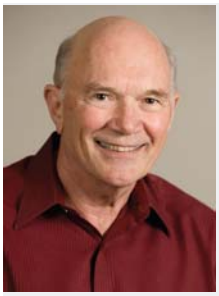

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From the Editors

Welcome to the first issue of ***The Journal of Rare Disorders*** (***JRD***). Currently, not a single journal in the United States is devoted to rare disorders. Now, for the first time, we bring together all aspects of rare diseases into one journal: the science, progress on new therapies, new drugs, and issues of drug regulation and cost. Papers on orphan drugs or rare disorders previously could only be published in specialty journals; however, our journal offers readers an open-access, online option to publish important new work in this field. We also anticipate contributing to awareness of the different types of rare disorders, and educating our readers about patient advocacy and the costs, both financial and personal, of these diseases.

Although, by definition, a single rare disorder affects fewer than 200,000 Americans, more than 7000 rare disorders have been diagnosed in the United States. Over 25 million patients are affected, of which approximately half are children. Information is often scarce, and physicians face many challenges in the diagnosis and treatment of these patients. To facilitate the exchange of information in this crucial area, the first U.S. journal dedicated to providing a forum for the discussion of rare disorders has been established. This peer-reviewed publication will focus on cutting-edge topics and is meant to expand the body of knowledge in this field.

Published quarterly, *The Journal of Rare Disorders* accepts for consideration original research, open-label studies, review articles, regulatory issues, case reports, and editorials. The journal is co-edited by Keck Graduate Institute faculty M. Ian Phillips and Timothy R. Coté. Dr. Phillips is the Norris Professor of Life Sciences and the Director of the Center for Rare Disease Therapies. Dr. Coté is the former Director of the Office of Orphan Products Development at the US Food and Drug Administration and currently holds the position of Professor of Regulatory Practice at Keck.

This inaugural issue covers Birt-Hogg-Dubé syndrome, familial and sporadic 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome), rare and orphan disease challenges, and an article by the Brain Vascular Malformation Consortium, entitled "Overview, Progress, and Future Directions."

In future issues, we hope to focus on specific therapeutic areas and to include useful tools for the practicing physician. Because diagnosis is a big hurdle that patients often encounter in the course of identifying their disease, we especially welcome clinical case studies that illustrate information about diagnosis. We are committed to making this new journal of value to the rare disease community, including physicians, researchers, regulators, patient advocacy groups, and pharmaceutical companies involved in developing orphan therapies.

We look forward to dynamic interaction with the rare disorder community and leaders in science and medical research. We encourage those in academia, regulation, and commercialization of therapies to send your manuscripts, case studies, suggestions, and feedback to us.

M. Ian Phillips, PhD, DSc, FAHA **Tim Coté, MD, MPH**