

# Knowledge Is Power

**D**ID YOU SEE THE ATRIAL SEPTUM?" MY WIFE'S QUESTION sent a shudder down my spine. As soon as the ultrasonographer and her student departed, leaving us alone in the dimly lit prenatal obstetrics suite, my wife's courageous voice and straightforward expression jarred the eerie calm I had managed to maintain.

We were there as beneficiaries of the newly unveiled recommendations of the American College of Obstetricians and Gynecologists, which now offer prenatal screening for chromosomal anomalies to all couples regardless of maternal age.<sup>1</sup> The physician in both of us firmly held on to the axiom that knowledge is power: as the director of a clinic for adults with Down syndrome and the proud brother of a young woman with trisomy 21, I had encountered many young adults with this condition and had shared so many poignant stories with my wife. Early in this our first pregnancy we had agreed to undergo serum quadruple screening during the second trimester.

As it turns out, we had received 10-fold higher odds of having a child with Down syndrome compared to the population average: 1 in 50, rather than 1 in 500 for a woman my wife's age. The genetic counselor had called my wife immediately—sooner than expected, while I was traveling in Europe—and had urged us to schedule this urgent ultrasound. Although the time to detect abnormal nuchal thickness had passed, it would provide information on any other congenital malformation that may increase our odds. In my absence, my wife's surgical background had helped her keep her composure. On my part, frantic transatlantic telephone calls had vainly tried to soothe; e-mails written with untold emotion could provide only transient comfort.

Wasn't it ironic, after all: the clinician for so many patients with Down syndrome now had to confront the same bombshell within our own nuclear family. The tables had turned, and the healer needed healing, badly. In my own wounded soul I was now living what so many parents who enter my clinic had once lived; but I was also experiencing the raw feelings shared by so many others who, hidden from me, had received the same news but elected never to live with Down syndrome.

"I understand them now," I had muttered to myself, thinking of those expectant parents (as many as 92% by some estimates<sup>2-4</sup>) who reach the latter decision. If I, a person who belongs in the other 10%, who on a regular basis palpates the uncanny warmth and love individuals with Down syndrome exude; who witnesses the amazing superhuman transformation brought about in so many families; who enjoys hearing the stories of struggles, achievements, joys, and un-

conditional affection that invariably permeate my patients' lives; if I, a person who admiringly tells them about my own sister's independent job, her advocacy work, her kind e-mails, her swimming prowess, and her deep insights into her condition; if I, who had confidently snapped to my medical school interviewer that I could not conceive of my family without my younger sister being part of it, was now drowning in pain; if all of that was happening to me, I could not begin to fathom the tumult of sorrow, dread, and despair that must overcome others who lack my clinical or personal experience.

My wife and I had sought the comfort of that acquired knowledge. And there, in that ultrasound suite, we grasped for data that might unload the final blow or begin to lift the cloud. For we knew that septal defects are much more common in Down syndrome, and that such a finding would seal our fate; whereas a perfectly normal ultrasound might halve our odds.

And so, in the vacuum of any formal radiology training, both of us had strained to inspect the four-chamber view when the images of our child's beating heart fleetingly crossed the screen; as her heart happily beat away my own had sunk, because I too had failed to see the atrial septum; and a cold shiver had made a mockery of the silent, fake smile that I'm sure contorted my face in a feeble attempt to display reassurance.

Now my wife had anxiously broken the silence, seeking that reassurance I could not provide. But knowledge is power: I could not lie to her. Holding her hand in my own, I looked straight into her eyes and heard myself say: "No, Sweetheart, I could not see the atrial septum."

The spasm traveled in both directions, and we held each other as best we could in that cold, mechanical, aseptic, dark, and lonely clinic room. She lay back on the hard table as tears flooded her eyes. I squeezed her hand in mine and whispered into her ear what I had already told her so many times. That as parents we had two goals: first, that our child grow up to be a good person so as to leave this world a better place; and second, that she be happy. We knew—intellectually—that for individuals with Down syndrome, more than half the battle was already won: that extra chromosome carries the gene for plain goodness, a trait that turns out to be contagious; and happiness seems, for them, oh ever so simple to achieve.

That certainty, and our headstrong yearning for the deepest recesses of our faith, helped us survive the interminable

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agony of our wait for the obstetrician who would officially interpret the ultrasound.

“Congratulations, you have a beautiful baby!” he blurted as he rushed in. “But what about the septum?” I managed to mumble. “What septum?” And methodically he showed us how it was all there, he reminded us of the existence of the foramen ovale (“basic embryology,” he said with a playful smile), and after our agreement he informed us that our baby was a girl (after all, for us “knowledge is power”!).

Since then, I have often reflected on how my patients who have Down syndrome may view these official recommendations, or their likely outcome. They know. They question and they may wonder why we think that their lives may be less worth living, when they certainly exult at the chance to be alive and to contribute. Their improved health and remarkable quality of life stand as great accomplishments of modern medicine. As they enjoy the protections of an enlightened culture that welcomes diversity, as they enter a society that more than ever goes overboard to provide services and support to people with disabilities, will they understand the paradox of a door that may not let them in in the first place? I glance at their inquisitive faces and cannot begin to argue cost-benefit societal analyses when the benefits—the kindness, affection, delight, innocence, service, tenacity, and loyalty with which they grace us—cannot be quantified. And one fears that, if strength lies in numbers, their dwindling communities will no longer be able to advocate for themselves, while the rest of us will cease to benefit from their presence.

But knowledge is power. And if a pluralistic society protects individual decisions, we can certainly conquer the loneliness, fear, and anguish brought about by ignorance while those irrevocable decisions are being considered. Two recent surveys illustrate the persistent deficiencies that pervade our health care system when it comes to accompanying, counseling, and supporting expectant mothers who face the situation we have just faced.<sup>5,6</sup> And in a rare bipartisan initiative that bridges the extreme divide, the Pre-natally and Post-natally Diagnosed Conditions Awareness Act was re-introduced on July 18, as an amendment to the Public Health

Service Act that would provide timely, accurate, and up-to-date information on everything we know about Down syndrome (and other anomalies) to expectant parents who receive such diagnoses.

We have been told that some persons do not want to gain that knowledge, even when volunteered by those who have first-hand experience in living with Down syndrome. But as a society we Americans often encounter such resistance in other situations and have decided that knowledge—whether on the facts of evolution, sexual physiology, or the equal rights of women—must be offered even to those who seem most reluctant to receive it.

As for me and my wife, the relief afforded by that new information has not made the doubt disappear. The certitude an amniocentesis would yield is not an option for us, because we have decided we cannot place this pregnancy—much desired and long awaited—at risk. But we now face the remaining weeks with the strength and wisdom the fullness of this knowledge has given us. And we relish in the confidence that when Carolina is born, whether she has 46 or 47 chromosomes, she will be welcomed with the unbridled joy she deserves, that joy our obstetrician colleagues and their new parents share every day.

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