

Doris T. Zallen  
Letter to the Editor, *Hastings Center Report*



To the Editor:

Emily Largent and Steven Pearson, in their article “Which Orphans Will Find a Home? The Rule of Rescue in Resource Allocation for Rare Diseases”, venture into difficult terrain: the allocation of scarce medical resources in these days when medical needs outstrip these resources and cost control is being sought. But their focus on the rare diseases requires comment. Rare diseases – or orphan diseases – are easy targets. Just the very label “rare disease” invites the view that such a disease is peripheral and unimportant to the health-care system. This is truly unfortunate. Not only does it seem to justify abandoning millions of people but it also can undermine the integrity of the entire research enterprise; it can reduce the chances of finding successful treatments for ALL diseases – common and rare alike.

Under the federal Orphan Drug Act of 1983, “rare disease” is defined as any disease that affects fewer than 200,000 people in the US. There about 7,000 such diseases. In all, rare diseases affect nearly 30 million Americans, two-thirds of them children. There are few or no treatments (under 400 rare diseases have approved FDA treatments on the American market) and there are few research efforts dedicated to finding more treatments.

Have individuals and families dealing with rare diseases resorted to “rule-of-rescue” tactics? Have they gone public with their painful personal stories to advance their place in line and deflect medical resources to their needs? A few have, of course. But the reality is that the vast majority of individuals and families facing rare diseases are invisible to the wider society. Their separate communities are isolated and small and, as is the case for many such disorders, their days are busy dealing their own medical challenges or those of their children. As a result, the vast majority of the rare diseases are unheralded and unknown to the general public. In fact, when it comes to publicizing disease stories and actually garnering media notice, it is celebrities battling common diseases (e.g., cancer, diabetes, heart disease, Alzheimer’s disease) who collect the lion’s share of attention. For those with rare diseases, it is simply not true that there is an orchestrated rule-of-rescue appeal for “unrestrained rescue”, as claimed by Largent and Pearson.

Largent and Pearson invoke the concept of “opportunity costs”, involving the loss to many who will do without if resources are used to help those with rare diseases. When considering opportunity costs, it is necessary to factor in the costs of the research dedicated to the discovery of treatments and cures. The investment in research being done on the common diseases is huge, and its cost is huge. By contrast, the current investment in research on rare diseases is very small. Further marginalizing this effort (because effective-treatment outcomes may be considered too costly to use) would threaten all of

rare-disease research and drive that limited research investment still lower. This would further discourage young scientists from doing rare-disease research, no matter how potentially fruitful such research might be, since it is impossible to build a scientific career without funding.

Reducing research on rare diseases would harm the broad scientific enterprise that supports modern medicine. Rare diseases, because they often have a singular genetic basis, present unique scientific opportunities for discovering and understanding gene action without the confounding factors that characterize the common disorders (factors such as contributions from multiple genes, environmental influences, diet, and smoking and other life-style choices). The study of rare diseases has already provided fundamental understandings of genetic systems, biochemical pathways, and DNA-repair mechanisms that have helped elucidate the basis of and improve treatments for common diseases. The blockbuster drugs Botox and Viagra were originally developed to treat benign essential blepharospasm and pulmonary hypertension, two rare disorders.

To overcome the already-existing research-funding shortfall, many patient groups try to raise money themselves. These groups often conduct garage sales, car washes, and bake sales. It can take five or more years for them to raise enough to fund even a single research effort associated with their particular rare disease. At a recent meeting of the Medical Advisory Committee of the National Organization for Rare Disorders (NORD), we struggled to decide which of two excellent research grant submissions should receive the available modest funding. Even with all its hard work, the patient group providing the funding had raised only enough money for one seed grant.

Those with rare diseases are not asking for “preference” when it comes to allocation of resources, as Largent and Pearson claim, but only a fair shake. The tax dollars used for governmental research programs come out of their pockets too, and society has an ethical duty not to punish minorities because of their small size. Not only they, but the wider community as well, would benefit -- since such research is essential to the health of the overall medical-science enterprise.

More attention should be directed to reducing the high cost of all medications and treatments. More effort could be given to encourage the many drug companies that are waiting to enter the medical marketplace with generic alternatives. Today’s expensive rare-disease drugs will become more affordable when generic companies compete and drive prices down. Since people with rare diseases are geographically dispersed, no single government or insurance company is overwhelmed by the cost of orphan drugs. In fact, insurers plainly admit that they could save more money by shaving a half-penny off the price of a cholesterol drug, used by millions of subscribers, than by slicing a few thousand dollars off the price of an orphan drug. The cost to society of orphan-drug treatments is far less than the cost of acne treatments.

As Largent and Pearson indicate, there are other ways to cut medical costs – ways that call for some sacrifice from all of us. That we will have to make tough calls about allocation of health-care resources is certain. However, there is no reason why those

dealing with rare diseases have to be first in line to justify their needs and defend their treatments.

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Reply to Dr. Zallen's comments from author Emily Largent:

“...Finally, Doris Zallen raised a salient point regarding the scientific value of rare disease research and how the benefits may accrue to all. A particularly clear example of this is the Undiagnosed Diseases Program at the National Institutes of Health, the stated aims of which are to provide answers to patients with mysterious conditions that have eluded diagnosis and to advance medical knowledge about both rare and common diseases. I support Zallen's call for further research, feel the government has an important role to play in both basic science and rare disease research, and furthermore speculate that comparative effectiveness research may strengthen the claim that many with rare diseases have on our limited health resources.”