THE UNIVERSITY of EDINBURGH
School of Social and Political Science

ECRD2016 in Edinburgh
Theme 6: **Game Changers in Global Society**
Poster: **127**

Learning from the History:
Means of Imagining the Community of Rare Diseases

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Objectives
1. Examines historically how rare diseases have became an important public health issue in the 21st century
2. Explores how they can remain to be recognized as such in the future

Methods
This is based on a sociological study on the history of orphan drugs in the United States and Europe. In this study, published materials and interviews were analyzed to identify critical factors in the history.

Results

Phase 1 – Origin of the Orphan Drug Problem
Originally, orphan drugs meant "drugs of limited commercial value," in which pharmaceutical companies showed little interest. Some clinical researchers successfully developed a drug to treat their patients but could not find companies to produce it commercially. The rarity of their diseases was one of several reasons for their unprofitability and hence the companies' lack of interest. In the mid-1970s, the US Food and Drug Administration (FDA) started the discussion internally about providing financial and non-financial incentives for commercial development of such drugs. It concerned mainly drugs that were already developed but not made commercially available. Because the companies had produced some unprofitable drugs as "service drugs" in the past, the FDA believed the situation could be improved by provision of some incentives.

Phase 2 – Victims of Orphan Diseases
When the issue was picked up in the US Congress in the late 1970s, some patients and their families were invited to take part in the discussion for the first time and speak about their problems in their own words. They became collectively characterized as "victims of orphan diseases." This rhetoric shifted the focus of the discussion from drugs to diseases, and allowed patients of rare diseases with no therapies to be part of the discussion too.

The Congressional hearings provided a 'platform' for actors representing different stakeholders to converse with each other. Each of them tried to define what 'orphan drugs' were and proposed potential solutions for them, reflecting their own beliefs. For example, the pharmaceutical industry, tried to present orphan drugs as a consequence of strict drug regulation by the FDA and insisted that what was needed was not a new legislation for orphan drugs but reformation of its drug approval process.

Phase 3 – Rare diseases became opportunities
In 1983, the discussions in the Congress resulted in the Orphan Drug Act and orphan drugs became clearly defined as drugs for rare diseases, but pharmaceutical companies were still reluctant to undertake development of such "service drug." The real game changer was the emergence of biotechnology firms in the late 1980s, which successfully re-conceptualized orphan drugs as attractive "business opportunities." They did so either by salami-slicing (suggesting the drugs are intended for rare diseases despite their broader application potentials) or setting very high price.

In the 1990s, rare diseases became a policy opportunity in Europe as it represented a merit of its integration. However, the idea of orphan drugs as "business opportunities" was crucial for starting the discussion about having equivalent regulation in Europe, which was enacted in 2000.

Discussion
Rare diseases has become an important public health issue *by imagining its community* – the community is 'imagined' in a sociological sense that, while its boundary has never been clear and its members do not know most others, a wide range of actions have been identified with the interests of the community. Little progress was made when the community was imagined exclusively as that of 'victims' and those who cared about them, but the community became successful after policymakers and commercial organizations began to realize not only patients and their families but also *they themselves benefit* from addressing the issues of rare diseases.

Implications
1. With the Internet and Social Media, raising awareness of and advocating for rare diseases are much easier now than it was in the 1970s and 1980s. They are powerful tools for the empowerment of patients but there is a risk of 'fragmenting' the community if too much emphasis is placed on divergent and potentially conflicting interests.
2. Recent advances in genomics are increasingly 'sub-categorizing' what have been thought as common diseases and making them eligible targets for orphan drugs. The potential explosion of orphan drugs can challenge the relevance of the imagined 'rare diseases' community in the public health domain.
3. As the community does not exist but is imagined, its future and hence the progress in rare diseases depend on the effort to keep all the stakeholders interested and this may require some strategic shaping of its communal identity.

Acknowledgement:
This research is supported by the Wellcome Trust Senior Investigator Award "Making Genomic Medicine" (Grant No. 100597/Z/12Z),
PI: Professor Steve Sturdy, Science, Technology and Innovation Studies, University of Edinburgh