. That was long before cappuccino and sushi went global, and e-mails from Cialis warning about 4-hour erections and good and bad cholesterol weren't daily worries. Although the title of our text did not reflect the coverage, we decided to let it stand. Steve Levin had, in the meanwhile, died of chronic myelogenous leukemia at an early age.

The story of why I wrote *Syndromes of the Head and Neck* is, in itself, rather bizarre. I had been quite influenced, during my early years, by Helen Curth, a German dermatologist who gave an apotheotic lecture at Columbia University Medical Center. That was post–World War II and pre-Korean conflict but definitely during the time of telexes and Watts lines and mimeographs and typewriters and three-martini lunches; Ollie was a one-toothed dragon, and Uncle Miltie was a household word. It was Helen who intrigued me by her discussion of the syndrome of acanthosis nigricans and gastric cancer. Having then been infected by Helen Curth with the concept of syndromes, I began to look for them on my own and reported a number of new ones in the early meetings of several oral pathology societies. It was at one of these that I first met Jens Pindborg. He expressed a profound interest in syndromes and suggested that we write a small monograph on the subject. I assured him that there was no way that I could get to Denmark, as he had suggested, because I had been but few years at Minnesota, and a sabbatical was out of the question. He told me to apply for a Guggenheim and a Fulbright fellowship, both of which I did, and both of which I received. So, off to Copenhagen. In retrospect, that was a huge mistake,

Received November 1, 2004; accepted for publication November 23, 2004; electronically published January 6, 2005.

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* Previously presented at the annual meeting of The American Society of Human Genetics, in Toronto, on October 30, 2004.

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0002-9297/2005/7602-0007$15.00

because our efforts should have been carried out on neutral ground. Jens was called away frequently to deal with things on his home turf and thus was not able to contribute very much to the writing of the text. However, the larger problem was in not being able to enter the stacks of their biomedical library. They simply didn't permit it. One could look at three periodicals every 15 minutes. That system did not work at all, at least for me, so I decided, after about 9 months, to return home to use my own library to finish the text. The final product was far different from the present-day effort, consisting of a few hundred pages on rather glossy paper in which we discussed a lot of conditions that were really inflammatory and not genetic but that involved the oral mucosa. All of this material was eliminated in subsequent editions.

It was my good fortune to meet Richard Goodman when he was a Genetics Fellow with Victor McKusick in the late 1970s. That was long before beach volleyball was an Olympic sport and before celebrity chefs and stock options. I had delivered several talks at Johns Hopkins and had met Richard during one of those trips, just as I had met Judy Hall and Charlie Scott. There was immediate rapport, and Richard (he abhorred the name "Dick") and I coauthored *Atlas of the Face in Genetic Disease* and *The Malformed Infant and Child*. Both had second editions, but when Richard died of intestinal cancer, also at young age, no further editions were essayed. This was about the time of Timothy Leary; Lucy was in the Sky with Diamonds, The Yellow Submarine was way off the charts, and Ringo was a household name.

Another happy union was joined with Bruce Konigsmark. Bruce was a neuropathologist who was persuaded by Victor to devote time for study of genetic deafness. He came originally to do neuropathology at Johns Hopkins, but Victor so captured Bruce that Bruce turned all of his attention to the genetics of hearing loss. Bruce always claimed that, having been trained as a pathologist and then as a neuropathologist, he knew nothing about deafness and less about human genetics. Victor encouraged him to start looking critically at what was available in the literature of the time concerning genetic deafness. This culminated essentially in a classic article that Bruce wrote for *The New England Journal of Medicine,* in which he talked about amassing a large collection of published material, separating it into appropriate piles such as "genetic deafness versus nongenetic deafness," discarding the latter, and then dividing genetic deafness into "isolated genetic deafness versus syndromal genetic deafness," finally separating the latter into "genetic deafness with endocrine problems," "genetic deafness with neurologic problems," "genetic deafness with muscular problems," etc. And so it came to pass—a plan that Bruce intended to follow.

