

The patient, the pediatrician and a national partner organization—an alliance

By Dianne Gray, Maggie Hood, MD and Stefanie Putkowski, RN



A rare disorder affects fewer than 200,000 people in the US at a given time. At least 50% of rare disorders affect pediatric patients. They are often lethal, leading to severe morbidity and mortality as the infant grows into early childhood or adolescence.

As a parent of a child born with a rare disorder, Dianne Gray writes: I'll never forget the day we received Austin's long-awaited diagnosis. In December 1995, Austin was diagnosed with a rare recessive genetic condition, **Neurodegeneration with Brain Iron Accumulation (NBIA)**. With five words, our lives were changed forever. We were told that day that his life would be short, his pain would be great, and that we had better get ready for a long difficult road ahead.

That day we became one of the million families worldwide diagnosed with a rare disorder, and it is then that our family began down a road of isolation that is still painful to describe. Our loving friends, family and community members, all wanted answers and certainty. 'Is it cancer?' they would ask, as though if I said 'yes' it would become a journey that they felt more secure with, and possibly something they, too, would know how to handle. When my answer was 'no' – that it was a rare disease, NBIA – they would shrug their shoulders and the conversation would end, not out of lack of care, but out of not knowing what to say. How often do we all have difficulty in conversing about something that we simply do not relate to?

During that time, though, I also turned to our pediatrician who admittedly was as baffled as we were with the diagnosis. To her credit, she became our champion. What she didn't understand, she researched. She took a 'let's roll up our sleeves' mentality and decided to become the team captain for our cause, and thank goodness for it, as we were stunned and probably more than a little lost in those first, all important weeks.

We trusted our pediatrician as she was a cherished part of our extended family and now she became an important safe haven for me, as she had been a part of our journey from the moment of Austin's birth. I not only counted on her professional experience but on her compassion, as I sobbed in her office during my then-infant daughter's well baby visits.

As our team grew to include a full array of pediatric palliative care specialists, our pediatrician asked for copies of reports and kept informed on Austin's progress. Together, they became instrumental as our family navigated a new medical regimen that dealt with symptoms that first appeared unmanageable. They all also connected us with the National Organization of Rare Disease (NORD).

Simply put, NORD became our lifeline. They taught me that we were not alone and that, indeed, we were a part of a rare disease 'family'. In addition, NORD provided Austin's pediatrician, other team members and myself with physician and patient-friendly rare disease reports, clinical trial information, and a listing of specialists who could treat Austin's symptoms. They also provided information on financial resources for specific situations and gave us materials on medication assistance programs.

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Through connecting us with physicians who understand and treat rare diseases and their symptoms, NORD provided access to research and effective care which gave us options for treatment...and in a world where we seemed to have so little choice, that was a blessing in itself.

Most important, NORD provided me with disease-specific association information, which opened the door to a community of parents where I was not only understood, but where my sorrow and determination were welcome.

In closing, I'd like to ask you where you, too, can help to forge affiliations and relationships for your patients and families, because we all feel a little lost in the dark at times and we need your help. I'm forever thankful for the light shared by those in the pediatric community and those serving the rare disease families they may never meet.

As a pediatrician who cares for children with life-limiting illnesses, Maggie Hood writes:

The pediatrician is clearly on the front line, as the child will typically be brought first to his/her office, often accompanied by an anxious parent. If a diagnosis of a potentially life-threatening, rare disorder is ultimately made – often by a specialist to whom the pediatrician has referred the child – the pediatrician still remains a most important and essential member of the child's team.

In fact, the presence of the pediatrician is needed now more than ever. He/she can serve as the anchor in the storm, as a most trusted advisor for a family going through a potentially nightmarish ordeal while still managing the routine medical problems of the child and siblings.

As Dianne so eloquently describes, the sense of isolation that ensues as the result of a rare diagnosis can be overwhelming for the child, family, community and health care team. As a member of an interdisciplinary palliative care team, I find the strength and variety of our partnerships to provide great value in reducing

that sense of isolation. In essence, Aristotle's quote – "The whole is greater than the sum of its parts." – aptly illustrates the world of hospice and palliative medicine.

As Clinical Information Specialist for NORD, Stefanie Putkowski, RN, writes:

NORD can serve as a lifeline for the families of children with rare disorders, and can provide the pediatrician with entirely dependable resources from our vast database of information – culled since NORD's inception in 1983.

NORD's services include:

- Rare disease reports that are in patient-friendly language, yet sophisticated and clinically accurate, reviewed by experts in the field.
- Referrals to reliable, trusted support groups for the family
- Help in locating potential clinical trials for the child
- Help in locating experts in the child's specific disorder
- Information regarding financial resources for specific situations and/or support for caregivers
- Medication assistance programs

NORD helps end that isolation by providing dependable resources, reliable medical information, and networking programs.

In summary, alliances among the patient, family, physician and expert organizations are more valuable than we realize. There are many such potential partners out there who can extend the expertise, access and reach of your practice, including that of your palliative care team. We hope that this article encourages all members of the pediatric health care team to continue to forge alliances that, in turn, will serve all those involved.

WEBSITE: www.rarediseases.org