CO-PRODUCING EVIDENCE: ETHNOGRAPHIC INQUIRY OF A "WILD" SEARCH FOR CAUSATION

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T105 "Wild research: Radical openings in technoscientific practice?"

“Wild research” or “Research in the wild” (Callon & Rabeharisoa 2003)

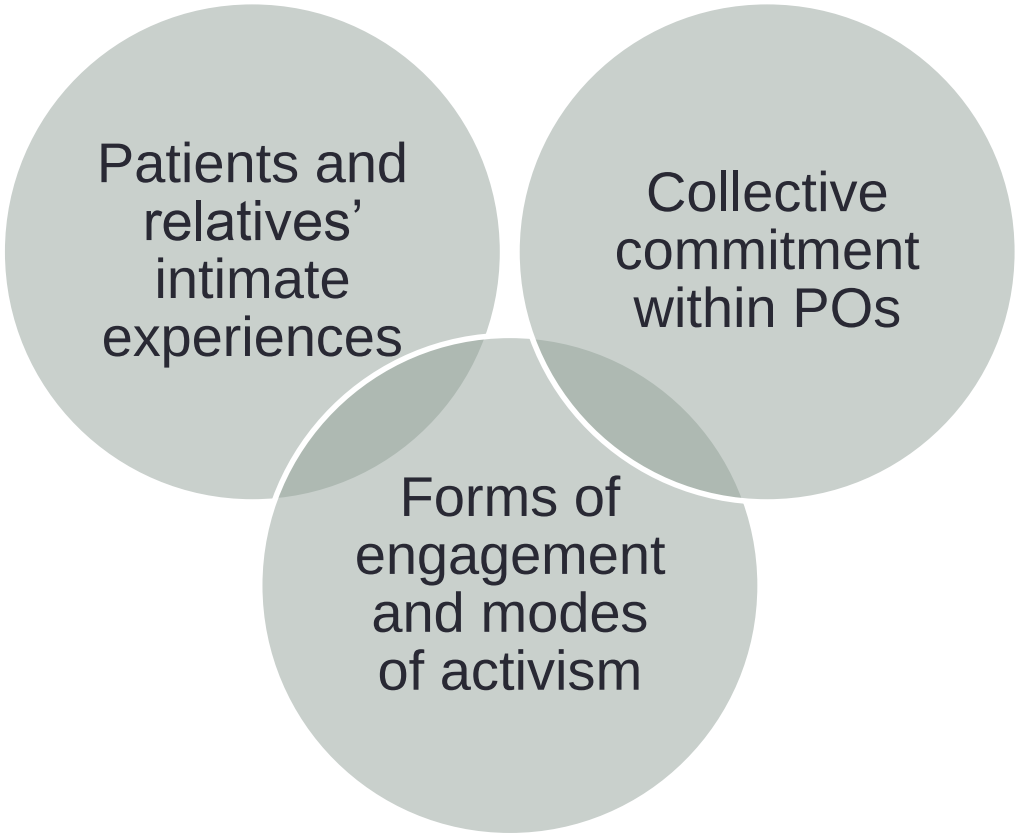
- **Collaborative forms of production, validation and circulation of knowledge**
  - Scientific researchers, medical experts, patients and relatives, patients’ organizations, etc.
    - > **“Hybrid collectives”**
  - // Laboratory research because the multiple types of knowledge produced by the collectives are complementary rather than contradictory, as well as not intrinsically different
    - > **Hybrid bodies of knowledge**

- **What are the forms, processes and implications of such collaborative projects?**

Research context

A **multi-sited ethnography** which explores the field of patients' organizations (POs) concerned with genetic disorders in French-speaking Belgium

- **Narratives from engaged patients and relatives**
- Participant observations
- Online and offline documentary research



Narrative of a “wild” search for causation

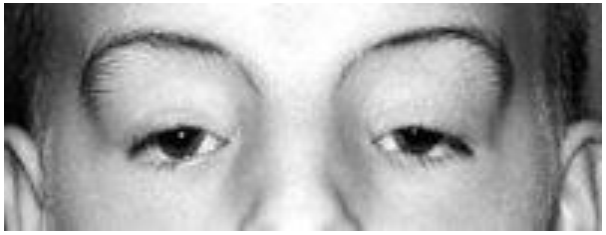
“Our daughter was born 19 years ago, in April. During her first year, our paediatrician told us that some babies’ evolution were slower than other ones. But she was not worried about our daughter. ***Yet we were realizing that something was getting wrong****, because we have 2 older daughters.* ***We knew this was something else*** *[…] When she was 9 months old, we were getting more and more angry and* ***we decided to do something.****”*