Bruce asked me if I would send him articles that had hearing loss as a component that I encountered during my preparation for a second edition of my own text. I told him that I would, but, in further communication, he indicated that he would like to send me everything that *he* had written so that I could edit it for him. I told him that I could not do that, since I was extremely burdened putting together the second edition of my own text. Nevertheless, he sent me several cartons of manuscript that he had completed. Within months, Bruce succumbed, as had Steve Levin, to chronic myelogenous leukemia. I put together Bruce's thoughts and a few of my own, arranged the material into decent format, and used the title that he wished, *Genetic and Metabolic Deafness.* This text was published in 1976, a few years after Roe v. Wade and long before airports became shopping malls and prior to everything being Googled; there were no billionaires in Moscow. In 1990, Mike Cohen and I asked Helga Toriello to share the writing and editing of a new version of this text, because so many new deafness syndromes had been published. We changed the title to *Hereditary Hearing Loss and Its Syndromes,* and that opus saw light in 1994. This was continued by Helga, Willie Reardon, and me for still another edition, vastly expanded by the addition of single-gene nonsyndromal hearing loss, following the molecular explosion of the 1993–2003 decade, during which the molecular people discovered hearing loss as an ore-rich lode. This edition appeared in 2003, 10 years after I had officially retired and the year in which the space shuttle *Columbia* disintegrated and SARS reached epidemic proportions in China and began to spread around the world.

My best-known effort in oral pathology was a graduate or reference text that we called *Thoma's Oral Pathology.* Kurt Thoma was a Swiss-born oral surgeon who had written monumental multiedition tomes on both oral pathology and oral surgery in the 1920s and 1930s. Although Dr. Thoma had long retired, my coauthor, Henry Goldman, a pioneer oral pathologist and periodontist from Boston, and I so named our text to honor Dr. Thoma. It was published in 1990. It was a great sorrow for me when the second edition, almost completed, never saw the light of day. The reasons for abandoning further editions are too sad to relate. Several dusty unpublished chapters still grace my office shelves, often providing me with useful references for rare facial or especially dental anomalies.

Had I even a touch of the metaphysical about me, I would do my best to discourage individuals from coauthoring any text with me for, as I have indicated, Bruce Konigsmark, Richard Goodman, and Steve Levin died untimely deaths. Richard and I had talked about doing a *magnum opus* on congenital malformations. It was with great satisfaction that this work finally saw the light of day, under the aegis of Roger Stevenson and Judy Hall, in early 1993; it was a wonderful memorial to Richard's

vision. That was 4 years after the fall of the Berlin Wall; it was the year that Bill Clinton was sworn in as the 42nd president and a *fatwa* was issued against Salman Rushdie.

The fourth edition of *Syndromes of the Head and Neck* was written at a point when each new weekly or monthly human genetic publication presented us with additional molecular information. It was also written at a time when a spate of new syndromes appeared each month. We tried to be as catholic in our approach as possible, the limiting factor being that the facies must be unusual—hence, the title that is so well known that a change of name was not deemed wise. It took approximately 5 years of almost full-time labor to bring this edition to fruition. The finished text consisted of almost 6,500 typewritten pages (i.e., almost 15 reams of paper). The 2001 edition has about 1,275 pages; within the confines of these pages, we tabulated over 350 syndromes of orofacial clefting alone. Mike and I took on Raoul Hennekam as an editor and the individual who will carry the text to further editions.

I have been talking about the "good old days"—days before everyone was either a hero or a victim, before channel surfing and spin doctors and power breakfasts and retreats (which had a different meaning) and breakout sessions; "thinking out of the box" might have referred to baseball. There were no side-impact air bags or Humvees or AIDS or El Niños to worry about. There was no Euro for us against which to measure our ever-shrinking dollar. It was before Harry Potter; before Schwarzenegger was even a Terminator, much less governor; before the World Wide Web; before the rise of religious fundamentalism; and before the spate of e-mails I receive on erectile dysfunction and penile extensions.

That was history—at least my history. Were the good old days really that good? Probably not! But I have so enjoyed the compilation of a plethora of remarkable syndromes and have described about 100 new ones. The joy of being almost 82 is in seeing—and, rarely, at times—contributing to the molecular understanding of a few of these. If the late '70s was the Age of RFLPs, and the '80s and '90s saw the emergence of the Molecular Age, then surely the first decade of the 21st century is the Age of Molecular Pathways. It's been fun. Perhaps this will be my last presentation—perhaps, but I doubt it! Thank you all!