Dimensions

One in a Million:

When Extraordinary Cases Occur in an Ordinary Practice

Contributed by

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My practice has never seemed to contain the seeds of the extraordinary, being established de novo in a small New England town whose economy has been supported by a mixture of tourism, fishing, and the 1980s New England real estate boom. Coming from Oklahoma, I initially found the ocean-related illnesses to be unusual,[1] but have since realized that every locale has its illnesses. (The physicians in New England had never heard of the brown recluse spider. I recently learned that *Centruroides exilicaude* antivenom is now available in Arizona.[2])

Although mine is a typical practice, truly "rare" cases have occurred at seemingly regular intervals. I am sure that any other family physician can also generate a list of rare cases (although with a different list of diagnoses). In the aggregate, rare cases occur commonly, and every practicing physician has to be alert to them.

Spells

The urgent tone of the mother's voice cut through my anticipation of an afternoon off. Only 15 minutes away from closing the office, I was suddenly 4 hours away from leaving the emergency department. "I'm a nurse and my baby's having a seizure," the mother said. The age alone, 14 months old, eliminated alcoholic seizures and posttraumatic epilepsy from the mental differential diagnosis I was forming. Thoughts of meningitis and febrile seizures accompanied me on the drive to the emergency department. On arrival, I was confronted with a nonseptic child with a blood sugar level of 26 mg/dL and an embarrassingly short differential diagnosis list. The decision to transfer my patient to a nearby tertiary care children's hospital was obvious. When my small patient, in whom nesidioblastosis had been newly diagnosed,[3] was returned to my care, little was needed but to follow along with the suggested treatment of diazoxide. But the case was not soon forgotten; the afternoon was counted as well spent.

Lumps

The oddest lump turned out not to be a lump at all, but a small *Dermatobia hominis*[4] worm trying to burrow into a warm spot of flesh. His host was a missionary, and the parasite quickly became an international traveler. In this case, the patient had consulted an ENT specialist at the university before coming to me. He had been no help at all. "We see such cellulitis around insect bites," he had told the patient. A telephone call to the patient's nursing supervisor in Belize, Central America, soon solved the mystery.

The saddest lumps have resulted in more common diagnoses but, these, I am glad to report, have occurred uncommonly. My practice has only had one 2-year-old patient with T-cell acute lymphoblastic leukemia (he is alive and well), only one adult with lymphoma (she was not so lucky), and one 39-year-old woman with metastatic colon cancer (the mother of one of my daughter's classmates, and likewise, she was not so lucky.)

Cough

Family physicians see many patients with cough, and a 17-month-old with a cough was not a red flag on my busy schedule that day. The child looked perfectly healthy, although she did have a 2-month history of cough with intermittent fever. On auscultation of her lungs, breath sounds on the right side seemed decreased, and even percussion seemed dull. A chest radiograph showed a large pleural effusion. The tumor behind it was not visible, however, until the pulmonary consultant tapped off the fluid. Her stage III-IV neuroblastoma[5] responded to MADDOC chemotherapy and hemibody radiation. So far she has not been left with any sequelae, but I have been left with a healthier respect for "cough."

Diarrhea

I already had a healthy respect for diarrhea, thanks to the occasional case of ulcerative colitis, giardiasis, salmonellosis, or campylobacter enteritis masquerading as "diarrhea." This woman's case was special, though. The patient herself was special: she was a sterling citizen, mother of three children under the age of 5 years, supportive wife to a boatbuilding husband, and owner of a great cookie store that catered to the tourists. With all of her family and seemingly all of the community suffering from diarrhea, her case seemed routine enough. Her diarrhea stopped each time she restricted her diet to clear liquids and resumed each time she tried to eat. The results of her laboratory studies were all normal; at least, I accepted her iron-deficiency anemia as normal enough for a young mother. But then she called and reported 29 bowel movements during the night! It did not stretch my imagination to believe her when she said she was too weak to have the outpatient small bowel study that I had scheduled.

I had already considered celiac disease[6] when the gastroenterologist suggested it, but I learned a lot from the case anyway. Besides the patient's iron deficiency, the other signs of nutrient malabsorption were impressive. Her serum calcium and vitamin D levels were low, and the parathyroid hormone (which we are supposed to measure with the intact molecule immunoreactive molecular

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assay [IRMA]) was compensatorily elevated. Her antigliadin antibody was even markedly positive. (I have yet to see a positive anticytoplasmic neutrophilic antibody for Wegener's granulomatosis, but I do not think I have missed any cases of Wegener's.) The possibility that mild cases of celiac disease are missed is sobering, and the availability of the antigliadin antibody is welcome.

