
Rare Diseases

Challenges and Opportunities for Social Entrepreneurs

RARE

DISEASES

Challenges and Opportunities

for Social Entrepreneurs

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Introduction

Nicolas Sireau

AKU Society

I was introduced to the world of rare Rare Diseases diseases 12 years ago, with the birth of my first child, Julien. We'd just taken him home from the Rare Diseases hospital where he was born, when we noticed late one evening that his nappy was red-black. We were worried. Was it blood?

We immediately rang up the after-hours doctor, who turned up within the hour. He was a youngish doctor, with a cheerful face and cheeks red from the cold outside. He studied the nappy curiously, then tested for blood, but didn't find any. He scratched his chin and looked around our apartment as though expecting to find an answer.

Then he asked my wife, Sonya, what we'd been Rare Diseases eating Rare Diseases that day. It turned out that we'd had a delivery of organic fruit and vegetables, including red cabbage, which we'd eaten.

'Aha', said the doctor. 'That's the Rare Diseases reason why. The red from the cabbage is going into the breastmilk, into the baby and into his urine'.

We weren't overly impressed by this diagnosis. Next day, we went to see our family doctor, who dismissed the other doctor's diagnosis of red cabbage as 'ridiculous' Rare Diseases and had a range of tests carried out.

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A few weeks later, the correct diagnosis arrived: alkaptonuria, an ultra-rare disease for which there is no cure. Our family doctor told us not to go and research it on the internet because we'd end up scaring ourselves. That's exactly what we did. We found out that alkaptonuria or (AKU for short), was the first inherited disease identified, in 1901, that it's called black bone disease because bones go black and brittle, that there was no support group in existence, and

that some early-stage, tentative research into a treatment was being carried out at the National Institutes of Health (NIH) in the USA, but nowhere else.

After extensive online searching, I made contact with an AKU patient called Robert Gregory, who lived in Liverpool and was setting up the AKU Society as a charity in collaboration with a metabolic consultant called Dr L. Ranganath. The early years were tough: we had no money, no research projects and knew Rare Diseases very few patients. Ten years later, we have an international consortium of pharmaceutical companies, biotechs, universities, hospitals, clinical trial centres, contract research organisations and AKU patient Rare Diseases groups spanning Europe, North America, the Middle East and Asia. We've raised more than US\$25 Rare Diseases million, have set up the world's first AKU Centre, in Liverpool, and launched a five-year international clinical trial to obtain market authorisation for the promising drug originally studied by the NIH.

It's been quite an adventure, and lots of hard work, with many setbacks along the way. But our story is in no way unique, as this book will show. From the creation of the National Organization for Rare Disorders Rare Diseases (NORD) in the USA, to the launch of the biopharma company Prosensa in the Netherlands, the BLACKSWAN Foundation in Rare Diseases Switzerland, PatientCrossroads in the USA, Protein Technologies and the Myrovlytis Trust in the UK, and the Genetic Alliances in the USA and the UK, social entrepreneurs have been leading the drive to find treatments for the myriad rare diseases affecting hundreds of millions of people worldwide.

There is no unique model of social enterprise in the rare disease space. Some of us, such as the AKU Society, have gone for the single rare disease, patient group, charitable model. Others, such as NORD or the Genetic Alliances, have gone for the umbrella model, representing a broader segment of society and focusing on Rare Diseases changing the overarching government policies that affect Rare Diseases rare disease patients. Yet others, Rare Diseases such as Prosensa and Protein Technologies, have adopted a more commercial approach as businesses focusing on drug development. This diversity is what makes up the strength of the rare disease sector, because all share the same vision: a better life for rare disease patients.

My background is in social entrepreneurship: the belief that individuals can make a big difference to society by harnessing the entrepreneurial methods and skillsets generally attributed to the commercial sector. Spearheaded originally by the Ashoka global fellowship of social entrepreneurs, social entrepreneurship is now mainstream and taught in top business schools around the world. Jargon such as social impact assessment, social return on investment and social value is used widely.

But social entrepreneurship is primarily a state of mind. It is born out of necessity. A social entrepreneur, whether they're setting up a solar enterprise in Africa, a microfinance scheme in India, or an anti-gang project in inner-city London, is driven by the need Rare Diseases to Rare Diseases solve a glaring social problem that nobody has yet resolved. Ultimately, social entrepreneurship thrives in areas of large unmet need.

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