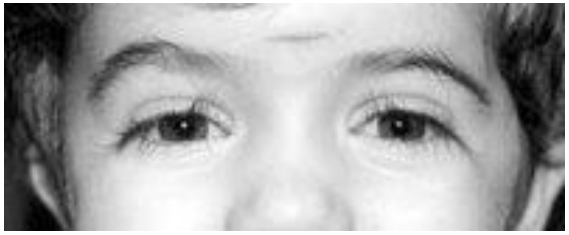
Narrative of a “wild” search for causation

“We consulted a geneticist. She told us that she could indeed notice some abnormalities, but that ***she did not know anything about them****. She told us: ‘I assure you, I attend many international conferences, I travel a lot… Believe me that maybe in 2, 5, 10 or 30 years we will be able to name your daughter’s disease, but nowadays we do not know anything about it.’* ***The disease did not exist yet****. Well, actually, it had already been identified in Japan, but it had not reached Europe yet.* ***It was not diagnosed yet****.”*

Kabuki/Niikawa-Kuroki Syndrome

- **A rare genetic disorder** (developmental delay, distinctive facial features, skeletal abnormalities and intellectual disability)
- Causation: mutations in the KMT2D/MLL2 gene (Ng & al. 2010) or the KDM6A gene (Lederer & al. 2012)
- Prevalence: 1/32 000
- Age of onset: newborn
- Diagnosis : **patient history and identification of the characteristic signs > clinical genetic testing to attest the certainty of the diagnosis of Kabuki Syndrome**





- Source: http://www.orpha.net/

Narrative of a “wild” search for causation

“Really, in our search for causation, ***we were lucky to hear via the French organization [PO] that some French geneticists were launching a research project about Kabuki Syndrome.*** *A blood test was done on Louise in Brussels, which was sent to Decker hospital in Paris. Our geneticist also asked us whether we agreed that she realized some other blood analyses in Brussels. We answered: of course!* ***Anyway, we were ready to attempt everything.”***

Search for information // Search for causation

⟹ **Issues of identities**

Narrative of a "wild" search for causation

"But Paris told us that Louise was not affected by the syndrome caused by the defect on MML2, and so that they did not understand […] ***Then, thanks to Louise, they were finally able to name the responsible gene: KDM6A****. They started searching for other similar cases. No one was found out in Belgium. But via some Italian and Swiss analyses, they were able to find out 3 new cases.* ***Results were presented at international level and got approved by the international scientific committee****."*

Deletion of KDM6A, a Histone Demethylase Interacting with MLL2, in Three Patients with Kabuki Syndrome

Damien Lederer[5], Bernard Grisart[5], Maria Cristina Digilio, Valérie Benoit, Marianne Crespin, Sophie Claire Ghariani, Isabelle Maystadt, Bruno Dallapiccola, Christine Verellen-Dumoulin

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Narrative of a "wild" search for causation

"I have some contacts, but honestly, I do not have strong knowledge in the biomedical field. And actually, I'm not seeking for it. Because at least, for us, it does not really matter. ***What matters is to know that [the cause of the syndrome] comes from a genetic accident and is not hereditary, does not stem from parents' responsibility****. And this is something that parents have to be aware of."*

- A success story of a (re)search for scientific causation or strong evidence, but not the end…

⇒ **Multiple understanding of evidence and diagnosis** (Jutel & Nettleton 2011; Brown & al. 2011)

⇒ **Production of hybrid bodies of knowledge and identities which are put into circulation or *enacted*** (Mol 2002) **through patients' organizations' knowledge-related activism** (Akrich & al. 2013; Rabeharisoa & al. 2014)

Radical openings in technoscientific practices?

- **Continuous processes of co-production, -validation and -circulation of hybrid bodies of knowledge and identities** through the interactions between the diverse involved actors

  - Entanglement of **intimate and collective, technoscientific and social, matters of concern and practices**
  - Entanglement of **sites**
  - Entanglement of **temporalities**

THANK YOU FOR YOUR ATTENTION!

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