

From the Publisher:

Welcome to the third issue of *The Journal of Rare Disorders (JORD)*. There have been many exciting advances in the field of rare disorders and we are proud to be part of the dynamic discussion. Researchers and clinicians are making huge strides in treating these diseases and we hope to advance that mission with insightful articles that raise awareness and offer new therapeutic options. We are looking forward to the second half of 2014 and plan on publishing even more meaningful articles for the practicing physician. We are especially seeking clinical case studies that offer information on diagnosing rare disorders as this is a particular hurdle that patients often encounter.

The first article of this issue by Dr. Vincent Scoglietti et al, discusses the development of a clinical protocol for the treatment of Idiopathic granulomatous mastitis (IGM). IGM is an uncommon benign chronic inflammatory breast lesion, characterized histologically by noncaseating granulomatous inflammation. It presents a challenging clinical scenario, as it can mimic breast carcinoma on imaging and physical examination.

Our second article by Katrina J. Llewellyn, PhD et al, investigates autosomal-dominant myopathic disorder associated with diaphyseal medullary stenosis with malignant fibrous histiocytoma (DMS-MFH) which is characterized by myopathy, bone fragility, and osteosarcoma associated with mutations in the MTAP gene. The authors report on the myopathy in two cousins with DMS-MFH.

Proper design and reporting of clinical trials in rare diseases typically require nonstandard statistical approaches that may generally be optional in conventional large clinical trials. In our third article Alemayehu PhD et al, discuss some key statistical points for consideration in the design, analysis, and reporting of such trials.

Ter Haar syndrome (THS) is one of the most deleterious disorders known in medicine today. THS is characterized by congenital glaucoma, craniofacial abnormalities, hypertelorism, kyphoscoliosis, skeletal dysplasia, congenital heart defects, and developmental delay. In this case report Dr. Eda Haznedaroglu et al, discuss treatment of a patient with extreme longevity.

Finally, we include the *Rare Disease Impact Report*. This report was produced by Shire in collaboration with an expert global panel of patient advocates, physicians, and policy experts in the rare disease field. This global panel conducted survey research in the United States (US) and United Kingdom (UK) to determine the health, psycho-social, and economic impact of rare diseases among patient and medical communities. Key findings published in this *Rare Disease Impact Report* identify and quantify the impact of rare diseases based on survey responses from a multi-stakeholder audience of patients/caregivers, physicians, payors, and thought leaders.

The coming months promise to offer dynamic developments in rare disorders. New therapies are in development that will offer help to many patients who are without effective treatment options. We hope the rare disease community will continue to meet this challenge and we encourage those in academia, regulation, and commercialization of therapies to send your manuscripts, case studies, suggestions, and feedback to us at www.journalofrareorders.com/ContactUs.htm