“GENETIC DIAGNOSIS AND EMERGENCE OF PATIENTS AND RELATIVES’ EVIDENCE-BASED ACTIVISM”

Fanny DUYSENS, PhD Candidate
Spiral Research Center, University of Liège, Belgium

“Inhabiting Postgenomic Worlds. Reconfiguring Responsibilities and Solidarities”
B.STS International Workshop – 07-08/12/2016 – KULeuven
“I finally had a diagnosis after 15 years of total wandering. And now we enter the era of genetic treatment” (Jeanne Pelat, ambassador of Téléthon 2016)

“This 30th Téléthon is launching the Generation of finders!” (Laurence Tiennot-Herment, President of the AFM-Téléthon)
Patients and relatives’ activism

- Patients and relatives’ organizations (POs) as “hybrid collectives” (Epstein, 2008) composed by “concerned members” (Callon & Rabeharisoa, 1999): patients, relatives, professionals (scientific, medical, allied health)

- “Evidence based-activism” (Akrich & al. 2013; Rabeharisoa & al. 2014)
  - “The development of modes of activism that focus on knowledge production and knowledge mobilization in the governance of health issues. We argue that such modes of activism are reconfiguring the role and dynamic of patients’ organizations, users’ and activists’ groups, and have broader consequences for the governance of the so-called ‘knowledge society’ in contemporary democracies”
The “medical diagnosis” as object of study

- A disruptive and a foundational moment in patients and relatives’ life courses, “that kind of experience where the structures of everyday life and the forms of knowledge which underpin them are disrupted” (Bury, 1982:169)

- Fundamental in the practice of Western biomedicine and healthcare (Jutel, 2009; Jutel & Nettleton, 2011)

- Main components (Blaxter, 1978; Brown, 1995; Brown & al., 2011):
  - **Categories**: the act by which a legitimate practitioner or diagnostician describes and categorizes a disorder based on specific symptoms
  - **Processes**: the continuous description and categorization, the ways by which the medical diagnosis is used
  - **Outcomes**: the practical implications for those who are involved (practitioners, patients and relatives in the foreground) > “a diagnosis can vindicate or blame, can legitimize or stigmatise, can facilitate access to resources just as it can restrict opportunities” (Jutel & Nettleton, 2011:797)

- The medical diagnosis as “enacted” (Gardner & al., 2011; Mol, 2002)
Narrative of genetic diagnosis

• “I got my diagnosis when I was 48 years old. So that is not a long time ago that I got the accurate diagnosis. Before, I got some diagnoses which were not accurate ones. Once I was told I was affected by Charcot–Marie–Tooth disease (CMT). Then I told Doctor V. about it. She fell to laugh: no, that is not CMT! Just by seeing me she told that was a stupid guess. But me, I had believed this for 20 years because I had always been told that I was affected by CMT. Someday, she told me that she had a new guess. I got tested and a problem with type VI collagen was detected. One test and Bethlem myopathy was diagnosed. This is a very rare disease. And this means that I had waited to be 48 years old to get THE accurate diagnosis” (narrative from member of a Belgian PO concerned with neuromuscular diseases, 2015)

• Bethlem myopathy is “an autosomal dominant form of slowly progressive myopathy and collagenopathy”. Prevalence 1/1,000,000. Autosomal dominant heredity or autosomal recessive (www.orpha.net)
Emergence of evidence-based activism

• The “social diagnosis” (Brown & al., 2011:942)
  • “It is not about the present, where people seek knowledge about the symptoms they experience. It is also about the past, whereby people seek the causes of problems that led to a current diagnosis. As well, it is about the future, where the medical and social sequelae of the diagnosis reside along with the treatment and prognosis”

• In the field of neuromuscular diseases, emergence of evidence-based activism focused on the search for genetic causes, development of medical genetics and genetic therapies

• “Despite the persistence of failures, the advocates of gene therapy […] say that ‘it will eventually work’, and have created such a social expectation that the ‘mystique of the gene’ imposes everywhere, even in the imagination of each” (blog of Prof. Jacques Testard, Research director at INSERM, 2011)
P Os and co-construction of “gene worlds”

- How is genetic knowledge made possible?
  - Production of genetic information
- What are the consequences?
  - Reception of genetic information through diagnosis

- **Gene worlds** as “networks of people whose cooperative activity, organized via their conventional ways of doing things and subjected to regulations and resource constraints, produces current genetic knowledge” (Timmermans & Shostak, 2015:3)
  - “upstream factors that make clinical gene worlds possible”
  - “downstream dynamics that shape their uses and meanings”
THANK YOU FOR YOUR ATTENTION!

Fanny.Duysens@ulg.ac.be