**Editor's memo:**

# PAH: Looking for Zebras

Experts have always speculated that pulmonary arterial hypertension (PAH) patients who are diagnosed earlier tend to fare better than those who present later in the course of their illness. For example, we know that children are more likely than adult idiopathic PAH patients to respond to acute vasodilator testing. Whether this indicates that younger patients have a more active vasodilatory component and less "fixed" disease is not known definitively, but supports the notion that later onset disease may have been latent well before symptoms emerge. We also have research to support the benefit of treating patients earlier. Unfortunately, experts now know that even with the emergence of cardiorespiratory symptoms, there is still a severe delay in the time to diagnosis. If early diagnosis can lead to earlier initiation of targeted treatment for PAH, then we need to focus on what the major barriers are to achieving timely diagnosis in the modern treatment era.

In this issue of *Advances*, authors emphasize the high frequency of delay in diagnosis, which remarkably has not changed despite the major advances in medical therapy over the past 2 decades. Authors also highlight the role of both the patient and practitioner in delayed diagnosis. In a disease that often presents with symptoms that mimic other more common disease states—the so-called zebra—coupled with the reality that many physicians trained before the current era of novel therapies, the PAH patient often falls victim. Only through education and advocacy can we raise awareness and put the diagnosis of PAH on the minds of the front-line practitioners who usually see the patient long before they meet a PH expert. With Dr Greg Elliott's commitment to the PHA's early diagnosis campaign, he has guest edited a terrific edition of *Advances* in which authors help us to understand why delays in diagnosis occur, and emphasize that although PAH is a rare disease, it's one we can't afford to miss.

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**Guest Editor's Memo**

The story is familiar. A young woman presents to clinic years after she noted breathlessness and fatigue. She has seen other physicians, and the inhalers, exercise, and diet have not helped. In fact she is worse, much worse.

Delays in diagnosis of PAH are not a new phenomenon. The median time from symptom onset to diagnosis of primary pulmonary hypertension was 1.3 years in the early 1980s. That was an era without effective treatments. Now, 9 medications are approved for the treatment of Group 1 PAH; and yet the delay from symptom onset to diagnosis has not changed.

Earlier accurate diagnosis of PAH is imperative in an era of effective treatments. The Pulmonary Hypertension Association (PHA) launched a public awareness campaign "Sometimes it's PH" (www.SometimesItsPH.org); and this issue of *Advances* addresses key aspects of earlier accurate diagnosis of PAH. Lynn Brown, MD, describes what is known about delays in diagnosis of Group 1 PAH, emphasizing the importance of considering PAH as a possible diagnosis. Many PAH patients have co-morbidities such as obesity or asthma that mask PAH. Health professionals must consider "zebras" like PAH, especially when symptoms are out of proportion to those seen with more common disorders or when patients are not responding to treatment. Dr Brown also stresses the importance of patient contributions to delayed diagnoses. Like physicians, patients often attribute their breathlessness to growing older or being overweight. Laura D'Anna, DrPH, explores the difficult subject of health disparities on early diagnosis of PH. Social risk factors including low-wage jobs and low educational attainment often lead to delays in diagnoses of serious illnesses like PAH.

Screening protocols achieve early accurate diagnoses and improve outcomes for many disorders such as hypertensive vascular disease, cervical cancer, and breast cancer. Peter Leary, MD, and Jeffrey Edelman, MD, examine the subject of screening for PAH before the arrival of symptoms. Their article identifies at-risk populations, available screening modalities including genetic testing, and the limited evidence for therapeutic intervention early in the disease.

In addition to the 3 key articles by Drs Brown, D'Anna, and Leary and Edelman, 3 leading physician – scientists share their perspectives in the Pulmonary Hypertension Roundtable. Drs Marc Humbert, John Newman, and Julio Sandoval remind us that delayed diagnosis of PAH is truly a global problem. They identify many challenges and opportunities for the pulmonary hypertension community to achieve the goal of earlier accurate diagnosis of PAH.

Matt Hegewald, MD, provides the perspective of a busy practicing pulmonary and critical care specialist in "Ask the Expert." As in every issue, the regular sections supplement the publication's theme for practitioners.

In my view, the present state of affairs is unacceptable. Change will require the effort of the entire PH community to raise awareness and to devise, test, and implement new strategies to reduce the time from symptom onset to diagnosis. This issue of *Advances* and the PHA campaign "Sometimes it's PH" offer an important start.

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