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When Missing a 'Zebra' Can Land You in Court

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A Delayed or Wrong Diagnosis Can Be Lethal

One of the most famous axioms in medicine is, "When you hear hoofbeats, think horses, not zebras."

Every medical school student is taught that most diagnoses are more likely to involve common conditions and diseases than rare ones. Focus on the likeliest possibilities rather than the obscure ones.

That makes good sense—except when the physician is confronted with a patient who may have one of 7000+ rare diseases listed by the National Institutes of Health, each disease affecting fewer than 200,000 people in the United States. Together, rare diseases affect almost 30 million Americans or about 1 in 10 people. Globally, an estimated 350 million people have rare diseases.[1]

Physicians are generally unlikely to face a malpractice suit for misdiagnosing a rare disease. However, even though a disease might be rare, the results of a delayed or wrong diagnosis can be devastating or lethal. Juries have awarded millions of dollars in cases involving both primary care physicians and specialists. The doctors and hospitals not only missed the right diagnosis but did too little to find out why the patient failed to improve after their early treatment.

"We just don't get a lot of these claims," said William S. Kanich, MD, JD, chief medical officer for MagMutual, a Georgia-based medical malpractice carrier. "It isn't usually the rare diseases doctors get sued for. It's the common ones—missing heart attacks, appendicitis, cancer, pneumonia, etc.

Misdiagnosis Is Common

"The standard of care in rare disease cases, frankly, is to miss them," said Dr Kanich. "Juries tend to give physicians the benefit of the doubt in most cases. The key question for the doctor is: How do you know what you don't know? In the first 2 years of medical school, we'd often hear about pheochromocytoma, a malignancy on the adrenal gland, and other rare diseases. But in 15 years of practice, I've never seen one. It's difficult to diagnose a disease like that."

"If the patient isn't responding the way you think he should, that's when doctors have to ask themselves, 'What am I missing? What else could it be?'"

Doctors May Think They Have It Right

No one is exactly sure how often physicians misdiagnose or are late to diagnose medical conditions. “In most diagnostic errors, the physician was pretty sure of what was going on, except it turned out to be something else,” said Mark Graber, MD, founder of the Society to Improve Diagnosis in Medicine.

“Most doctors don’t even think about their track record in making diagnoses,” he said. “Autopsies have virtually disappeared. Doctors don’t get high-quality feedback. Often, you’ll never even hear if you injure a patient, who may go see someone else after you. Few healthcare organizations are measuring the incidence of diagnostic error in their own practices.”

A 2014 Institute of Medicine study[2] estimated that 5% of US adults, or 12 million patients a year, are misdiagnosed. Many of the errors are inconsequential, but some lead to serious complications and death.

It takes an average of 7.6 years for a US patient with a rare disease to receive the proper diagnosis, the 2013 Shire Disease Impact Report found.[3] Such patients typically visit up to eight physicians before they get the right diagnosis. Shire is an Ireland-based global specialty pharmaceutical firm.

Listen to the Patient and Family

About half of physicians Shire surveyed said that professional medical organizations don’t give enough attention to rare diseases. More than half said that there aren’t enough opportunities to network with other physicians who treat rare diseases. Primary care physicians “may miss the indications of a rare disease because they may have never seen a particular rare disease before, or the disease presents the signs and symptoms of a more common disease,” the report found.

The biggest mistake physicians make is not listening to the patient or his family when they say something is wrong, said the experts we asked.

“When the family tells you something isn’t right, listen carefully,” said Dr Kanich. “If a parent tells me their child isn’t well, I tend to believe them. Nobody knows their child better than the parents.”

Dr Graber agrees. In the cases he’s studied, parents “kept insisting that something was wrong and that the assigned diagnoses didn’t seem correct. We hear this over and over from patients that they weren’t listened to.”

Being Careful in Early Diagnosis

Not listening to the patient was a crucial element that led a Missouri jury to award $29 million last year to a young Springfield woman with Wilson disease that went misdiagnosed for 9 months.[4]

The college senior went to an internist, complaining of fatigue, tremors, panic attacks, insomnia, and other issues. The physician thought that her problems were related to anxiety and depression, and the young woman did have emotional problems, said her attorney Grant Rahmeyer.

Despite repeated requests from the patient and her mother for an MRI and a referral to a neurologist, the physician declined, preferring to adjust the dosages of her anxiety medications. Nine months after the patient first saw the doctor, she complained that her condition had deteriorated, with issues such as acting drunk, falling due to balance problems, and difficulty with handwriting due to the tremors.

The physician relented and ordered an MRI, which showed severe damage to the patient's basal ganglia caused by Wilson disease, a disorder that causes too much copper to accumulate in the liver and brain, according to the lawsuit.

As a result of the delay in diagnosis and treatment, the patient has severe brain damage, quadriplegia, dysarthria, dystonia, tremors, and sporadic contraction of her extremities. It took the jury only 2 hours of deliberation to reach its verdict. "She'll need caregivers for the rest of her life. Had the disease been diagnosed earlier, it could have been easily managed," said Rahmeyer.

"We're not saying that a primary care doctor has an obligation to diagnose Wilson disease. It's hard. But this doctor just wrote off her problems as anxiety when it was clear she wasn't getting better.

"It ought to be med school 101," he said. "Listen to the patient. If something is odd, you need to get to the bottom of it. If the doctor can't do it himself, he needs to make a timely referral. The doctor just anchored herself to the wrong diagnosis of anxiety and depression and then filtered everything through that."

