What’s New in Critical Illness and Injury Science?

Case reports: The first step on a path toward cure

Case reports, brief scientific manuscripts which present the salient details of an interesting or extraordinary case, its diagnosis and management, and a discussion of relevant literature, are often denigrated by the scientific community. Claims are made by their detractors that case reports are the lowest possible level of “evidence” in evidence-based practice that they do not truly add to the scientific literature, they are the realm of those that have nothing novel to study and report, or they are merely padding for one’s curriculum vitae. However, while these claims may be true in selected instances, case reports often have distinct value and may serve as the first step on a path toward achieving cures for human disease.

We live in a time where many life-threatening diseases have been cured or rendered into chronic, manageable conditions by medical or surgical therapies. If you couple this mental framework with the daily demands of a typical, high-volume clinical practice, it is easy to lose sight of the path a disease takes from identification to cure [Figure 1]. In my basic science laboratory, we study Hirschsprung’s disease, a congenital condition in which the distal enteric nervous system (ENS) fails to form because of failed migration of enteric neural crest cells. Currently, we establish the diagnosis based on the combination of clinical history, characteristic radiographic findings on contrast enema, and histology obtained by rectal biopsy. Treatment consists of surgical resection of the “aganglionic” segment that lacks an ENS, and multiple options tailoring surgical approach to the patient’s presentation are available. Furthermore, recent studies revealing suboptimal long-term outcomes are resulting in iterative improvements in our treatment approaches. Finally, basic science advances have brought us to the point of evaluating cell-based therapies to replace the missing ENS. All of these were made possible by the initial description of two patients with this disease by Professor Harald Hirschsprung.

Speaking in front of the International Congress for Children’s Disease in 1886, Hirschsprung described the cases of two children with congenital megacolon. This presentation, which appeared in the scientific literature the following year, led to a flurry of other case descriptions and series in the following years. Over time, these descriptions of a novel clinical entity coalesced into recognition of a true disease process, ultimately setting off years of intense study by some of the best and brightest minds in medicine and surgery. The next century was marked by steady advances punctuated by occasional setbacks in methods of establishing the diagnosis, medical and surgical treatment approaches, and basic scientific understanding of the underlying pathophysiology. However, our current ability to treat Hirschsprung’s disease can be directly traced back to Professor Hirschsprung’s case report.

Finally, case reports may help bolster the scientific workforce. Case reports are often written by a group of authors, typically with an undergraduate, medical student or resident as the lead author and a more senior clinician as the corresponding author. These brief manuscripts are an ideal venue for introducing trainees to methods for literature review, scientific writing, and the technical requirements of formatting and submission. Perhaps most importantly, acceptance of the manuscript and appearance of their case report in print provides positive reinforcement for the student – a tangible demonstration of their contributions to scientific knowledge on the path toward cure. Case reports represent an opportunity to therefore engage the best and brightest and encourage their careers in scientific discovery.
Therefore, I encourage the readers to consider the cases within this issue carefully and in the context of their own clinical experience. Perhaps these cases, viewed through the readers' lens, will provide a springboard to identification of new disease processes and reveal new paths toward cures.

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