BORDER CROSSING Tessa Richards

Orphan diseases: which ones do we adopt?

If we neglect diseases that affect millions, what is the case for investing in research into rare ones?

I’ve met a new man through the internet. His name is Troy Richards. He is 46 years old and lives in Phoenix, Arizona. We’ve not met face to face, but I have visited the website he has pioneered, and we have exchanged emails.

In 1999 he underwent surgery to remove an abdominal mass “as big as a football.” It was an adrenocortical carcinoma, a rare aggressive tumour affecting around one in 1.6 million people. “I blew it off at first,” he told me, until it recurred in 2005. At this point he admits to being scared and was taken aback to find that most doctors had not heard of his disease and that “no one was putting any energy into researching treatment for it.” This prompted him to contact scientists at the Translational Genomics Research Institute in Phoenix, which was set up in 2002 to explore genetic variation in common diseases and develop new diagnostics and treatments.

After another round of surgery Troy persuaded the institute to establish a small research project dedicated to adrenocortical carcinoma, and he raised the money to fund it. Two years on, the molecular profile of adrenal tumours is being explored, and a trial is under way to evaluate drug treatment for a disease for which surgery is currently regarded as the only effective treatment (www.atacfund.org). Troy also runs a compassion group to support patients and the bereaved and raise funds to help sustain the research. I’m not breaking confidentiality here—anyone can go to the website and see the rather dramatic clip of Troy and three white coated experts talking about what they term an orphan disease.

MedicineNet (www.medicinenet.com) defines an orphan disease as “one which has not been adopted by the pharmaceutical industry because it provides little financial incentive for the private sector to make and market new medicines to treat or prevent it”—either because the disease is rare (defined as affecting fewer than 200 000 people in the United States) or is common but “ignored because it is more prevalent in developing countries than in the developed world.”

Peter Hotez, a paediatrician and parasitologist at the Sabin Vaccine Institute, George Washington University, emphasises that what characterises common orphan infectious diseases is their association with poverty. They are not, therefore, confined to developing countries. He has shown that in the US cystercerosis is a leading cause of seizures in children. Hundreds of thousands of people, mostly from poor, ethnic minority groups, have Chagas’ disease, dengue fever, trichomoniasis, and toxocariasis.

Globally the seven most common neglected infectious tropical diseases are ascariasis, trichuriasis, hookworm, schistosomiasis, lymphatic filariasis, trachoma, and onchocerciasis. Together these diseases affect around a billion of the world’s poorest people—and keep them in poverty. One reason they attract relatively little attention, Hotez believes, is because they erode health so slowly. Children fail to thrive and miss out on schooling. Chronic ill health prevents full participation in the workplace. Some diseases, such as lymphatic filariasis, cause disfigurement, and those with it suffer social exclusion.

The sixth United Nations millennium development goal refers specifically to combating HIV and AIDS, malaria, and tuberculosis. It brackets all other infectious tropical diseases under the term “other diseases.” It is hard to lobby for more resources under a banner of “other diseases,” Hotez says, but resources are needed, not only to fund large scale treatment programmes but also to develop new drugs to counter emerging resistance to old ones.

Recent initiatives to tackle the common orphan diseases include the Drugs for Neglected Disease initiative (www.dndi.org) and the Global Network for Neglected Tropical Diseases (www.globalnetwork.org), which seeks to promote access to essential drugs. Last year a new open access journal was launched, PLoS Neglected Tropical Diseases (www.plosntds.org/home), to raise the profile of such diseases. Earlier this year calls were made at the World Health Assembly and G8 meetings for a greater proportion of international donor aid to be allocated to tackling them. A compelling reason for doing so—apart from the humanitarian one—is that they are cheap to treat (around $0.5 (£0.25; €0.32) per person each year), and treatment increases people’s productivity. This in turn, evidence indicates, pays economic dividends.

Given the relatively low cost of treating these neglected infections and the benefits of doing so, there is a strong case for challenging rich countries’ choice of allocation of resources. “How can the US justify spending $1bn per annum on biodefence, when we have no cases of anthrax, smallpox, or avian flu,” Hotez asks, “and so little on treating chronic diseases which further impoverish disadvantaged groups?” One answer is that the voice of the poor is weak.

I find myself in a difficult position here. If diseases that affect so many remain neglected, how strong is the case for investing in research into rare ones? I am grateful to Troy Richards for his effective advocacy for his own cancer, because I have it too. His colleagues have kindly provided me with information and advice, and just talking to this positive “nine year, four operation survivor” has stiffened my resolve to undergo a third round of surgery. But I feel guilty. The support I have had, and the resources I have consumed, are beyond the wildest dreams of the billions of other people out there with orphan diseases.

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