Headaches

Family physicians commonly see patients who complain of headaches. Generally, we try to relieve the suffering and help the patient deal with stress, or give advice on how to live with cervical arthritis, or try to find a trigger for the migraine. Yet, in the back of our minds, like the veteran detective, we try to remain alert for clues of a more serious cause. Is this the patient with the tumor, the atriovenous malformation, the leaking aneurysm? My practice has yielded seven primary brain tumors over the last 10 years (four meningiomas, one astrocytoma, one acoustic neuroma, and one pituitary adenoma). Only one of these patients presented with headache as a major complaint. Seven cases in 10 years is less frequent than "common," but more frequent than "rare." Nevertheless, there is still something breathtaking about discovering a brain tumor,[7] and it is not soon forgotten. A careful search for atypical aspects of the headache and neurologic or endocrine symptoms provided clear-cut clues in all seven of these cases.

Pregnancy

Pregnancy is common enough, but this young lady had been uncommon from the beginning of her life. Abandoned as an infant on the streets of San Salvador, her unresponsiveness was accepted as part of the malnutrition suffered by such a waif. The story of her diagnosis of a urea cycle defect predated my knowing her, but the story became more complicated when she tested for pregnancy. A quick call to her gastroenterologist led to another call to the Amino Acid Disorder Laboratory. (As a family physician, I continue to be amazed at how subspecialized physicians can become, and how useful that obscure knowledge can be when you really need it.) Luckily the HHH syndrome[8] (hyperammonemia-hyperornithinemia-homocitrullinemia) is recessive. There had been two other known pregnancies in patients with HHH, and both had normal infants.

Later in the pregnancy the patient had a seizure, which is described as part of the HHH syndrome. A CT scan of her head showed intracerebral calcifications consistent with cerebral cysticercosis. My amino acid physician consultant had a Brazilian pediatric neurologist working in her laboratory, and I soon learned that in Central America (and probably much of the Third World) neurocysticercosis is the main cause of late onset epilepsy.[9] My horizons expanded.

How did I end up with the third pregnant HHH patient ever documented? Such cases prove the immense variety of the human condition and, despite all the criticisms, the beauty of our complex, scientific medical care system. Probably the greatest problem with that patient was a more typical family medicine issue: how can an 18-year-old woman with limited mental capacity and material resources possibly provide a healthy environment for a child?

My list of rare cases could continue. The first 10 years of my practice have also included a patient with vertebral osteomyelitis, two with primary biliary cirrhosis, one with an atrial myxoma, one with bacterial pericarditis, one with an ocular melanoma, one with amyloidosis, and one with a dissecting thoracic aneurysm. The occurrence of 20 unusual cases over 10 years in my typical private practice suggests that such odd cases will continue to regularly occur.

All practicing physicians should maintain appropriate suspicion when a patient presents with atypical symptoms or an apparently common disease that follows an atypical course. Unusual cases like these are intellectually rewarding to me, but do take additional time for the research necessary to generate an expanded differential diagnosis beyond that which quickly comes to mind. When the patient has been guided through a complex illness that he or she never dreamed existed, however, the patient-physician rapport that develops is tremendous. My relationship with consulting physicians and my position in the medical community have been strengthened by the successful management of these unusual cases. My humility and my respect for the difficulty of the practice of medicine have also been strengthened, but by those cases with poor outcomes. All of the cases have increased my wonder at the mystery of our existence and the vastness of the human condition.

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Cards, Cakes, and Homegrown Tomatoes

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The year 1993 will be a pivotal one for medicine in the United States. Bill Clinton has promised quick action on the problems of health care cost and access. The changes are likely to be swift and imperfect. To date, most of the debate has narrowly focused on how the country can provide more medical technology, to more of the people, at a lower cost. There has been no articulation of the need to provide each American with a personal relationship with a primary care physician. Instead, physician care—usually referred to as "physician services"—has been viewed in this debate as a generic commodity that can be bought, sold, reallocated, or traded to the lowest bidder. In our attempt to provide CT scans for all, will we develop a system that ignores the central role of the human relationship in the fragile balance between health and disease? Will the doctor-patient relationship survive the health-care-crisis solution?

