Roberto Carlini, Federico Carlini – Genesy Project Stafano Dalla Palma – JANS Academy of Data Science Remo Pareschi – University of Molise

GENESY

The blockchain of evolution

background



Recent technological advances, coupled with changing patient needs and increasingly obsolete business models, are compelling the pharmaceutical industry and global healthcare systems to innovate through increased R & D efforts, acquisitions and / or new partnerships.

The market of **Direct-to-Consumer genomic data services** is evolving towards new technologies and business models, thus providing a scenario rich of important opportunities for innovative solutions in the domain of genetic sequencing and IT platforms.

opportunity

In the future, health care will increasingly be based on personalized medicine. Risk factors and strengths can be identified in advance and prevention, diagnosis and treatment of diseases can be targeted to have the best effect on each individual. With regard to the general economic effects, this means significantly improving the productivity of health care and social well-being.





Down-spiralling of sequencing costs

The first human genome was sequenced in 2001 at a cost of \$ 3 billion. Today, the complete sequencing (WGS) of the human genome costs less than \$ 1000, by using low-pass sequencing technologies, \$150 or less.

Therefore, complete sequencing of the genome will soon be widely adopted as it allows for broader analysis, disease prevention and personalized therapies.

Genesy general design

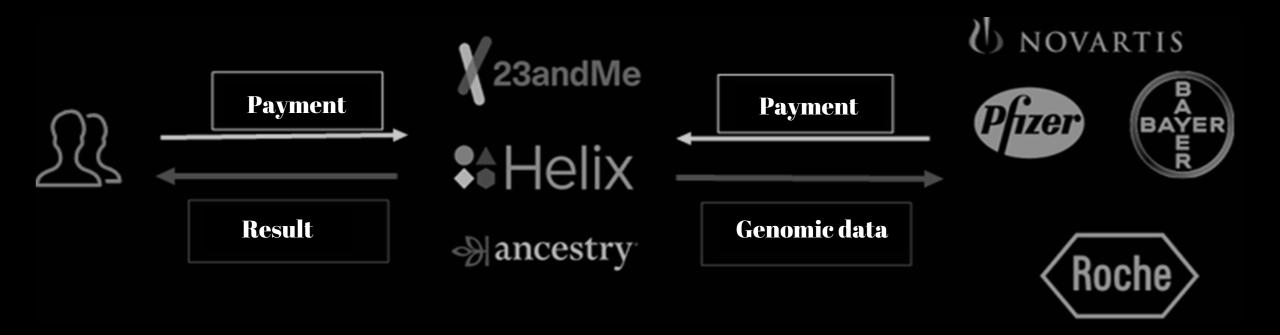
Genesy sequences DNA at various levels (complete genoma and genetic panels) in Italian and US laboratories through Illumina medium to high-end machines.

Users can share their information, if they wish, through controlled access mechanisms and download their results locally from Genesy and an app will let them analyze and interpret their genomic data in complete autonomy

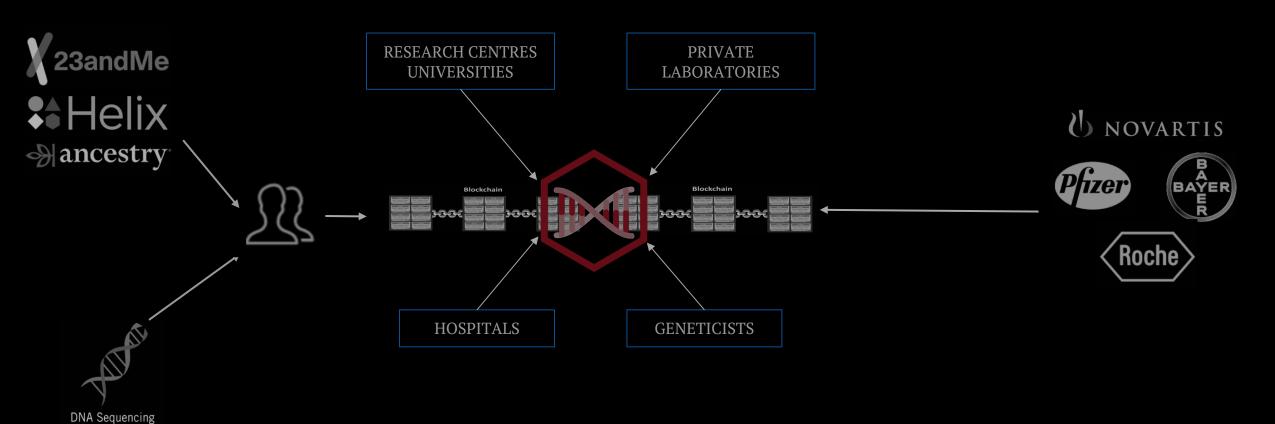
Access to data from either users or professionals or academic or pharmaceutical facilities are enabled, with identity management guaranteed by dual-key systems and cryptographic functions.

