NEW PERSPECTIVES: PART I

A Day in a Transition Practice
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As an academic internist trying to find my niche, I began seeing adults with Down syndrome as they transitioned from pediatric services. I see patients age 16 to 65 and beyond. There is a new demand for internists caring for patients who have special needs and are living longer and healthier lives. Life expectancy for people with Down syndrome, for example, has increased from nine years in 1929 to almost 60 years today. 1 If the general population enjoyed the same increase, we might live to be 350.

When I began our transition clinic, I had limited experience with transitions or patients with special needs. I did, however, have enthusiasm, an outstanding mentor, and great support from colleagues and faculty. Our clinic began accepting patients once a month nine years ago, and today it operates one-half day per week. This article provides a view of the needs of this special population from the perspective of real patients.

Valerie
“Don’t shake her hand,” the care-giver reminded me the first few times I saw Valerie, a very pleasant woman who is in her 30s and has Down syndrome. To Valerie, the handshake signifies the end of the doctor’s appointment and prompts her to gather her things to leave, even if she just started the appointment. I instead begin our visits with a wave hello.

Theresa
During her first visit to our clinic, Theresa, a patient who is in her early 40s and has Down syndrome, eloquently described a problem with chronic diarrhea that had persisted for decades. Her condition created daily issues and fostered a fear of leaving the house and not being able to locate a bathroom. Despite an extensive workup for liver disease, no diagnosis had been found. She had been told, “That’s just a part of Down syndrome.” Of the few medical problems we find to be common in patients with Down syndrome, key among them is celiac sprue disease.2 After treatment for this disease, Theresa’s diarrhea resolved completely, and she remains symptom-free.

Brenda and Terry
Because Down syndrome is known to be associated with Alzheimer’s disease, the caregivers and physicians of patients with Down syndrome often fear that any decline in the patient’s function might indicate the onset of dementia. Since Alzheimer’s disease is a diagnosis of exclusion, it should not be made without a thorough workup.

Brenda is a nonverbal woman I began seeing in her 50s, when she was referred for “new psychosis and onset of Alzheimer’s.” She had reportedly begun trying to remove her shirt in public—a new behavior for her. Discussion with her caregiver revealed that Brenda had recently experienced a 15-pound weight loss despite her recent dietary habit of drinking milkshakes and avoiding other foods. An x-ray revealed a hiatal hernia. It turns out that Brenda had been trying to loosen her collar to the point of ripping off her shirt due to reflux. Her new behaviors were reversed with ranitidine.

Terry was reported to have experienced memory loss and a significant decline in skills. During his first visit, he was nonverbal and asleep in his wheelchair. Testing showed an initial thyroid-stimulating hormone level of more than 250 uIU/mL. Treatment of severe hypothyroidism and sleep apnea dramatically improved his memory and function.

Many of our patients are referred to us by Kishore Vellody, MD, who runs the Down Syndrome Center at Children’s Hospital of Pittsburgh. In most cases, the major medical conditions associated with Down syndrome present early in life. While about 50% of our patients have a congenital heart defect,3 these defects have invariably been corrected before I see the patients as young adults. While leukemia is common, the majority of cases occur before age 20.4

I am always happy to see referrals from Dr. Vellody’s clinic for two specific reasons. First, I can see the patients in a routine visit. Transitions are remarkably harder in a time of crisis. It is difficult to deal with urgent issues, refer to multiple new subspecialists, and begin invasive diagnostic workups when meeting a patient for the first time. Pouring through hospital records and sorting out laboratory results while trying to build a relationship with someone in crisis is much more difficult. Second, all of Dr. Vellody’s patients come with a comprehensive progress report that summarizes past problems and outlines ongoing issues that I may need to address. This is invaluable for making successful transitions and avoiding gaps in care.

Some of the challenges of the transition include the loss of access to services that are easily obtained continued on page 2
for patients under age 21. The structure of school is invaluable for our patients, and there are not always resources available to provide daily routine once they have graduated from the school system. Finding day programs or job programs is vital to the quality of life of our patients. Services to help maintain existing opportunities and introduce new ones are just as crucial.

Manny
Manny is a 26-year-old patient who has made a smooth transition to our clinic. As I do with all of my patients, I make sure that I address Manny first before I begin talking with family members who may have joined him for the office visit. I also make sure not to assume that I am more of an expert than the caregivers, whose coping strategies I gladly pass on to others.

Manny’s body mass index is 24, and he takes great pride in his healthy appearance. I was happy to learn that he has a personal trainer who works with him twice a week. In fact, I talk about Manny frequently with other patients, many of whom have been told that being overweight is expected of individuals with Down syndrome. Hearing Manny’s story inspires other patients to exercise.

David
David lives in a group home. One day, he became so agitated that he was admitted to our local psychiatric hospital. I never determined the cause of his acute agitation myself, but I learned from a caregiver that someone had come to vacuum David’s room, had encountered several piles of David’s belongings on the floor, and had moved these belongings to David’s bed not knowing that David had organized his belongings in a special way. The loss of organizational structure was debilitating to David.

I am always comforted to know that I do not need to be the one to diagnose every problem or solve every challenge that arises. Using the resources available—caregivers, family members, and trusted specialists—I never feel alone in caring for my patients. For example, while I might suggest that a person has sleep apnea, I appreciate the patience of our pulmonologists who work to find a creative diagnostic and treatment plan. Finding a plan is not always straightforward.

People come to our clinic for the first time with great trepidation. Those with special needs often are highly resistant to change, and the transition visit represents a large change. Many patients have had difficult experiences with new doctors and have a great fear of the unknown. Many are leaving the comfort of long-term pediatric care in addition to embarking on the changes of adulthood. After new patient visits, I often hear patients and their caregivers describe an enormous feeling of relief and gratitude.

While I feel a great sense of obligation in helping a population in need of good care, I also feel a great sense of reward in offering care, acting as a liaison with other medical providers, and becoming a stakeholder in the team of caregiver support that each patient develops. Though the patients who use our clinic may seem specialized, they demand only the same level of diligence, attendance to best practices, strong communication channels, and empathy that other patients demand and receive.

My work in the Down Syndrome Center has garnered a great benefit: More than any other aspect of my job, this work has allowed me to develop closer relationships with patients and their caregivers and to learn more from them. For example, I know now that when a caregiver suggests that I not shake a patient’s hand, there is a good reason.

Editor’s Note: Pseudonyms are used for all patients described in this article.

References