DNA digest

open access and secure data digest

Open Data on the Web
Fiona Nielsen, April 24, 2013

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DNAdigest enables data access

- DNA sequencing
- Genome research today
- Why genomics data is not open
- How DNAdigest will enable data access

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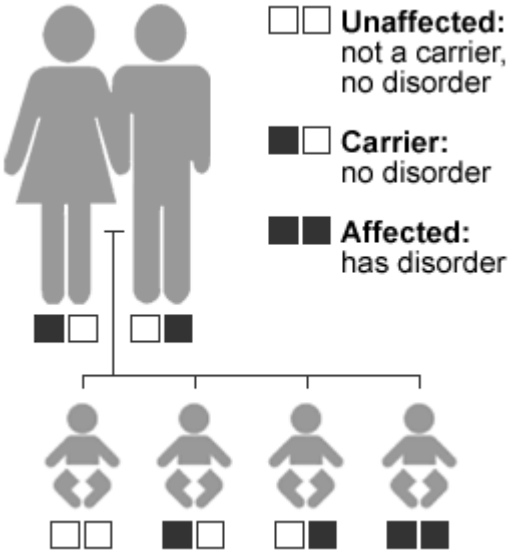
DNA sequencing

- cancer research
- heritable traits and illness
- rare diseases



images :http://www.pbs.org/newshour/rundown/2012/10/new-rapid-test-can-diagnose-babys-genetic-disorder-in-days.html
and http://www.nidcd.nih.gov/health/hearing/pages/usher.aspx

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New technology, new opportunities

The technology is developing fast, promising a **Genomic Revolution**,

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New technology, new opportunities

The technology is developing fast, promising a **Genomic Revolution**,

But the research progress is not as fast as it could be...

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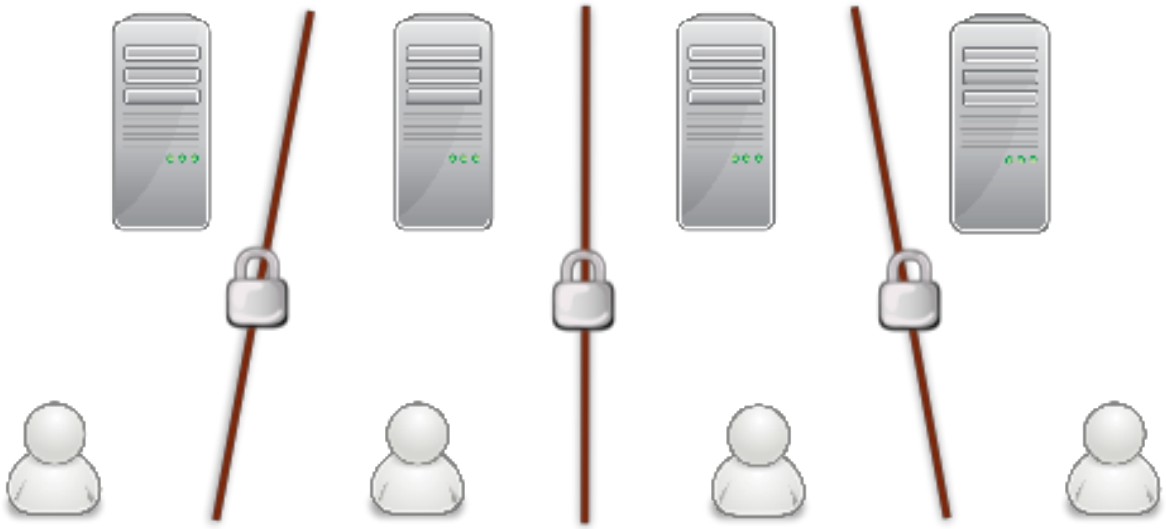
New technology, new opportunities

Why is research lagging behind?

- Because researchers are not sharing all their data!