The Genesy token utility based on the Stellar platform supports the sale/purchase of data and facilitates the monetization of the Genesy services.

the traditional business model



the GENESY ecosystem



GENESY operates as a gatekeeper and manager of the ecosystem, ensuring property and exclusive user access to genomic data

Services

ethical vision

Genesy aims to educate consumers to better understand test results in order to make informed decisions and support the human rights approach to data sharing, in conformance with the indications of the GA4GH (1) for the responsible sharing of genomic data, that must be conducted so as to minimize damage and maximize benefits not only for those who contribute their DNA, but also for society at large and for health systems as a whole. Ultimately, Genesy plans to build a new genomics ecosystem based on the belief that all people should:

- have possession of their genomic data
 - have direct access to their platform
- be in control of the access by third parties to their own data
 - be sure that their genome is safely stored
- be able to leverage their genomic data to improve their future health
- have the opportunity to anonymously donate their data for the public good
 - be able to benefit economically from the use of their genome by third parties

(1)The Global Alliance for Genomics and Health (GA4GH) is a policy-framing and technical standards-setting organization, seeking to enable responsible genomic data sharing within a human rights framework. Broad Institute - National Institutes of Health (NIH).

users in control of their privacy

Users can manage their privacy (partial or total). This is an essential element of the Genesy business model, as research bodies may be interested in contacting user categories for in-depth studies. These studies could represent additional revenues for users, who would thus be encouraged to share their information

This business logic is implemented through a process that guarantees to all the participants of the ecosystem the absolute transparency of the procedures and the privacy and ownership of the circulating data.

The regulation of the information procedures on the processing of personal data will be provided in a document that must be signed by the user via secure e-mail systems.

privacy and treatment of genetic data guidelines

data acquisition will be in accordance with European regulation (GDPR)

the right to data erasure (Article 17 of the GDPR) will be ensured through the hash-off approach, made possible by the fact that genetic data are kept off-chain in the cloud, and are therefore erasable, while the blockchain maintains the metadata of the owners of the data

DNA management on a blockchain platform

Immutability

Security

Transparency

Non-duplicability



technology



The Blockchain solution offers significant advantages over classic centralized databases:

Disintermediation: direct management of one's own data

Privacy: it guarantees maximum protection of one's own identity

Security: data cannot be modified or tampered with

Permanent register: structured and time-stamped data

Decentralization: no access point to hackers, no central authority

Smart conytracts: transactions take place automatically via predefined agreements

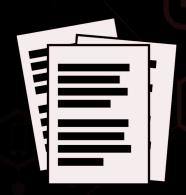
Traciability: consensus mechanisms allow transactions to take place without errors

components of the platform

Hyperledger **Blockchain** (Linux Foundation) for the management of transactions and of user metadata, integrated with:

