Diagnosing rare diseases

Here are some facts about rare diseases and physicians’ ability to diagnose and treat them:

- **7.6** Number of years it takes the average patient with a rare disease to receive a proper diagnosis
- **8** Average number of physicians seen before a patient receives a diagnosis
- **2 to 3** Average number of misdiagnoses received before a correct one is made
- **43.6** Percentage of patients with a rare disease who believe that because of a slow diagnosis, treatment was delayed, which may have had a negative impact on their condition
- **40** Percentage of specialists who rate their training on rare diseases as neutral, ineffective or very ineffective
- **56.7** Percentage of primary care physicians who rate their training on rare diseases as neutral, ineffective or very ineffective
- **53** Percentage of patients who rated the rare disease knowledge of the physician first seen at symptom onset as poor
- **92** Percentage of physicians who feel it’s difficult to address the needs of a rare disease patient in a typical office visit
- **60** Percentage of physicians who said they’ve received conflicting information about treatment of a patient with a rare disease
- **67** Percentage of patients and caregivers who said they have to educate their health care providers about a rare disease


Awareness aids orphan drug development

One aspect of getting funding for orphan drug development—convincing funders of the need—is easier than it used to be. At least that’s the impression of James Cloyd, Pharm.D., director of the Center for Orphan Drug Research at the University of Minnesota. The cost of doing the requisite basic science, animal studies and phase I, II and III trials hasn’t come down and investigators still must compete with those working on treatments for more common conditions. But now there’s more awareness of rare diseases than in the past and that’s having a positive impact. “Ten years ago, you may not have heard the phrase ‘orphan drug.’ Now, even in the popular media, the term shows up,” Cloyd says, pointing to recent articles in publications such as the Wall Street Journal as evidence.

That awareness is in part because of an international campaign to draw public attention to rare conditions launched in Europe in 2008. Now Rare Disease Day is observed around the world on the last day of February.

For the last three years, the University of Minnesota’s College of Pharmacy has organized Minnesota’s version of the event. Last year, Gov. Mark Dayton proclaimed February 28 Rare Disease Day, and the U hosted speakers who addressed improving care of those living with such conditions.

Cloyd says the campaign has increased awareness. “You don’t have to start out by stating, ‘I think we need to address therapies for rare disorders,’” he says. “There’s now an understanding that we should.”