

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

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*“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)*



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« CE 30E TÉLÉTHON LANCE LA GÉNÉRATION DES TROUVEURS ! »

**MERCI**

Le Téléthon affiche pour sa 30e édition un compteur de 80 319 113 d'euros au terme des 30 heures d'émission.

*“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 & Rabearisoa, 1999): patients, relatives, professionals (scientific, medical, allied health)
- “**Evidence based-activism**” (Akrich & al. 2013; Rabearisoa & 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](http://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)

# POs 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!***

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