Shirley's Case

I met Shirley 5 years ago. She had been regularly seen at the Rheumatology Clinic during the previous 2 years with complaints of swelling and pain in her joints, especially those in her hands. Each of these visits had been accompanied by detailed history-taking by medical students and drawing of blood for many laboratory tests. These often repeated tests invariably resulted in the same conclusion at the next rheumatology visit: "Findings are suggestive of rheumatoid arthritis, but the patient's rheumatoid factor is negative."

By the time I met her, she had been forced to quit her job as a seamstress. She simply could no longer sew. She was 28, married, had two children, and a very large extended family, all of whom I was later to get to know. She had been referred to me by her friend, who was also my patient.

Her rheumatologic history had been accurately and extensively recorded. Not noted in the chart was the fact that she was a tough woman who had rarely missed a day of work before her hands started to swell and hurt. She could not understand why her problem had remained so perplexing and why no one had been able to help. I reached out and took her hand. Her PIP and MP joints were obviously swollen, hot and tender.

The diagnoses were obvious: (1) seronegative rheumatoid arthritis; (2) reliance on a test result rather than the patient's signs and symptoms; and (3) lack of continuity of care in a subspecialty clinic.

I shared the first of these three diagnoses with Shirley and asked her if she would be willing to try an experiment. We would treat her with prednisone for 7 days. If my hunch was right, we could confirm the diagnosis in a week without drawing a single tube of blood.

I knew the results when I walked into the room that next week. She had a wonderful, little-girl smile. She presented her hands like presents to me—no warmth and no swelling.

My experiment had secured a trust that would endure. Not that it has not been challenged. Shirley has since had gold-induced nephrotic syndrome, azothioprine-induced diarrhea, and NSAID-induced gastritis. But Shirley is doing very well on 10 mg of prednisone every other day. She is sewing again.

Shirley and I share what is euphemistically referred to as a "doctor-patient relationship." She agrees to let me experiment with her care. I agree to be there when she is sick. She agrees to take her medicines, even when she does not like the side effects. I agree to think about her "case" sometimes when I am driving to work in the morning. She agrees not to call me unless it is important. I agree to get right back in touch with her when she calls. She bakes me cakes. I eat them.

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Patients as Voters

I worry that the people involved in the political debate cannot answer for themselves the simple question, "Who is your doctor?" They seem all too eager to replace the personal relationship between patients and physicians with micromanaged, bureaucratic, and inflexible "programs." Efficiency will be gained by "managed competition." Overutilization will be eliminated by "clinical guidelines." Medical care will be organized around "health care systems," not patients and their doctors.

Family physicians have been rare voices in calling for attention to this issue. Dietrich showed that primary care physicians manage 75% of patient care in a fee-for-service setting.[1] Franks, Clancy, and Nutting[2] have identified the doctor-patient relationship as an essential element for avoiding unnecessary treatment. These are undoubtedly the reasons why family physicians have been shown to be more cost-efficient providers of care than other specialists.[3] Mold and Stein[4] have described the "cascade effect" that occurs when patients are cared for by "systems."[4] Brody[5] has eloquently argued that the doctor-patient relationship must be the basis for ethical decisions about a "good death" that avoids prolonged pain and suffering.

Each medical specialty has its own therapeutic technology. Gastroenterologists have their endoscopes. Gynecologists have their laparoscopes. Cardiologists have their balloon catheters. For family physicians, the fundamental therapeutic technology is our relationship with our patients. Further compromise of this relationship will have a profoundly demoralizing effect. We will be like surgeons without scalpels.

Every year my patients send me cards, bake me cakes, and bring me homegrown tomatoes. This year I am going to ask them to write letters instead. If every family physician asked just five patients to write one letter each, every member of the House and Senate would receive approximately 1000 handwritten letters from "voters back home." Politicians respond to public opinion; therefore, this simple approach could have tremendous impact.

I am sure that it will be easy for you to identify five patients like Shirley in your practice. At their next visit to your office, tell them that you need their help. Sometime in 1993, you will ask them to write: "Dear Senator: I am writing on behalf of my family physician . . ."

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