MANYEE LIEU

Support communities involved in disease studies

Lack of continued help for poor families involved in Huntington's-disease research has sown resentment and mistrust, says **Ignacio Muñoz-Sanjuan**.

After decades of research, a genetic therapy for Huntington's disease is being tested in clinical trials. Sponsored by Swiss pharmaceutical firm Roche and United States-based Ionis Pharmaceuticals, this trial targets the gene that causes the disease. If the new treatment works, it could offer a way to halt progression of this genetic disease — an awful neurodegenerative disorder that attacks mainly the brain. The disease is caused by a single gene mutation transmitted in a dominant fashion, so a child has a 50% chance of inheriting the disease if one of their parents carries a single copy of the defective gene.

Before the human genome sequence was available, the study of Huntington's disease relied on the help of generations of people in Venezuela, where it is highly prevalent. They donated samples of skin, blood and semen, and handed over organs of their deceased relatives, including their own children. Yet what have these communities got in return? Despite efforts by pioneering scientist and Huntington's advocate Nancy Wexler, who led the research in the Maracaibo region over two decades and founded a clinic there, they have received little or no benefit from the research they enabled. Because of inaction by local governments, they largely lack access to genetic diagnosis and counselling, have inadequate medical care and scant legal protection.

Does the biomedical community have a moral responsibility to ensure sustained support for people who were crucial to its research? I argue that it does. As a scientist dedicated to treating Huntington's disease, I struggle with the knowledge that the current quality of life of those affected is deplorable. I have seen people shunned and neglected by their relatives, sitting alone in darkened rooms, devoid of medical or social support. I have met the children of those affected, who are afraid of what will become of them. Tragically, suicide is common.

Some of the largest clusters of Huntington's disease in the world nestle in Maracaibo townships, especially Barranquitas and San Luis, where roughly a third of families have a history of the disease. Wander the streets of these shanty towns, and you will find symptomatic patients on every street corner; to the uninitiated, their numbers are staggering. Many other families with Huntington's live in similar conditions elsewhere in Latin America, particularly in Colombia, Brazil and Peru.

Many of the people I met there now resent and distrust scientists. They had hoped for treatments, and had expected help with palliative medication and improved living conditions. At the very least, they wanted feedback on how their contribution had helped.

Research, and especially basic research, is fundamentally disconnected from the realities of vulnerable populations. Is it unreasonable to expect investigators and their institutions to assume some responsibility for ensuring adequate care for volunteers and their quality of life? Perhaps studies in vulnerable populations should not be conducted at all, unless a comprehensive, long-term plan is drafted in cooperation with the research institutions involved and local and national governments.

At a minimum, and as described in 2002 by the Council for International Organizations of Medical Sciences and the World Health Organization, sponsors have a responsibility to ensure that people recruited for research from vulnerable populations "will ordinarily be assured reasonable access to any diagnostic, preventive or therapeutic products that will become available as a consequence". In the case of the recent Colombian trial for familial Alzheimer's disease, sponsored by Roche and California company Genentech, patients participating in the study have been guaranteed access to the medication. But this is not enough. Sponsors of drug trials should also support development in the wider community.

What infuriates the people in the Latin-American clusters of Huntington's disease the most, is that they still lack ready access to the genetic tests that could tell them whether they or their children will develop the disease.

Here, all scientists can help. The biomedical community can lobby and pressure national governments to include Huntington's disease in legislation on rare disorders, which guarantees access to tests and treatments, and then enforce these regulations. Although some legislative framework exists in some of the countries involved, it is hard to access, particularly for poor people.

Governments need to offer free genetic tests to everyone at risk, and to provide adequate genetic and psychological counselling, and recognition of their disease status, even in remote communities. To do this efficiently, a proper census of communities with suspected cases of Huntington's disease is necessary, as many of these communities are unknown to government institutions.

Without support, the cases of Huntington's disease in these communities will increase and create an even worse public-health issue. Governments can develop effective family-planning and gene-carrier identification programmes to curtail the prevalence of the disease. Such an approach has been successful in, for example, diminishing the incidence of β-thalassaemia in the Italian island of Sardinia. And because Huntington's disease only affects a few thousand people in each country, there is an opportunity to make a real difference.

The clinical trial of the new therapy is terrific news. But we must not forget or ignore the needs of those who made it possible. ■

THEY HAVE RECEIVED LITTLE BENEFIT FROM THE RESEARCH THEY ENABLED.

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