

DIRECT-TO-CONSUMER GENETIC TESTING: USERS' PERCEPTIONS



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Direct-to-consumer genetic tests (DTCGT) represent a new online market, since their appearance in the early 2000s.

About 40 private companies worldwide offer this kind of services (about 700 different tests are available): the customer can purchase the tests online, within a price range of 60 to 2 000 Euros (about US\$ 80 to 2 800). [1] This entitles her/him to receive, by post, a test kit, allowing him/her to send back to the company's lab a sample of saliva.

After 6 to 10 weeks, the customer is informed via e-mail that the test's results are ready in his/her private account, on the company's website. S/he can then access online various information — depending on the precise package s/he purchased — that can be gathered under three labels:

- diseases susceptibility assessment (common multifactorial diseases, monogenic disorders, mental disorders, metabolic diseases);
- ancestry information;
- intellectual and physical performance.

Being part of the company's panel, the user may then be entitled to receive complementary results (sometimes free

of charge), when new tests are made available within the company's catalogue.

Thus, one can consider DTCGT as new "personalized genomics" devices. Worldwide, public health authorities and bioethics committees question the uses of DTCGT: healthcare systems have to face a novel situation, since they are currently regulating general use of genetic tests, but the DTCGT e-economy challenges national as well as regional attempts of control. [1] When investigating the broader context, one can observe the concomitance of two trends:

- ① an increased importance of informed consent in medical settings, linked with a growing tendency of health practitioners to encourage patients to rationalize their health decisions: thus, individuals are more and more empowered;
- ② the development of precise commercial strategies by DTCGT companies: emphasizing healthism and relevancy of claims for individual empowerment; encouraging curiosity for genomics; promising biosocial gatherings. [1]

Objectives

- > investigating users' motives for purchasing DTCGT;
- > investigating users' expectations and perceptions of DTCGT;
- > investigating the potential integration of DTCGT into itineraries of health management and identity quest;
- > examining the potential echoes between the companies' commercial strategies and users' perceptions.

Method

This study has been conducted through qualitative socio-anthropological methods, based on a grounded-theory approach [2]:

- simultaneous data gathering and analysis;
- follow-up of emerging themes through early analysis of fieldwork data;
- integration of these themes within a theoretical framework that broaches causes, consequences and conditions of the observed processes. [3]

The approach is thus illustrative and explanatory, but not quantitative: representativity is not a goal of representativeness, but rather "information saturation". This socio-anthropological approach is not focused on the sole content of users' views (as any questionnaire study could be), but rather on the building process of these views (and especially their connections with perceptions of science, family and identity).

Recruitment - Since the studied devices are purchased through the Internet, users of DTCGT do not represent a constituted and "easy to locate" social group. They are scattered all over the world; this leads to a necessary adaptation of the interviewees' recruitment protocol. Thus, online social networks have been used to locate users and standard snowballing method has then been applied (by means of informal contact between interviewees and other users).

> It is noteworthy that the term "sample" designates the specific set of interviewees or observed individuals. It does not involve any technical reference to sampling theory. Socio-anthropology works rather on networks or "tree structures" than on samples.

Data collection

- > through **in-depth semi-directive interviews** with users of DTCGT (N = 20 as of March 2011, targeted N being 30 for the exploratory phase);
- > through **analysis of relevant websites and forums.**

Interviewees (as of March 2011):

- Country: USA (5), UK (2), France (5), Germany (3), Portugal (1), Switzerland (1), Canada (1), Netherlands (1), Italy (1)
- Gender: male: 12; female: 8
- Age range: 26-63

Results presentation

The exploratory interviews of this research reveal several major poles within users' perceptions of DTCGT. They represent analytical paths, to be followed during the coming post-exploratory phases of the research. The present results are thus a first analytical approach of the data and aim at stressing main emerging research questions.

Different motives, different user's profiles

- Several interviewees share an intense interest for genealogy and history. They present this interest as the main reason leading them to order a test.
- Another group of interviewees is characterized by a professional involvement in ICT activities (communication, e-marketing, e-commerce, software development, etc.). Among them, several informants emphasized that they were not at all interested in health or ancestry information, but rather in the very

- process of getting tested and obtaining a new type of data: genomic information.
- A third group, less "typical" but significant, is made of groups 1 and 2 members' relatives and friends, who have been encouraged to take a test or who have been presented with it. Several of them do not express a strong interest for DTCGT but rather a will to please the suggesting family member/friend.

- **Could users' motives be the basis of a relevant profile typology?**
- **How could the potential intersections of profiles be described?**

"What's health got to do with this?" - DTCGT and medicine

■ "Health packages"

DTCGT use biological lab technologies that are also used by medicine. Thus, they are often tackled by authorities and literature as potentially medical devices. Their status is different in our interviewees' views, though. Most of the informants who purchased health tests packages made this decision for financial reasons: a "special offer" made a set of tests available for a low price, and included health tests. About half of the interviewees declared having no anterior interest in health information, but got a "health package" as a "bonus" to the ancestry tests they were interested in. Thus, potentially sensitive health information is made available without clear informed consent process (see for example [4]).

- **Do many users' views "disconnect" genetic testing and medical information?**
- **How do they experience accessing this information about disease susceptibilities?**

■ DTCGT and medical diagnosis/research

Some of the tests results state individual genetic susceptibilities for specific diseases. This can be experienced as a medical diagnosis. In users' discourse, statistical risk indeed is often framed individually (for example as a validation of one's family medical history, and not as a neutral figure). It can thus lead to modifications of their health behaviour (diet, physical activity, for example). Besides, the companies' marketing stresses the interest for users to get involved in a new kind of research [1] but this does not echo so far the main concerns of our interviewees.

