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Family Medicine

NARRATIVE ESSAY

Ninety-ninth Percentile for Hair

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After years of infertility, my wife and I, who are both physician-educators, were finally expecting. The pregnancy was remarkably unremarkable, at least from a medical standpoint. Just 3 days shy of her due date, my wife nudged me awake in the early morning hours: today was the big day. We grabbed our hospital bag and cautiously approached labor and delivery. Our son was born 5 hours later.

We were immediately struck by his full head of thick, wavy hair. We quantified it at about the 99^{th} percentile. He was born ready to star in a hair conditioner commercial. I breathed a huge sigh of relief; our background worry about complications was unfounded. During the first set of vitals, my initial awe of fatherhood switched to my physician sense of concern. Something was not quite right.

He was small for a full-term baby, weighing in at 5.5 pounds. His testes were undescended. He had an extra toe, so he didn't meet the full criteria of the common expression "a healthy baby with 10 fingers and 10 toes." Individually, I knew these findings were not uncommon or particularly worrisome. I felt like the stereotypical "worried well" first-time parent that I so often teach my residents to reassure. Hours later, my uneasiness resurfaced as I reflected on the growing number of clinical findings. I could rationalize most of the physical features with the glaring exception of his disproportionately wide thumbs. We took a deep breath and prepared to do the very thing we discourage our patients from doing: we asked Dr Google.

I slowly typed the term "big thumbs" into the search browser. An expletive burst out of my mouth as the results began to load; we stumbled upon my son's diagnosis within seconds. Images of children who shared a striking resemblance to my hours-old son filled the screen. They all had Rubinstein-Taybi syndrome (RTS), a diagnosis we had never heard of but would quickly become experts in.

My chest tightened as I skimmed websites and read the words: ocular abnormalities, congenital heart defects, and finally cognitive disability. I looked over to the hospital bassinet where my son was napping and quietly wept. My expectation of raising a "normal" child, a "healthy" child vanished. Many people would later comment on the fact that as dual physicians we had educational and financial resources to care for the health needs of any child. We still felt completely inadequate.

The denial stage quickly came after the initial shock. Maybe he didn't have RTS. I often teach my residents not to jump to "zebra diagnoses" when evaluating patients, as these findings could be normal. Even if he had RTS, maybe he would be the exception and not have any cognitive delays. But, by his 2-month well child check, his developmental delays were already apparent. At 9 weeks of age and several specialist visits later, the diagnosis of RTS was genetically confirmed. My hope retreated, at least temporarily.

I next went through a prolonged mourning period. Certainly, for the present, but more for the painful acceptance of the loss of my dreams for him. I would never have that special moment of adjusting his tuxedo on his wedding day or watching him rock his own child to sleep.

We felt like we were finally managing our new parenting roles and their unexpected challenges. Just as we were finally getting a handle on parenthood, then came Tuesday, September 13, 2016; our son had just turned 4 months old, and it would turn out to be the most terrifying day of my life.

My after-work routine of making a beeline to hug him felt different that day. He wasn't moving his right arm as usual. Erring on the side of caution, my wife and I carefully placed him in the car seat and drove to the emergency room. Several hours later, I followed the ER resident to the nurses' station to view his skeletal survey. Her voice started to shake as she pointed to the dozens of lytic lesions infiltrating his skeletal system. I couldn't begin to process what my eyes were seeing; no part of his tiny body was untouched.

I often teach my residents the art of giving bad news to patients. Empathetic honesty and allowing time for patients and families to process the diagnosis is vital. Despite my training and teaching, I was not prepared to be the recipient of this devastating news. A pathological fracture to the right proximal humerus explained his arm weakness, but there were far greater issues.

Lying awake at 1 am on the hard sleeper chair in the hospital room, I thought about the differential diagnosis: lymphoma, langerhans cell histiocytosis, and sarcoma. Sleep never came that night.

The final diagnosis was a mouthful: multicentric infantile myofibromatosis, his second rare diagnosis. The lesions were shockingly widespread, but thankfully his vital organs were spared. He would survive this.

We started chemotherapy the next day. We were told, the child whose hair was in the 99th percentile would likely lose all of it. I looked out of the window of the oncology floor onto the large cemetery below and thought about his future. Facing that mortality during weekly chemotherapy only strengthened my hope for his recovery. He had no hair loss in 5 months of successful treatment.

The jump from relatively little personal medical and insurance engagement to now was staggering. The several cumulative weeks spent on the phone with insurance companies and service providers, in physicians' offices, and hospital rooms was eye-opening to how little I knew about this side of medicine.

My residents frequently discuss their frustrations with the medical system while trying to care for their patients. As physicians, my wife and I have spent hundreds of hours talking with insurance companies to advocate for patients. As parents of a special needs child, we have cumulatively spent thousands of hours doing the same for our son. I teach my residents how to advocate for their patients, especially special needs patients. I want them to be knowledgeable about age-appropriate resources, insurance barriers that can impact every patient's care, but especially patients with special needs and their families. Professionally, I became a more empathetic and passionate physician because of him.

Our now-former 99th percentile for hair son turned 6 years old this year is doing remarkably well and now gets regular haircuts. We celebrate his successes in "inchstones" as opposed to milestones. We are so thankful for the immeasurable joy that he has given us and for the knowledge that we have gained to share with other special needs families and the residents we teach.

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