



Génome en Poche

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A team of molecular biologists and computer scientists from Bordeaux has recently invented an application that will enable anyone to carry its genetic code in a smartphone, keeping up with much information concerning their health status.

Shortly, smartphones and tablets owners will be able to carry around their genetic code everywhere, as they already do with their favorite songs or holidays pictures. The application "Portable Genomics" already exists as a prototype for Apple's iPhone and iPad. It is the work of a small informal team located in Bordeaux, gathering geneticists and computer specialists. So far, they had been working on a voluntary basis.

Patrick Merel, the project initiator, is a molecular biologist working as a researcher in a famous hospital in Bordeaux. He dragged in this adventure two biologist colleagues and Macha Nikolski, researcher in bio computing at the CNRS and at the LaBRI (Bordeaux Laboratory in computing research). The iPhone application has been programmed by one of Mrs. Nikolski's PhD students.

The power of new smartphones and notebooks now makes possible projects that were unimaginable only a few years ago. In computer language, the human genome –composed of three billion base pairs – only represents a three-gigabyte file. And this is lighter than a movie on a DVD, only 10% of the new iPhone storage capacity. Moreover, several US companies on Internet put forward the human genome sequencing, without administrative procedures or prescription. The customer simply needs to

spit out in a tube, send it through parcel post and pay a few hundred dollars. The laboratory extracts the DNA from the saliva, sequence it and analyze a part of the customer's genome, selecting markers corresponding to the most desirable information.

A few days later, the customer receives via Internet tons of information on his predisposition to various diseases, his ethnic and family origins, his physical and psychological attributes. The customer can also download his genetic code in its raw data to run his own research, or to entrust it to an association, a school, a group of researchers...

Progress is so fast that the complete human genome sequencing, which used to cost hundreds of thousands dollars a few years ago, will soon become affordable: \$50,000 in June 2009, \$15,000 a year later and probably \$3,000 in 2011, maybe even \$100 by 2016... In theory, this industry is illegal in France, but facing Internet, authorities are powerless.

Requirements are satisfied for the advent of "portable" genomics. As says M. Merel: "Medicine is about to fall radically in a new world, the one of in vitro and in silico molecular diagnostic". His ambition is to appeal to the general public, passing representative bodies (**). Faced with sustainers of a permanent administrative control on these eminently confidential data, M. Merel refers to a superior rule: "The inalienable right for every citizen to keep the ownership of their genetic code".

For an iPhone or iPad owner, the application Portable Genomics will be very easy to use. Once the genetic code will be transferred on the device, the application is going to analyze and display these results as in the iTunes software, already used by hundreds of million people to organize their music and videos. If the user has a predisposition to diabetes, the icon "Diabetes" becomes red: to be taken into consideration. On the other hand, if there is no particular risk of developing cancer of the stomach, the icon stays green: no worries. When you press the icon, you get a list of genes involved in the diagnosis, statistics on the disease, how the calculation of risks was done, the symptoms description and so on.

Beyond that, M. Merel already imagines a wider range of services: the medical file storage in the smartphone, direct access to Internet web-sites dedicated to various diseases, genetic data sharing with networks of "citizen research", reminders and alarms for patients who need to have some tests performed or need periodical treatment. Furthermore, by automatically connecting to Portable Genomics' mainframe, the smartphone will continuously integrate every new discovery on the human genome. Thus, this will enable refining or revising diagnostics. The user will never be truly at peace. Indeed, the green icon (reassuring) can turn red anytime because on the other side of the world, a geneticist has just discovered that the appearance of a cancer of the stomach can be influenced by a genetic combination, hitherto not discovered. On a different note, the analysis of the Y chromosome, which every man inherits from his father, can suddenly reveal family secrets well kept until now...

M. Merel also wishes to use the GPS in smartphones for patients geolocalization. If necessary, during a trip for instance, the system will be able to give the customer a list of specialists in his disease, in his neighbourhood. "The patient will go to the doctor's with his genetic code in the pocket and they will look at the analyses together". M. Merel also plans to use "Medical doctor ranking websites", which rate doctors on a scale of 1 to 5. "In France, this will be hard to swallow, doctors will fight back but they will eventually get used to it".

Backstage, the system is quite complex. Mrs. Nikolski plans an increase in power/rise in different steps: "First, data analyses will be contracted out by external companies that will use mainframes and send results on the cellphones. But on the long run, the goal is to develop algorithms which will make possible the data analysis directly on the cellphone. The user will be completely autonomous". If everything goes well, Mrs. Nikolski would like to formalize her collaboration with M. Merel: "It's a nice journey, if it works, I'll be diving in". Her only concern: "Are people ready to welcome such a radical innovation? Americans will probably be found of this kind of service. Whereas the French, maybe in a different style or in a few years only..."

In fact, France does not seem to be in a good mood to let go with it. A spokesman of the French National Consultative Ethics Committee reminds that the real problem is far upstream. Since in France it is forbidden to order a genetic sequencing without a prescription, it is natural that services formatting these data are forbidden too. Furthermore, he warns potential customers who will walk around everywhere with their genetic code in their pocket that they might be victims of hacking, or forced to show their data to their relatives or to an employer, without their consent.

Nevertheless, M. Merel long kept hoping that he could develop his invention in France, despite constraints set by the French legislation, very strict in terms of bioethics. In 2009, he submitted his project to OSEO (public institution in charge of supporting innovation and the growth of small businesses): “I had been really careful in drafting the project. On the European market, genomic data would be shown only to the general practitioner. For the rest of the world, users would have a direct access to their data”.

Despite his precautions, OSEO refused the project. “I’m sure that it is because of the bioethics implication that bothered them, even if they did not clearly want to say it. Instead, they preferred telling me that my idea was not innovative – whereas nowhere, something similar exists. Then they clearly suggested and encouraged me to start it in the US”. M. Merel also discovered that when OSEO rejects a project, it shuts every other doors. “Useless to call upon the regional council, agencies dedicated to start-up support, or innovation contests. The same thing happened with the INRIA. They refused to support him. He admits that he is a bit embittered: “The bioethics of the French legislation strangles research. It will eventually disappear but we will have fallen behind against fierce competition from other countries that get less in the way of researchers”.

M. Merel grew tired of waiting and decided to follow OSEO’s advice: put forward his project to the United States. In May 2010, he went to a conference on “Genomic Information Alliance”, held in Seattle by the genetic company Illumina, in which participated representatives of Google, Amazon and Microsoft. “They allowed me to present my project. I insisted on the fact that it is necessary to develop clinical use of wireless technologies and to entrust genomic data to the general public”. They understood right away his innovative character and accepted him, with no criticism or reluctance.

In October, M. Merel came back to the US. But this time he went to San Diego, California, major worldwide center of biotechnology, where he had already lived in 2005 as a researcher. During two weeks, with the help of a few American friends, he founded the company Portable Genomics, started to meet venture funds and initiated patent application. He even made contact with some people in order to present the project to Steve Jobs himself, Apple’s leader. “If he likes our project, we will be catapulted on the personal genomic trade in the wink of an eye”.

Back in France, he applies for a working visa in the US as an entrepreneur, which compels him to deposit \$150,000 on the bank account of the society. “With my hospital researcher salary, it has been a hard time. I had to borrow money from my friends and family. I didn’t want to invite foreign investors to invest in the project at this stage”. M. Merel already plans to hire nine people – a few are French – if they are willing emigrate.

(*) Kindly translated by C.M.

(**) Portable Genomics did not caution this sentence.