- **What leads users to tackle DTCGT results as medical diagnosis?**
- **Do users change their lifestyle on the ground of the tests' results?**
- **Do users consider the DTCGT companies' activities as research activities?**

Perceptions of medical risk and figures

A relative risk is given to the user, for every tested trait or disorder. The result is generally expressed within 3 categories: “decreased”, “standard” or “elevated”.

Thus, the results can present an “elevated risk” for a disease whose prevalence is very low, this leading to potential misunderstandings. For some users, health indeed appears as an acceptable deviation from a population genomics standard profile, and not a complex frame taking individual specificities into account.

➤ **What personal perceptions of risk are involved when accessing one’s results online?**

Origins & “Recreational Genomics” [5] - DTCGT and Ancestry

DTCGT companies also offer Y-chromosome to identify paternal descent, and mitochondrial DNA test to identify maternal descent. This ancestry information is then displayed (see screen capture) as a map or as percentages of different ethnic backgrounds. This is a problematic issue since this type of results conveys a sort of “ready to think” view of one’s origin, to users who often are in search of a lost (or fragmentary) family memory. Interviewees described various combination processes between DTCGT results and previously known genealogical data or family archives. Some consider the test’s results as a confirmation of their family history, some as a real blurring of their own identity.

- **How do users connect their existing genealogical knowledge with the ancestry information provided by the tests?**
- **How do users deal with non matching family history / test results?**

Curiosity and “Early Adopters” - DTCGT and ICT milieu

For some interviewees, who defined themselves as “early adopters”, DTCGT is a new online technology they are eager to test and comment. It is part of a broader whole that includes other aspects of their online activities (social networks, regular blogging, etc.). Thus, the produced data and information may be treated as other personal digital information: individuals tend either to openly display them (sharing in specialized forums or even publish raw DNA sequence under a Creative Commons Licence) or to protect them carefully (using pseudonyms when ordering the tests).

- **Is sequenced DNA part of one’s digital identity?**
- **Can one talk about a “DNA trend” in the ICT milieu?**

Diversity of legal contexts and impact on users’ perceptions

While, in the USA, DTCGT market is not regulated as a medical field, is it targeted by the Food and Drug Administration since mid-2010, when a DTCGT company announced its intention to sell tests over-the-counter. [7] A dedicated FDA meeting is planned for March 2011, in order to examine the applicable regulations — especially regarding the potential mandatory involvement of a physician in the DTCGT process. The paternalistic GP character and the threatening shadow of health insurance companies lead many of the American interviewees to be very cautious regarding the disclosure of their test results.

In France, the law (*Civil Code*, Article 16-10, alinéa 1) states: “*The examination of the genetic characteristics of an individual can only be undertaken for medical purposes or scientific research.*” Selling DTCGT on French soil is thus prohibited. In this context, even if no user has been prosecuted, the French interviewees are concerned about possible legal consequences of their online purchase.

- **What is the impact of various legal environments on DTCGT users and users’ profiles?**

(e-)Biosocialities? - DTCGT & Social Interactions

■ **Electronic biosocialities**

The sharing of DTCGT users' experiences has resulted in the appearance of virtual bio-communities, where users may search for fellow creatures with similar genetic profile. Thus, these sociality phenomena are based on a shared biological condition and are called biosocialities. [6] Interviewees mentioned these communities as "genetic Facebooks" where they could "meet" people. But it is noteworthy that none of the interviewees actually decided to organize a face-to-face meeting with relatives found through ancestry forums or with individuals sharing genomic specificities.

- ↳ **What kind of long-term social interactions occur in these virtual communities?**
- ↳ **Do these biosocial phenomena have "palpable" consequences in real life?**

■ **DTCGT & "real life" interactions**

DTCGT do not seem to create non electronic strong ties between strangers. But they appear to be a sensitive family discussion subject. Several interviewees mentioned having discussed DTCGT before or after having purchased their own kit. Some even bought supplementary kits to get some of their close relatives tested. These situations lead to a common re-examination of the familial past, to the exposition of the family members' views regarding genomics and sometimes to the disclosure of previously hidden information.

- ↳ **What are the stakes of familial discussions dealing with DTCGT?**
- ↳ **Do DTCGT results have an impact on familial historiography?**

Conclusion

This exploratory phase has shown a complex entanglement between users' perceptions of scientific knowledge and technology, identity and health.

Their motives to get tested are heterogeneous and their apprehension and understanding of the results is shaped by these very motives and perceptions.

Secondary phase (to be conducted until Summer 2011) will broaden the explored themes and investigate the dialectic links between autonomy of choice and access to genetic knowledge, consumerism and "healthism" (health considered as the most important value), personal risk management behaviours

TeGALSI research project (Tests Génétiques en Libre Accès sur Internet,

i.e. French for "Direct-To-Consumer Online Genetic Testing") has been selected within the framework of a call for proposals published by the French Institute for Public Health Research (IReSP).

- > The project will be conducted during the 2010-2013 period.
- > Dr. Pascal Ducournau (Senior Lecturer in Sociology, currently at U558-Inserm, Toulouse, France) is the scientific director of TeGALSI.
- > The project also involves researchers from:
 - Copenhagen University (Denmark),
 - Lancaster University / Cesagen (UK),
 - Andalusian School of Public Health (Spain),
 - Katholieke Universiteit of Leuven (Belgium).

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