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Genome research today

the patient

the researcher

the data

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Genome interpretation is hard

To make sense of his data, the researcher needs more samples for comparison

- He will look in databases of genomic variation
- He will search in literature describing the disease

But still the common case is a large number of ***variants of unknown effect***

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More data needed to validate results

At the same time, other researchers around the World are facing the same problem:

- They want to learn from other data sets
- But they do not make their own data openly accessible

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Why is genomics data not shared?

Data sharing is a problem:

- medical data confidential
- ability to identify individuals from genome data
- bulky data sets

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Current practice for sharing

Patients give consent for their data to be published and shared only if:

- de-identified
- aggregated in research result

Current practise is to publish results in scientific journals, **no open sharing of raw data**

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How to open up access to the data?

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How to open up access to the data?

Solution:

Not all research questions require access to entire data sets

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How to open up access to the data?

Solution:

Not all research questions require access to entire data sets

Example:

Does this mutation occur at higher frequency in cancer than in healthy samples?

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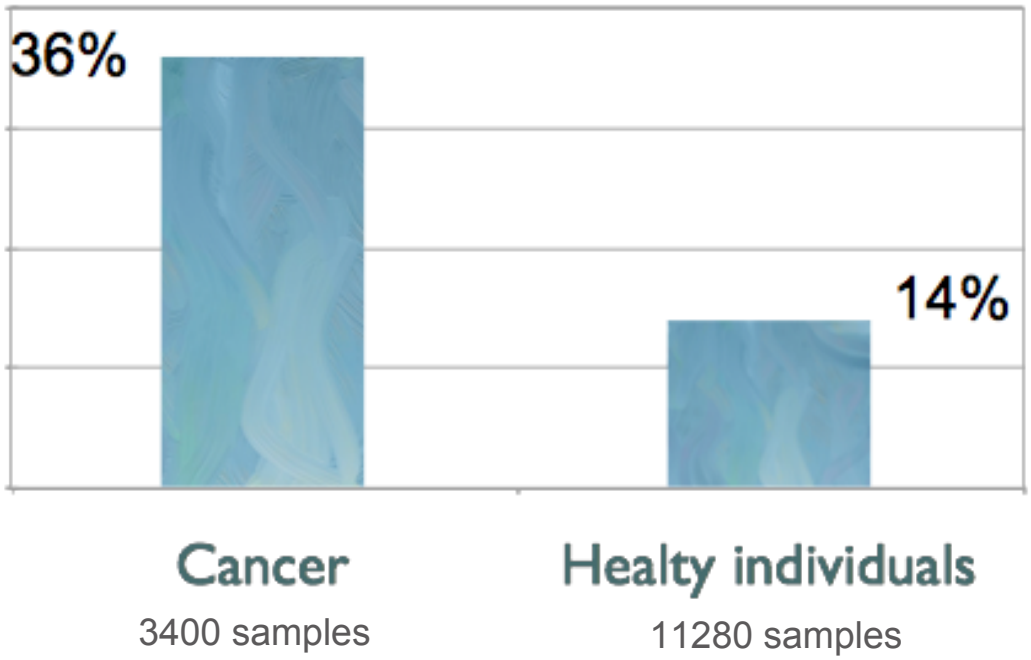
Example Use Case

"What is the frequency of this mutation in disease samples versus reference samples?"



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DNAdigest allows data access

Advantages:

- Anonymisation by aggregation
- Access can be made open for all users
- Queries for direct hypothesis testing

Challenges:

- Connect existing repositories with the DNAdigest API
- Create incentives for institutions to connect
- Build sustainable business model

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DNAdigest needs your help

We propose a solution, but we need your help to make it happen:

- Inform patients that their data is not shared sufficiently today
- Encourage researchers to share their data
- Support incentives that promote data sharing

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Summary

DNAdigest will allow interactive queries, collecting data from multiple repositories with results presented aggregated and anonymised

DNAdigest is a not-for-profit organisation, founded for the purpose of enabling secure open access sharing of genomic data.

Please visit us at DNAdigest.org

Thank you!