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hen you hear hoofbeats, expect horses, not zebras." Every medical student has heard this adage used to teach a simple principle of diagnostics: consider common diagnoses before rare ones because common ones are just that. This summer, I discovered the literal embodiment of this medical metaphor. In Lancaster County, Pennsylvania, the sound of hoofbeats is the sound of travel, the sound of Amish families arriving at a country clinic to place their health and their hope in the hands of modern medicine.

HOOFBEATS AND HEALING: MODERN MEDICINE MEETS THE AMISH

The Clinic for Special Children is a non-profit organization founded by Dr. Holmes Morton and his wife Caroline that serves as a medical home for Old Order Amish and Mennonite children with inherited disorders. For the past two decades,



the clinic team has been quietly revolutionizing the landscape of genetic medicine with their groundbreaking advancements in the treatment of metabolic diseases. Because the Amish end formal education after the eighth grade, they have no licensed physicians or nurses of their own. Despite this lack of medical professionals and their rejection of modern technology, the Amish choose to embrace genetic medicine. Amish parents have come to place their trust in the capable hands of Drs. Holmes Morton, Kevin Straus, and Nicholas Rider – physician-scientists who have received worldwide recognition for their research while remaining country doctors at heart. During my tenweek fellowship at the clinic, I studied glutaric aciduria type 1, or GA1, a rare genetic disorder characterized by the inability to metabolize protein. With an incidence of one in two hundred thousand live births in the non-Amish population, GA1 is a medical zebra. But among the Lancaster County Amish, one in two hundred babies is affected. Infants with undiagnosed GA1 grow and develop normally for several months until the stress of a typical childhood illness such as influenza causes an acute metabolic crisis and irreversible damage to the motor center in the brain. If diagnosed at birth, the crippling effects of GA1 can be circumvented through dietary

restriction and careful treatment during times of illness. Over the course of the summer, I studied scientific literature on GA1, shadowed Dr. Strauss during patient appointments, accompanied him on rounds at Lancaster General Hospital, met with patients and parents in their homes, and helped organize a GA1 family conference. I then used the information I gathered to create a guidebook to provide the parents of GA1 children with accurate, accessible information about this complex medical problem.

Each evening after a full day at the clinic, I got in my car and drove along the meandering roads of Amish country. When I reached the home of Elam and Barbie Stoltzfus, my Amish host parents, their four children drove up in their pony cart to greet me enthusiastically. The family and I spent many evenings around a campfire – chatting, firefly-catching, and eating whoopie pies. There was something about the crackling firewood and dancing firelight that gave my body a sense of warmth not thought to be missing on a 95-degree day without air conditioning. The casual and intimate conversation between friends, free from the pressure of a ticking clock or buzzing cell phone, did the same for my soul. Each night as I headed to bed and turned out my lantern, I was inundated with darkness – not the unsettling kind, but rather the kind one longs for when sleep is precluded by the glare of streetlights or an alarm clock display. Despite injuries from mistaking walls for doorways in the dark, my body appreciated the day's natural rhythm of darkness and light. At times, I forgot that my host family was Amish. I began to realize that people are simply people. But every now and then, something would remind me that I was living in a different time and place – like the time Stephen (age nine) asked what the Toyota emblem on my car meant. I told him it was the brand of car, like Honda or Ford. He stared at me blankly; I searched for a more



meaningful answer.

"You know how the breed of your driving horse is a Standardbred?" I said. "Well, the breed of my car is Toyota."

Stephen's eyes lit up and he nodded. "Oh, I get it!" Nearly every "English" (non-Amish) person I spoke with about my summer had heard of Rumspringa (or "running around"), the period beginning at age sixteen in which Amish youth are allowed to experience the world before deciding whether to be baptized into the Amish church, or to abandon that life for the lure of technology, convenience, and independence. I consider this summer to be my own reverse Rumspringa. I gave up electric lights, air conditioning, skirts above the knee, television, Internet, and equal status to men. But I received much more in return – lifelong friends, a reminder to slow down, and proof that medicine can be powerful yet personal. The Clinic for Special Children has shown me that genomic science can be incorporated seamlessly into smallscale primary care, resulting in a truly innovative form of medicine. As I strive toward a career in pediatric genetics, I will use the extraordinary work of my mentors at the clinic as a standard for my own medical practice, and as a guidepost for every experience I have along the

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Pennsylvania

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