

CURRENT SCIENCE

Volume 116 Number 3

10 February 2019

GUEST EDITORIAL

Patient advocacy

'You really don't know anything. You don't know about the system. You don't know about the drugs. You don't know about the science. You don't know how the government works. You don't know the FDA from the NIH. You're just out there yelling and screaming.' So said a housewife in New York city, who had trained as a biochemist, talking with a group of early AIDS activists of the AIDS Coalition to Unleash Power (ACT UP). She offered to teach them what she knew, and they took her up on it. These and other details of the strategies of AIDS' activists were analysed in a report entitled 'Back to basics. HIV/AIDS advocacy as a model for catalyzing change', brought out by HCM Strategists and FasterCures in 2011, and freely available on the internet.

In retrospect, the key features of the highly successful movement were (a) getting attention to the cause; (b) obtaining various types of technical and regulatory knowledge and proposing solutions; (c) building a strong community; (d) demanding accountability from the government and (e) building both inward-facing and external-facing leadership. Let us briefly look at a few of the things they did. For garnering attention, they had headline-grabbing manoeuvres such as chaining themselves to the boundary fence of the Food and Drug Administration (FDA), lying down on Wall Street in their thousands, and throwing condoms into a cathedral. For obtaining knowledge and providing solutions, they attended scientific conferences and learned about the minutiae of the science, and how the government and regulatory process work. They also actively engaged with senior officials of the FDA and National Institutes of Health, questioning the status quo and provided common sensical alternatives. One of their biggest accomplishments was to build a strong community. Long before any social media campaign was possible, they held nightly meetings of one or the other of their committees, so that 'There was always a meeting you could go to'. Strong bonds were forged between all those who met regularly, enabling all the action. They also built strong bonds with many others who were not directly affected, such as doctors, nurses and other activists. They managed to hold the government accountable by an unrelenting combination of street-activism and dialogue. They built leadership not only within the organization, reaching out to activists in other cities and keeping everyone motivated and focused, but also the leadership that spoke with various agencies of the government. The activists' knowledge, strategies and per-

sistence earned them the respect of many of those they confronted, who can even today recount happenings of that era with appreciation.

This movement had many impacts: It caused orders of magnitude more funding for research into this disease; it compelled the government to actively engage with patients; it changed the way drug approval happened for this and some other diseases; it saved millions of lives; it created a model of how patient advocacy could run, and it was a movement that spread across the world. Advocacy by patients, their families and others has a venerable history, and this is accompanied by a strong 'do it yourself' attitude. We narrate a few other examples here.

Another American housewife, whose son was born in 1968, ended up having a strong impact on the rare disease movement. Starting at about two years of age, the child started behaving somewhat abnormally, and this got worse over the years. The doctors did not have a label for it, and it was only after the mother chanced upon an article in a magazine that she realized what the problem was: Tourette syndrome. There was no satisfactory drug on the market then, and the doctor they were consulting put the child into a clinical trial. This was most satisfactory, except that the pharma company closed down the trial, abandoning the patients who were responding to the drug. This incensed the lady so much that it started her on a life-long mission to tackle the issue of rare diseases and the lack of orphan drugs. Starting with no knowledge whatsoever, and raising three children on the side, she joined whatever efforts she could, took on whatever tasks were necessary and got to know everyone who could help her. In due course she founded the National Organization for Rare Disorders (NORD), and occupied key organizational positions for over 25 years. She was a prime mover for the efforts that led to the passage of the Orphan Drug Act in 1983. Her book *Orphan Drugs: A Global Crusade* describes their journey, and is freely available on the internet.

Another American parent also found that his children had a rare malady, Pompe's disease. He changed jobs to know more about the pharma industry; chased down researchers across the US and Europe; started his own company to try and find a cure; saw it acquired by a large biotech company; drove the research to the point where it got into a trial and then did everything he could to get his children onto the trial. Another ordinary person who went to unbelievable lengths to help his children. The story of this family has been captured in a book and also in a film.

Yet another effort was that of a couple who set up a company to help find a drug for Niemann-Pick type C-1, which affected their twin daughters. Their story has been profiled in a documentary and on television.

The examples above are from the United States, but today there are plenty of efforts in India. The indefatigable energy of patient advocates, often in the face of personal tragedy. We describe some of their efforts below.

