I'm Worried About This Daughter

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Lloyd had worried the residents since he came to the neonatal intensive care unit 3 days ago. He was a robust and vigorous baby at 3100 g, fighting the nurses and doctors with every procedure. One might wonder what he was doing there, surrounded by the shriveled bodies of the excessively premature, strapped to their machines and tubes. Lloyd could be heard regularly, crying for his next bottle above the din of whooshing ventilators and ringing pumps.

Lloyd had a cardiac condition that was thought to be life-threatening at the hospital where he was born. Soon after birth, he turned blue and nearly stopped breathing. The doctors then noticed that he had a loud heart murmur, and concluding that Lloyd had some awful cardiac condition, shipped him to the nearest tertiary care facility. As it turned out, the cause of this episode was that Lloyd had aspirated some milk. But he indeed did have a heart murmur, which was diagnosed as a VSD. He was now waiting for additional testing on his heart before he would be allowed to go home with his mother.

When Lloyd arrived at our hospital, the residents in the unit noted that he had a rather impressive hypospadias and he looked a bit strange. A genetics consult was called. The diagnosis of Opitz syndrome seemed quite appropriate, as the child had the hypertelorism, widow's peak, and mildly rotated ears to go with his hypospadias and heart defect.

At the time, I was a senior resident, just beginning my elective rotation on the genetics service, and this case was my first assignment. I found the salient articles, read them, and learned that Opitz syndrome is sometimes associated with mild developmental delay, and further, that it is dominantly inherited. I prepared myself to face the family fully armed with all available information by reading every word written about Opitz syndrome—the historic BBB/G syndrome debates, the laryngeal clefting, etc.

The next day, I sat in the neonatal unit's conference room, waiting to hear our geneticist explain the situation to the mother. As the mother, just released from the other hospital, was ushered in by the social worker, I was taken aback. There was something. I couldn't figure what—Wait! I quickly realized that she looked just like her son! Her eyes were widely spaced, she had a widow's peak . . . she had Opitz syndrome! I suddenly remembered reading that some female cases were missed because the hypospadias and other male GU findings, obviously absent in affected females, are often what initiate a genetics evaluation.

What to do? I was reeling. Fortunately, I was not doing the counseling; the attending geneticist was. I looked at him, and his face was serene, unchanged from a moment ago. Perhaps he didn't get it, I thought, so I leaned over and whispered to him, "I think she's got it, too." As he rose to greet the woman, he smiled without looking at me and gently nodded his head. I guess he had figured it out, after all.

The session went smoothly. He explained each piece of information to the mother, calmly explaining the findings, what genes are, how they control the way a person develops, and how they can be passed on from parent to child. The woman, from the lower socioeconomic stratum of our great city, seemed to understand. We left that meeting with plans for a follow-up the next day.

"Why didn't you say anything?" I asked later.

"It wasn't the right time," he responded. "We'll talk more tomorrow, and we'll mention it then."

That next day, the geneticist reviewed with the mother all that we had told her. She was able to repeat it back to us, but more by rote than from understanding, it seemed. He explained again the inheritance, and that these features can be passed on. Sometimes, he added, a parent may not even know that he or she has the syndrome, and it's only when the child is diagnosed that it becomes evident to the parent. To this she nodded. However, when he began to explain that he saw some of the same features in her that he saw in Lloyd, she balked.

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"No, I do not have anything wrong with me. I understand that my boy has something with his genes, but I am fine, my other children are fine. They each have different fathers, so they inherited different genes, right?" I was impressed that she had understood that much of what the doctor had told her, although recessive versus dominant inheritance obviously had eluded her. She appreciated our help for her son, but as for herself and her three daughters, they didn't need it.

The days passed, and Lloyd was transferred to the infants' ward for evaluation and correction of his laryngeal cleft (an additional associated feature of Opitz syndrome, whose discovery was prompted by the genetics service). Lloyd's mother continued to stand her ground. While she clearly understood the situation, it was equally clear that she was not as sharp as she appeared at first. She repeated ideas verbatim, but without an understanding of anything beyond the immediate facts, unable to think in even the slightest abstract terms. I was soon finished with my genetics rotation and said good-bye to Lloyd and his mother. I wished her luck and joked that I hoped to never see them again—in the hospital, that is. At first she was insulted, but when I explained what I meant, she smiled, thanked me for my trouble, and said good-bye.

Five months passed, and it was my turn to be the supervising resident for the infants' ward. As the patients were being signed out to me, I stopped my predecessor suddenly. To my surprise, Lloyd was still there. His laryngeal reconstruction had not gone well, I was told. It had broken down, and they would wait for healing before trying again. In the meantime, his mother was too scared to take him home. No one pushed the issue because the decision when to do his surgery was almost day to day.

After I received the sign-out, I made my way about the ward, meeting all the patients and their families. I saved Lloyd and his mother for last. She was delighted to see me again, she said, and I sensed that she meant it. She proceeded to tell me all that had happened to Lloyd in the past few months, the surgery, then pneumonia, now the waiting. I told her that I was replacing Dr Low, the previous senior resident, and that made her happy. "You, I trust."

Later that week, during one particularly hectic day, Lloyd's mom came to the ward, followed by her three daughters. She saw me, hesitated, and then started toward me. I've too much to do to spend time talking with her, I thought, but I could never be rude to her. So I put down what I was doing and prepared to listen.

"Doctor," she began, "I've done some thinking about what you said a long time ago, about how what Lloyd has can be passed on." Could it be true? Was she about to acknowledge that she, too, was affected with the same condition? "Well, it's about my daughter, my oldest one," she said as she pointed toward the three young girls standing by their brother's door. The two younger ones were carbon copies of their mother, almost certainly affected. The oldest, standing off to the side, was a tall young lady with few of her mother's features. Clearly, she did not have Opitz syndrome. "Like I said, I know I'm okay, and as you can see, my two younger ones are okay. But it's my older one I'm worried about. She's so... different from the rest of us."