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## 'The Needs Are the Same'

NIH Rare Diseases Director  
Discusses Research Challenges  
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*Wall Street Journal reporter Amy Dockser Marcus spoke with Stephen C. Groft, director of the National Institutes of Health Office of Rare Diseases, about the challenges of testing drugs and treatments for illnesses that affect only a tiny proportion of people. Below, an edited transcript of that discussion.*

### **WSJ: What are some of the key challenges involved in launching clinical trials in rare diseases?**

**Stephen C. Groft:** There is often a lack of availability of patients at any one particular spot. The difficulties of getting patients to research sites is not something we have been able to overcome easily, and trials often require a lot of travel. We are trying to facilitate this issue by having multiple investigators work on a common research protocol so that the trial can open at more sites.

There is also a need for a better understanding of the rare diseases, and much better diagnostic criteria to help researchers and clinicians identify patients more easily. And I think another major component is the lack of information or lack of access to the information about the clinical trials themselves. Information about clinical trials is readily available from a number of sources, including [clinicaltrials.gov](http://clinicaltrials.gov), but it is hard getting people to understand that there may be interventions available.

### **WSJ: How critical has it been for patient advocacy groups like the Progeria Research Foundation to play a role in getting clinical trials started?**

**Dr. Groft:** I think over the years we've seen the evolution of the patient advocacy groups. Particularly in rare diseases, it seems that the patient advocacy groups are the ones that have to lead a coordinated effort to achieve successful product development or rare diseases research.



Stephen C. Groft

The patient groups are the ones that are in constant contact with the pharmaceutical and biotech companies; they work with the academic research organizations and the individual researchers in academic institutes, they work with medical specialty societies, they are in touch with the federal government through the NIH in trying to gain access to researchers and to work with the FDA to get research protocols approved. With strong medical and scientific advisory boards, the groups are able to navigate through the complex resources and move research forward.

In progeria and other diseases, these groups provide a model for getting research completed that can lead to interventions. From what we have seen, the most successful product-development programs involve strong advocacy groups.

**WSJ: The NIH established a Rare Diseases Clinical Research Network in 2003, and its first clinical trials were launched last year. What was the idea behind that network?**

**Dr. Groft:** We are trying to facilitate clinical research in rare diseases by supporting collaborative studies at multiple research sites, utilizing multiple academic researchers, creating a common research protocol and then extending that to a number of research sites. Another need many people see is to try to find a way to increase the number of investigators who have a research emphasis on rare diseases.

The hope is that the studies that the network is doing could lead to trials. Some of the studies are natural history studies that follow patients for five years, eight years, or even longer. This is another weakness in rare diseases, that we often don't know the natural course of the disease. By following the patients over time, we hope to be able to make better conclusions as far as what we should be measuring regarding the disease.

Each of the consortia in the network was required to have a patient advocacy group that represents the disease working with them. We recognize that patient advocates are critical players. What we've noticed over the years is that active participation by patient advocacy groups is an essential component for stimulating rare disease research. There has to be a presence of patient advocates not only to help find the patients but also to convey information back to

**THE ORPHAN DRUG ACT**

The Orphan Drug Act awards companies a seven-year monopoly on a treatment as a way to encourage pharmaceutical companies to come up with cures for rare diseases. Learn more from the FDA.

patients and families as well as to the patient's local physician, who may not have ready access to these investigators or the research.

**WSJ: What needs to be done to further increase the attention and focus on research and trials in rare diseases?**

**Dr. Groft:** We need to generate the interest of the research investigators to focus on rare diseases, and

then after we have identified compounds, we need the assistance of the biotech industry to develop the products. Research can only go so far, then industry needs to get involved in order to take the product to the market.

The good thing is that as we are approaching the 25th anniversary of the Orphan Drug Act, we have developed models where we can help facilitate this relationship with industry and keep this moving forward. Right now we're also starting to see the globalization of research. There is more focus now in Japan, the European Union, South Korea, Australia, and some other countries in rare disease research. There is a tremendous interest in orphan disease product development in these countries and that can benefit all patients by stimulating more interest in rare diseases.

**WSJ: In a time of concern over the costs of health care, is it harder to justify research into diseases that affect fewer people?**

**Dr. Groft:** If you are a family member of someone who has a rare disease or you have a rare disease yourself, then the disease is no longer rare for you. Patients and their families spend a good deal of their lives living and coping with rare diseases. The needs are the same for patients of rare diseases and common ones: They are looking for an intervention to improve their lives and help them better cope with the disease.

Frequently we have found with rare diseases that new advances end up being useful for both rare and more common diseases. And one thing that we have been noticing, too, is that when the diagnostic criteria for a particular disease improve, the number of people who have that particular rare disease increase. Many times, since there is no intervention currently available, it is thought that there aren't that many patients available to be in trials. But through the globalization of research and the work of patient advocacy groups, we are finding that there are many, many more patients out there that can benefit from research on rare diseases than was previously thought.

There are a lot of people who have rare diseases that we simply don't know about yet. Once there is a critical mass of information and publications and research out there, more people with rare diseases come forward.