Editorial
Rare diseases of the digestive system

In 1932, Crohn et al. described a new disease, named “regional ileitis” in the JAMA as a case report [1]. The disease now is known as Crohn disease (CD). It carries a 2.6-4.5 fold increased risk for colorectal cancer, compared with the general population [2]. Regular surveillance is recommended for the patients with a history of CD for more than 8-10 years [2, 3]. Recognizing CD is critical for the associated mortality, treatment plans and comorbidities. CD involves approximately 0.5% of the population in the developed countries, with an estimated prevalence ranging from 0.6 to 322 per 100,000 persons in Europe, 16.7 to 318.5 in North America, and 0.88 to 67.9 in Asia and the Middle East [4]. According to the definition of rare diseases by the European Union (no more than 5 in 10,000 persons) [5], CD may not qualify for the designation of a rare disease in Europe and North America, but it may in Asia and Middle East. Nevertheless, CD has been included in the databases of the OrphaNet [6] and National Organization for Rare Disorders [7]. Its rarity has indeed been an ongoing challenge to conducting clinical trials [8].

Similar phenomena hold true in oncology research and practice. For example, Lynch syndrome was first described when studying two families of hereditary colorectal cancer in 1962 [9], which had been the research interest of late Professor Warthin at University of Michigan [10]. Nowadays, more than 100 cancer-prone syndromes and many genetic and molecular mechanisms have been discovered [11]. A whole body of clinical and scientific evidence supports regular screening and surveillance for Lynch syndrome [12]. Several “classics in oncology” also started from case reports or small case-series, including Kaposi sarcoma [13], post-mastectomy lymphangiosarcoma [14], lymphoepithelial tumor [15] and pheochromocytoma [16]. Undoubtedly, case reports played an important role in promoting the clinical practice and scientific research in oncology.

It appears clear to the editors that rare diseases involving the digestive system may have significant impact on the individual patients, public health, medical education and scientific advancement. There are currently 7,000 rare diseases identified [5, 17]. CD and Lynch syndrome are just two examples where case reports eventually led to critical scientific and clinical research findings, and benefited numerous patients. Many rare diseases, however, are poorly studied. Their etiopathogenesis, clinical presentation, diagnosis, management and prognosis are poorly defined. We therefore believe that there is an under-recognized need for more reports and studies on rare or uncommon diseases. A decision of accepting case reports in American Journal of Digestive Disease is thus made to meet such a need. While many journals have started eliminating the category of case report, we hope that this journal will offer a platform for publication of rare, yet interesting, cases with exceptional
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educational values. We are particularly interested in publishing cases with unique clinical, pathological and molecular features, ideally exceedingly rare yet clinically important. It is our hope that the publications will increase the awareness of rare diseases and thus strengthen the world-wide efforts in combating these diseases [5, 18].

In line with our thoughts, oncologists have already started paying more attention to rare diseases (rare tumors in their specialty), in part because there is a lack of clinical practice guidelines and personal experience. Reimbursement is also an issue associated with rare tumors [19]. Collaborative efforts are being made in accumulating experience on managing rare pediatric tumors [20]. Expanding clinical trials of novel therapeutics for rare diseases based on similar molecular mechanisms is also under way [17]. It is clear that an orchestrated effort involving policy-making, social support, patient education, physician education and research is needed to achieve the goal of better diagnosis and treatment of rare diseases [5, 18, 21]. We hope our decision of publishing case reports will contribute to the effort.

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References


