

Scientific Acts of Love: *Time-Intensive Detective Work*

GRACIE COLEGROVE, AGE NINE, PRACTICES GYMNASTICS AND plans to join her YMCA team next year. Her brother Harrison just won his orange belt in karate. Two years ago, be-

fore they were diagnosed with Dopa-responsive Dystonia (DRD), a genetic disorder that presents similarly to cerebral palsy, they both needed to be strapped into their wheelchairs to keep them upright so they could be fed. "The life change is so extraordinary," says their mother Ellen Colegrove. "People couldn't believe it when Harrison started walking again. . . it would make them cry to see the change in him."

Dr. Brian Kossak, a neurologist from Dartmouth-Hitchcock Manchester, was treating the two siblings who presented similar cerebral palsy symptoms and just kept getting worse. He and other providers thought there was something unusual about this and ultimately they were referred to DHMC. CHaD's Neurometabolic Team ran the children through a battery of tests and confirmed that they actually were suffering from DRD.

DRD is, in fact, a children's version of Parkinson's Disease, but for reasons we can't yet fully explain a very treatable version. Untreated, it is a progressive disease that steals all mobility as muscles stiffen up. Harrison was twisted, his limbs drawn inward. Gracie's spasticity sent her arms and legs outward. While their minds were totally intact, they were trapped inside their own bodies. Every single muscle was affected and becoming paralyzed. "They were going downhill so fast," says Colegrove. "If they hadn't shown improvement from the medicine (dopamine), they would have needed in-home nursing care."

Referring doctors receive the benefit of an array of CHaD specialists who participate in the diagnosis. They include: Susan Berg, genetic counselor; Val Hani, genetic counselor; Nurse Practitioner Elaine Johnson (and previously Lynn Wolfe); and geneticists John Moeschler and Mary Beth Dinulos. Pediatric neuroradiologists, our pathology service, and fibroblasty (skin grafts) specialists provide related services. Thanks to their sleuthing and commitment, patients with rare disorders are re-

ceiving treatment that is dramatically altering the course of their lives. Gracie and Harrison Colegrove are striking testimony to the power of their work.



"These efforts from so many providers are, in effect, scientific acts of love," says James Filiano, MD, Associate Professor of Pediatrics and of Neurology, and Director of CHaD's Neurometabolic Program. "The multi-layered regional complex of the Neurometabolic Program is the very essence of a tertiary care center, providing an important collection of resources for our neighboring and collaborating practitioners."

The chronic long-term, but effective, disease management achieved with the

Colegrove children emphasizes the importance of CHaD's regional care. Gracie and Harrison are now symptom-free, and can do everything most children can. . . and more. Last June, Gracie and Harrison won two first place medals each in their respective equestrian events at the Special Olympics held at UNH in Durham.

C O N Q U E R I N G D R D

Pediatric Neurology care providers have three wishes: to prevent the diseases that severely afflict some children; if unable to prevent, then to treat such diseases; and, if untreatable, to help the family care for their children. Sadly, the current level of technology keeps prevention and cure still out of reach for most genetic diseases. Most effort is expended making children more comfortable and managing symptoms.

But there are exceptions. CHaD's Neurometabolic Program, established in 1993, performs labor-intensive diagnostic evaluations with the hope of finding rare treatable disorders. Astute, observant, committed primary care and referring specialist physicians, nurses, and specialists both in private practice and state programs send patients to us to be tested. The meticulous, compassionate efforts of nurses, laboratory and radiology technicians, radiologists, genetic counselors, secretaries, and other personnel are required to make sure that nothing "slips through the cracks." And, parents and patients must have enough fortitude to proceed with the long diagnostic process.

DRD is just one of the rare treatable disorders that Dr. Filiano and his colleagues are working hard to surface. Mistaken diagnoses have typically been either movement disorders or degenerative neurological diseases or cerebral palsy. Patients typically respond very well to treatment. . . as long as the real disease is identified. The cocktail of medicines required to treat DRD must be adjusted as the patient heals since the drugs themselves have side effects. Treatment may also require dietary management or giving other drugs to counteract symptoms. An initial work up consists of first ruling out more obvious other conditions with urine, gene and blood tests, or a skin biopsy. For DRD specifically, a spinal tap ultimately diagnoses this disease, but it is part of one big day of testing. Our CHaD team can provide the kind of thorough work-up necessary for accurate diagnosis without interfering with the care private physicians give their patients.

CHaD is currently seeking philanthropic support to help expand this program's investigative work in assessing potentially misdiagnosed DRD cases.