IN THIS ISSUE

Our genetic legacy

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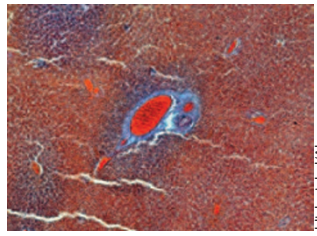
At present, it is accurate to declare that there are thousands of genetic providers involved in modern health care. However, not very long ago, the number of us was quite small. Whence those early few? Is it worth knowing their stories? And would we rather hear it from each of them or wait to read about them and their legacies in a journal section titled "In Memoriam?" *Genetics in Medicine* has opted for the former, providing a venue titled "Genetic Legacy," wherein those involved in clinical genetics are invited to provide a vignette about themselves. The point is to share how the person became interested in medical genetics, how he or she got started and made progress, and, finally, how he or she interprets their own story and finds its relevance for the future.

The premise is that where we are going reflects both where we have been and how those who got us here see the future. Hopefully, these vignettes will encourage incipient geneticists to take the plunge and will provide inspiration to those who have already taken their first steps. Ultimately, of course, it is the newbies whose efforts will have the most direct impact on our future, but sharing the past in the present—realizing the genetic legacy in the instant—is likely to have a wholesome indirect influence as well. Starting us off in a manner that epitomizes the evolution of health care–directed genetics, Judith Benkendorf shares her transition from ninth-grade awe to formulating the "counselome." —*Vincent M. Riccardi, History Editor*

First meeting of the ACMG Board of Directors, 14 September 1991, Lowes Hotel, Santa Monica, CA. Top row (left to right): Stephen Goodman, Patricia Murphy, Michael Watson, Lynn Fleisher, Michael Kaback, Rodney Howell, Robert Greenstein, James Hanson, and Kurt Hirschhorn. Center row: Jessica Davis, Maimon Cohen, David Rimoin, and Elaine Strauss. Front row: Sherman Elias, Laird Jackson, Arthur Beaudet, and Reed Pyeritz.

New: ACMG guidelines for management of GSD I

see http://www.nature.com/gim/journal/v16/n11/full/gim2014128a.html



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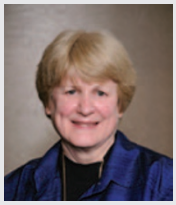
With this issue, the American College of Medical Genetics and Genomics offers the first published guidelines (available online only) for management of glycogen storage disease type I (GSD I), a group of rare diseases characterized by excessive accumulation of glycogen in the liver, kidney, and intestinal mucosa. GSD I is caused by either deficient activity of the glucose 6-phosphatase enzyme (GSD Ia) or a deficiency in the microsomal transport proteins for glucose 6-phosphate (GSD Ib). Patients with GSD I have a wide spectrum of clinical manifestations, including hepatomegaly, hypoglycemia, lactic acidemia, hyperlipidemia, hyperuricemia, and growth retardation. Individuals with GSD Ia typically have symptoms related to hypoglycemia, beginning in infancy. Developed by a team of experts in diagnosis and management of GSD I, the guidelines specifically address evaluation and diagnosis across multiple organ systems. The research team presents considerations for differential diagnosis stemming from presenting features and diagnostic algorithms. Also addressed are nutritional and medical management, including care coordination, genetic counseling, hepatic and renal transplantation, and prenatal diagnosis. The guideline is designed to allow health-care providers to recognize all forms of GSD I, to expedite diagnosis, and to minimize adverse events due to delayed diagnosis and inappropriate management. It also identifies gaps in scientific knowledge and suggests future studies. —*Michael S. Watson, Corresponding Author*

NEWS BRIEFS

Lasker award winner shines spotlight on breast cancer gene testing



In a bold move, Mary-Claire King, discoverer of the first gene identified as predisposing women to breast cancer, *BRCA1*, issued a public call for expanding breast cancer gene screening beyond the narrow group now considered at risk. King, currently at the University of Washington, Seattle, was awarded the 2014 Lasker Foundation award for her achievements in biomedical research. She seized the moment to suggest that population-wide screening in the United States could result in identifying more than a quarter million women at risk. In an opinion piece published in *JAMA* [2014;312(11):1091–1092] in conjunction with her award, King drew attention to a population-based study in which she and an Israeli research team screened an Ashkenazi Jewish population and found that the risks of breast and ovarian cancer for *BRCA1* and *BRCA2* mutation carriers identified through screening were as high as those identified through family history. "Women do not benefit by practices that 'protect' them from information regarding their own health," King wrote in her editorial. "They should have the choice to learn if they carry an actionable mutation in *BRCA1* or *BRCA2*." Any change in the