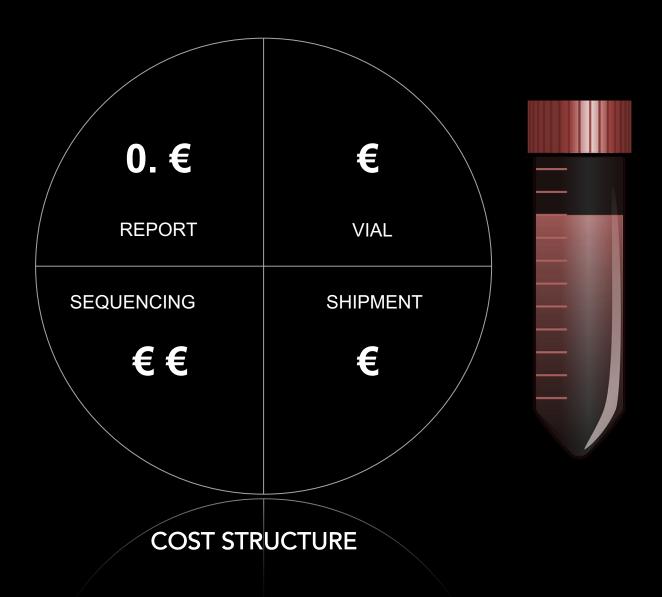
- NoSqL Database for the analysis and management of sequenced DNA data
- IBM Watson as a tool for creating reports by interpreting the produced data
- Cloud Object Storage for storing off-line genomic data
- Stellar (XML) digital currency for the management of the sale/purchase transactions within Genesy





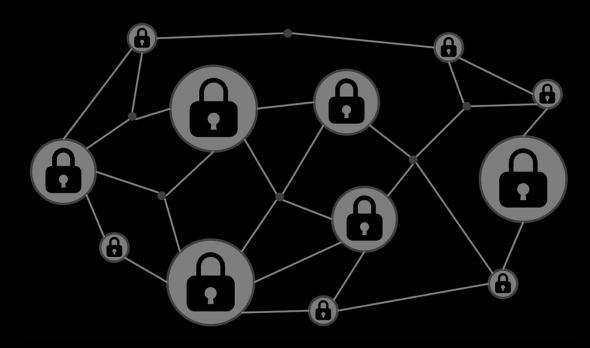






interfacing the physical and the digital world

- Through a small, light and thin vial that will preserve dry saliva intact for months with reduced logistics costs
- Delivered to the customer in a simple personalized packaging with the Genesy brand
- The vials are regularly insured and sent by batch to the sequencing centers, further containing shipping costs
- Users receive results within a few weeks
- Users are regularly notified about the possibility of acquiring new reports applied to his DNA, to be produced on a par with the development of new genetic panels and related medical research progress.



results so far

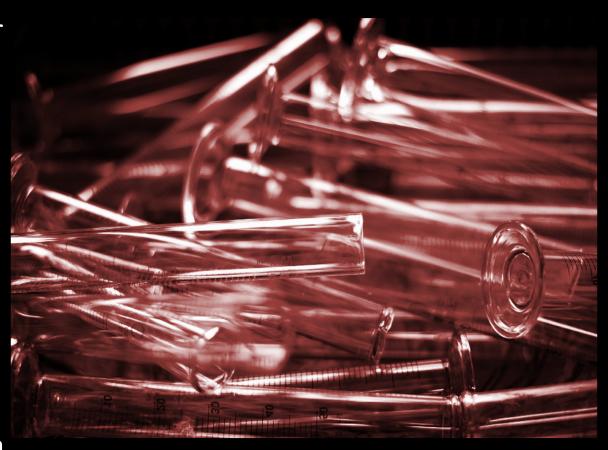
- implemented dedicated blockchain platform on Hyperledger Fabric
- Development of chaincode (smart contract) for sequencing data
- Storing off-chain files in the cloud
- Completed the pipeline (in the cloud) that uses raw DNA data to create reports for users
- Created the first reports, highlighting the first somatic traits and predisposition to certain diseases and neurological conditions

WHAT'S GOING ON

Rapid completion and testing of the platform for graphical and tabular management and visualization of the user's genomic information

Creation of a network that includes "interested" geneticists and researchers who can contribute with evaluations or suggestions to the construction of the medical / scientific framework to the project

Implementation of an internal "encyclopedia" based on the most important and reliable genomic databases, which will allow us to automate the preparation of our reports through the use of new DNA sequencing algorithms and their impact on research





team

We are structuring and expanding a team of both blockchain/big data/Al developers to manage the development and maintenance of our platforms and a team of geneticists and consultants, mostly of university extraction.

We are partnering with specialized centers, health facilities and academics operating in the field of human genetics and DNA sequencing.

The Genesy philosophy in one motto:

OWN WHO YOU ARE!

THANK YOU!