Journal of
Bacterialogy and Infectious diseases
Editorial

The Challenge of Rare Diseases

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Rare diseases pose particular challenges to patients who are affected, to the clinicians who care for them, and to the investigators who study their conditions. Although individually uncommon, rare diseases are common in the aggregate, with approximately 7,000 described rare diseases affecting 25 to 30 million US adults. Challenges posed to affected individuals and their families largely regard being diagnosed, receiving optimal care, and affording disease-specific medications. Challenges facing clinicians who care for affected individuals include gaining knowledge and experience in caring for such patients, and the availability of local experts and of expert guidelines. Finally, challenges to investigators regard the difficulty and expense of assembling large cohorts of affected individuals for study, and garnering funding for research. Fortunately, in the face of these challenges, the steadfast resolve of patient and clinical/scientific communities to enhance care and generate new knowledge has fostered a large inventory of countermeasures to offset these challenges. Although further progress is surely needed, successes to date include the formation of powerful patient advocacy groups which have brokered collaborations between the patient, scientific communities, the government, and pharma/device communities in service of detection, optimal care, and research; procurement of funds to support research; formation of consortia of clinicians and scientists to collaborate; and general activation of the respective patient communities to perpetuate these successes. Persisting needs include enhanced detection strategies, dissemination of knowledge regarding optimal care, and research to prevent, treat, and cure disease. The challenge of these rare diseases is captured in the names of organizations charged with their oversight, that is; the branch of the Food and Drug Administration that oversees rare disease treatments is called the Office of Orphan Products Development, the designated group of the National Institutes of Health is called the Office of Rare Diseases, and the legislation that directs regulatory requirements for drugs and devices for these conditions is called the Orphan Disease Act. Orphan diseases may lack support and advocacy just as orphans lack parents. This perspective piece considers the challenges posed by rare diseases through three lenses: the challenges for patients with a rare disease, the challenges for clinicians who care for them, and the challenges for investigators who study their diseases. In considering each of these issues, I submit that generalizable observations emerge from experience with AATD, an important rare disease for which substantial progress has been made since its description in 1963.2 After considering the challenges, I review countermeasures to offset these challenges and some of the successes born by these measures to date. Challenges to Patients: The challenges to patients with a rare disease are threefold: they may experience the manifestations of the disease but struggle to find physicians knowledgeable about their condition to manage them; they may suffer the consequences of the disease and go completely unrecognized; and they may be faced with very high costs for disease-specific medications. Lessons from AATD amply demonstrate all three phenomena. First, of the estimated 100,000 US adults with severe deficiency of alpha-1 antitrypsin, < 10,000 have been recognized.3, 4 Such underrecognition appears to be a global problem, similarly observed in every country in which the issue has been examined.5 Furthermore, patients with AATD often experience long delays between their first attributable symptom and initial diagnosis (frequently called the diagnostic delay interval). Multiple series between 1995 and 20136, 7, 8, 9 report a persisting diagnostic delay interval of 7 to 8 years for individuals with AATD. Compounding this delay is the fact that patients with AATD commonly see multiple physicians with AATD-attributable symptoms before the diagnosis is initially made. In one series,6 43% of patients with AATD reported seeing at least three physicians before initial diagnosis, and 12% reported seeing between six and 10 physicians.