The Indian Institute of Cerebral Palsy (IICP) was established way back in 1975 by the mother of a child with cerebral palsy, who wanted to provide a positive environment for such children by empowering them with education and skill training. The institute has a formal course for training special educators. It has also established the National Resource Centre for Augmentative and Alternative Communication (NRCAAC) in Kolkata, and works closely with IIT Kharagpur to develop devices which can aid in communication. This has helped many CP-affected persons lead a full life.

Delayed diagnosis and the lack of a knowledgeable and supportive ecosystem continue to be challenges for rare disease (RD) patients in India today, and this has compelled many parents of RD-affected children to create their own support groups in recent years. The following accounts were narrated to us in-person.

It took seven years for one patient to be correctly diagnosed with Pompe disease. This disease needs enzyme replacement therapy (ERT), which can cost up to Rs 1 crore per person per year. This was beyond the means of the patient's father, and taking care of his daughter was a huge challenge. He first established the POMPE foundation and later the Organizations for Rare Diseases India (ORDI), an umbrella organization. ORDI along with another patient group, Lysosomal Storage Disorder Support Society (LSDSS), took every possible measure from filing Public Interest Litigations to appealing to the pharma industry to provide accessible and subsidized treatment. As a result, in 2017, 240 patients with lysosomal storage disorders (LSD) received free ERT in India. ORDI was also instrumental in setting up the first Centre of Excellence for Rare Diseases at the Indira Gandhi Institute for Child Health, Bengaluru.

Another father, whose son has Duchene Muscular Dystrophy (DMD), took upon himself the humongous task of setting up a research facility to find a treatment for his son's condition. Although not a scientist, this parent understood that simply importing a drug from another country would not be helpful because a personalized treatment needs to be developed based on each patient's mutations. He set up the Dystrophy Annihilation Research Trust (DART), which is a patient support group and a research centre for the development of genomic interventions for DMD.

There are numerous cases where parents did not receive proper counselling on time, leading to multiple affected children in the same family. One couple unfortunately lost three sons to an inborn error of metabolism (IEM). The first two babies died undiagnosed within a few days of birth. The father consulted many doctors

before the birth of his third child, and the infant was screened immediately after birth. The baby was found to be positive for an IEM. It was the first in India, and the 38th in the world, to be diagnosed with pyruvate carboxylase deficiency. However, the child could not be saved because IEM diets were not available in India at that time, and doctors and nurses lacked the expertise to manage such patients. Following this tragedy, the father set up the Metabolic Disorders and Rare Diseases Organization of India to raise awareness about IEMs, and make newborn screening and IEM diets available in India. Due to his efforts, many hospitals and states have made such screening mandatory. Further, the Food Safety and Standards Authority of India (FSSAI) has started a 'Diet for Life' project, which has enabled several multinationals to market IEM foods locally. A domestic company (Pristine Organics) has also started manufacturing these diets, and this has reduced prices significantly. Other organizations too have been successful in making their disease-specific treatments, such as anti haemophilia factor (AHF), intravenous immunoglobulin (IVIG), iron chelating agent (ICA) and ERT available and affordable.

The activities of different RD patient groups in India have helped raise awareness in the government, the medical community and among the general public. Most notably, their collective efforts have resulted in various policy changes such as the National Policy for Treatment of Rare Disease, 2017, although this Policy has recently been put in abeyance.

Let us step away from rare diseases for the moment and talk about activism in general. In India in particular, we see ourselves surrounded by an intransigent system, and so we resort to a philosophy of 'adjust'. But we forget the power of civil society which is the strongest democratic force in our hands and has been a driving force in the country since pre-independence. In Bengaluru, we have recently seen a movement against the construction of a steel flyover and another in favour of suburban trains. Both were successful. Activists fight for what they believe in and are, by definition, a pain in the neck for those they are up against. Of course, there are different types of activists – the spirited lone rangers, the good-humoured throng united by indignation or anger but also camaraderie, the boors it is difficult to sympathize with, the ultra-brave souls who take on powerful interest groups, and so on. Whatever their stripes or colours, they can be a force for good.

To return to the rare disease crusaders, support them: Stand with them, network them, fund them, collaborate with them and write about them. There are many ways to support these pioneers.

Mohua Chakraborty Choudhury
Gayatri Saberwal*

Institute of Bioinformatics and Applied Biotechnology,
Bengaluru 560 100, India
*e-mail: gayatri@ibab.ac.in