Rule Out the Worst Possibilities

"We don't take malpractice cases just because a doctor made a mistake," said Malcolm McConnell, a plaintiff's attorney in Richmond, Virginia. "Doctors are human. But was the mistake a reasonable one? Did the doctor do a proper differential diagnosis, prioritizing according to the likelihood and severity? A doctor had a duty to rule out potentially lethal or life-changing conditions."

McConnell represented a patient who died from hemochromatosis, where too much iron is absorbed, builds up in the skin and liver, and can lead to cirrhosis of the liver, cancer, and death.

"This patient went to his primary care doctor for routine blood tests at least once a year for 12 years," he said. "On every single study, his liver enzymes were elevated. I don't say that the doctor had to diagnose hemochromatosis, but he needed to follow up on why the liver enzymes were elevated, which he never did. When the diagnosis was finally made, the patient had cancer and died. If he'd been diagnosed earlier, the condition could have been managed. A confidential settlement was reached."

"In another case, we won a $6.5 million verdict for failure to diagnose myelitis," said McConnell. "The patient, an attorney in his early 30s, complained of numbness in his hands and fingers. Over several visits and phone calls over months, the primary care doctor wrote it off as anxiety. Not once did she do a neurologic exam. The patient is totally disabled."

"Physicians sometimes have a bias when they make a preliminary diagnosis," he said. "It's like they have blinders on. They don't maintain an open mind. If initial therapy isn't effective, you

really have to do more to get to the bottom of the issue. If you follow proper protocols about testing and referral, she could have been led to the right diagnosis even if she never saw myelitis before. Doctors don't get a pass because it's a rare disease."

What to Do When You Don't Know What's Wrong

Charles P. Hehmeyer is a plaintiff's attorney in Philadelphia who specializes in inborn errors of metabolism, rare genetic disorders in which the body cannot turn food into energy.

He's represented clients when a heel stick test for newborns was done to test for genetic diseases such as phenylketonuria (PKU). Untreated PKU can lead to intellectual disability, seizures, behavioral problems, and mental disorders. The blood test is generally performed when the baby is 24-48 hours old.

"Every baby is screened, and most findings are treatable if caught early. You withhold or reduce the substance they can't metabolize," he said. "Negligence can occur in several ways. Labs may design a screening process that's flawed and leads to false negatives. The phlebotomist who collects the sample may do it at the wrong time, such as 8 hours after birth, when the child will more likely test negative."

"Some hospitals, especially smaller ones, will batch the samples, waiting until they have 20 or so before sending them to the lab. By that time, days have gone by, and the lab may not report the result in a timely fashion. By then, the child is seriously ill. After 72 hours of feeding, a child thought to be wellborn may become lethargic, vomits, has acidosis, and ketones in the urine. The doctors tend to think sepsis rather than a metabolic problem. They assume the lab will tell them if it's metabolic. Some kids get sick very quickly before the results are available, and by that time the child is dead or brain damaged. There's a need to refer to a pediatric intensivist much sooner."

"I have a case involving a baby who had a nasolacrimal duct obstruction. It's common to see blockages in part of the tear ducts. Rarely, the duct is plugged at the top and the bottom. There's a severe danger of infection. The pediatrician conceded that he'd never seen a case like this before. But he did nothing. He told the mother to come back in 2 weeks if there's no improvement. By that time the child suffered septic shock and brain damage," said Hehmeyer.

"The child's mother is a pediatrician herself. She knows how incredibly rare the condition is and didn't fault the doctor for not knowing how to treat it. If he had said honestly, 'I don't know what this is,' she would have called a pediatric ophthalmologist. But the doctor said, 'No big deal, we'll check it in 2 weeks.' That's negligence not to find out more."

Enlist an Expert

As the other attorneys suggested, physicians must consider the "worst first," Hehmeyer said. "If the problem is most likely to be A, but B or C can kill you, you have to rule them out. A doctor isn't required to know everything. That's impossible. But you need to know where to go when you don't have the answer. If you don't know, don't just assume. Know your own limitations and get an expert to help you."

"Defense attorneys always argue the hoofbeats theory, and it's often effective with juries. While doctors were taught about hoofbeats and horses, they were also taught that if you hear

hoofbeats but also see stripes, it just might be a zebra. I speak at meetings for several rare disease organizations. Many parents wear T-shirts that say 'Think Zebras.'"

Physicians also win malpractice cases involving rare conditions. "After their mother died of breast cancer, the family decided to sue the doctor," said James Lewis Griffith Sr., a veteran malpractice attorney in Pennsylvania. "We pulled all the mammograms, and the doctor properly interpreted each one. There was no evidence of breast cancer. It turned out the tumor grew on the chest wall itself and metastasized to the spine."

"It wasn't until the patient complained of back pain that they found it. The doctor testified that he can make diagnoses based on findings and symptoms. There was no back pain until much later. You can't x-ray everything. The doctor won the case. Jurors felt that his treatment was within the standard of care."

Where to Go for Help
"I grew up in the Midwest," said Dr Graber. "When there was a problem local doctors couldn't figure out, they'd consult with the Mayo Clinic. Most doctors these days don't have enough time to do a decent history and physical. But Mayo and many other centers of excellence around the country are available for consults and advice."

Here are some other resources:
The National Organization for Rare Disorders has a database with information for patients, families, and physicians about more than 1200 rare diseases. New topics are added regularly along with contact information for physicians who specialize in treating them.

The Genetic and Rare Diseases Information Center (GARD) is a program of the National Center for Advancing Translational Sciences and is funded by the National Institutes of Health. GARD provides the public with access to information about rare and genetic diseases.