

Virtual Mentor

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PERSONAL NARRATIVE

Through the Patient's Eyes: A Maddeningly Complex Disorder

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Though I don't remember his name, I will never forget his face. I was 9 years old, in the emergency room with two broken femurs. My parents—well-versed in fracture care, given that I had already had about 30 fractures—suggested that the doctor wait until I was sedated to remove my long leg braces. The braces were helping to stabilize the fractures, and minimize my pain. The doctor's response? "I'm the doctor here."

My whole family remembers this doctor. In the dozens of other trips we made to the emergency room, there were no doubt many more gentle, more compassionate doctors and nurses. But we don't remember any of them. We remember the one who was arrogant and insensitive to a child's pain and her parents' hard-earned wisdom.

I have osteogenesis imperfecta (OI)—a genetic disorder of type 1 collagen that causes fragile bones and a host of other problems. OI occupies an unfortunate place in the medical glossary of most physicians. It is common enough to merit some attention in textbooks. Most doctors I've met have heard of it. They ask to see my eyes (the sclera are blue—a telltale sign of OI). But it is rare enough that most physicians, even orthopedists, have never treated anyone with OI. Their knowledge is limited to a paragraph or two in a medical textbook, perhaps accompanied by a photo of a severely affected child.

OI is a maddeningly complex, variable disorder. It can be so mild it is never diagnosed, and so severe that newborns die within hours. I have a 30-month-old daughter who inherited OI from me. She has had one fracture. She has significant delays in physical development and just started walking this week. By the time I was her age, I had had seven fractures—six femurs and one radius. But I walked at 18 months. I spoke to a new mother earlier this week whose infant daughter has had as many fractures in her 6 months of life as I have had in my 34 years. I, my daughter, and that little girl all have OI. But for everything we have in common (fragile bones, blue sclera, loose joints) there are innumerable ways we are different.

Perhaps the most unpredictable facet of OI is the fractures themselves. My older brother accidentally tipped me out of my baby carriage when I was 2 weeks old. I was fine. One day when I was about 4, I sat on the floor and broke my femur. I've seen pictures of a teenage boy with OI snowboarding. This week, I heard about a

little girl whose arm broke while she was sitting—yes, just sitting there—in a grocery cart.

Few physicians have enough contact with the disorder to understand these variabilities. They tell parents whose babies are prenatally diagnosed with OI that they should prepare for their baby's death. Never mind that the most severe form, in which neonatal death *is* quite possible, is the least common of several types of OI, and that it is difficult to prenatally distinguish this type from the other severe but nonlethal types. Other doctors report parents whose baby has repeated unexplained fractures to social services, refusing to consider an OI diagnosis because they have that textbook picture in their mind of a severely affected infant with c-shaped femurs, a large head, and a tiny body. They can't believe that a child who looks completely "normal" (except for that funny blue tint in her eyes) could possibly have OI.

My family has been fortunate not to have either of these all-too-common, horrifying experiences. We have, by and large, been blessed with gentle and kind physicians who, although they are not OI experts, understand that we are, in our own never-been-to-med-school way. They understand that, though we can't explain the intricacies of OI genetics or the properties of type 1 collagen, we can tell when an injury requires an X-ray and when it just requires an Ace wrap. We know when our children have reached the limit of their endurance—when they need a sedative to calm them before the umpteenth IV stick, when they need mom or dad to position them on the x-ray table instead of an inexperienced X-ray tech, and when they need to keep their braces on bilateral femur fractures until anesthesia takes away the pain.

So we've been lucky. Our worst doctor-patient encounters have been like the one with Mr. "I'm the Doctor Here" in the ER 25 years ago. I wonder, did that doctor enter the profession hoping to be the kind of doctor his patients remember the rest of their lives? He has succeeded in that, though I'm sure not as he intended.

Ellen Painter Dollar is a freelance writer and former director of public relations and events for the OI Foundation. She edited *Growing Up with OI: A Guide for Families and Caregivers* and wrote *Growing Up with OI: A Guide for Children*, both published in 2001.

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