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AmbryShare: A Model for Sharing Genomic Data to Understand all Human Disease

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It’s old news; How is AmbryShare different?

Let’s get personal; Why share genomic data?

The challenges ahead; Ok, so what’s next?

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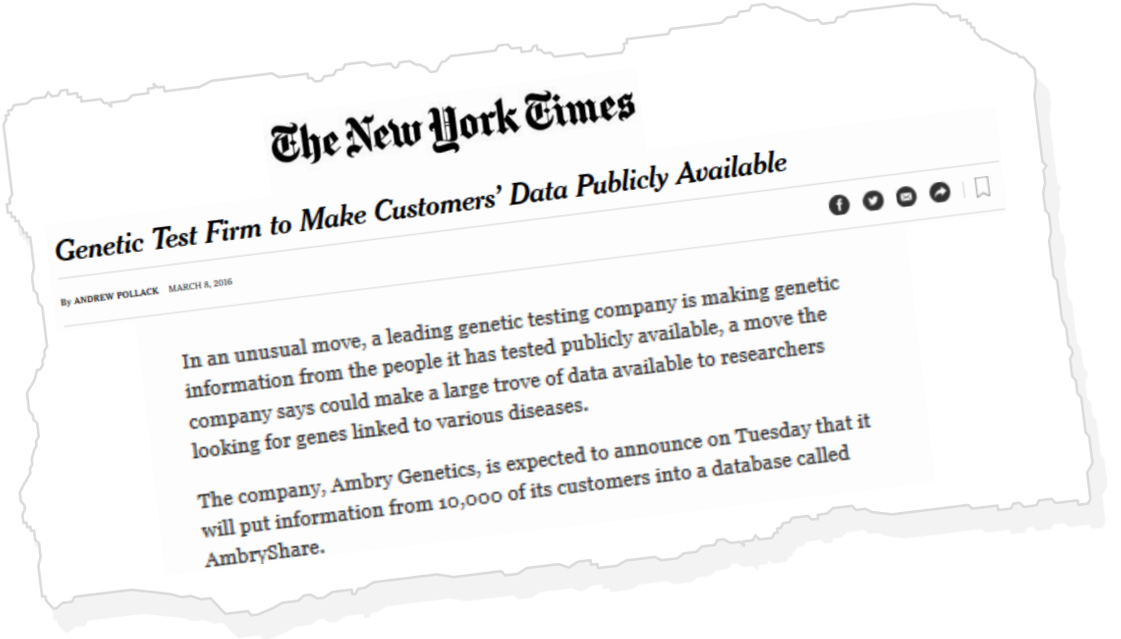
Excerpt from
March 8, 2016
NY Times article:

Charles Dunlop, founder and chief executive of Ambry, said he was approached by drug companies, but decided to make the company's data freely available to expedite research.

"I've got Stage 4 cancer myself," he said, referring to advanced prostate cancer that is in remission. "I don't want to wait an extra day."



The New York Times

Genetic Test Firm to Make Customers' Data Publicly Available

By ANDREW POLLACK MARCH 8, 2016

In an unusual move, a leading genetic testing company is making genetic information from the people it has tested publicly available, a move the company says could make a large trove of data available to researchers looking for genes linked to various diseases.

The company, Ambry Genetics, is expected to announce on Tuesday that it will put information from 10,000 of its customers into a database called AmbryShare.

REUTERS

CBS

FierceMedical Devices

The Boston Globe

FAST COMPANY

NBC

YAHOO! NEWS

Pharmaceutical Executive

Forbes

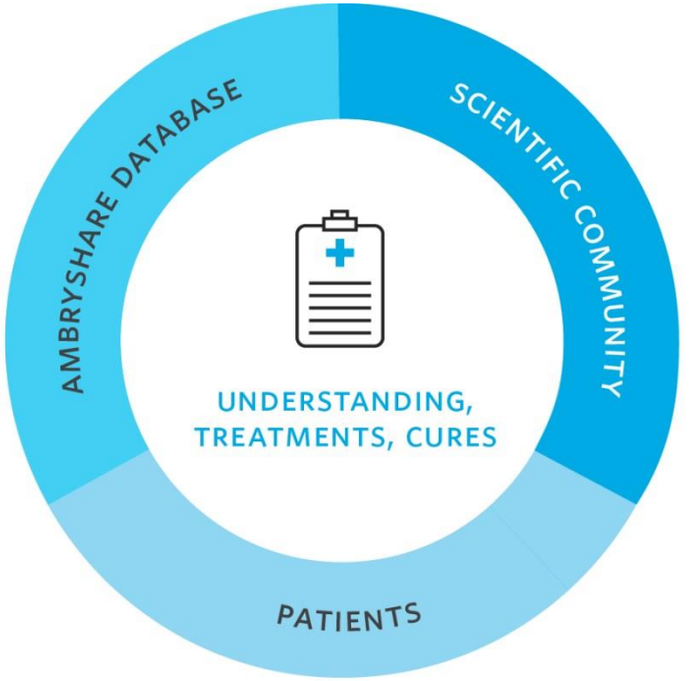
FORTUNE

nature

Ambry Share™

The world would be a better place if all human disease was understood.

AMBRYSHARE DATABASE
One of the largest allele frequency databases with anonymized aggregate human genome (exome) data on 10,000+ patients with disease.

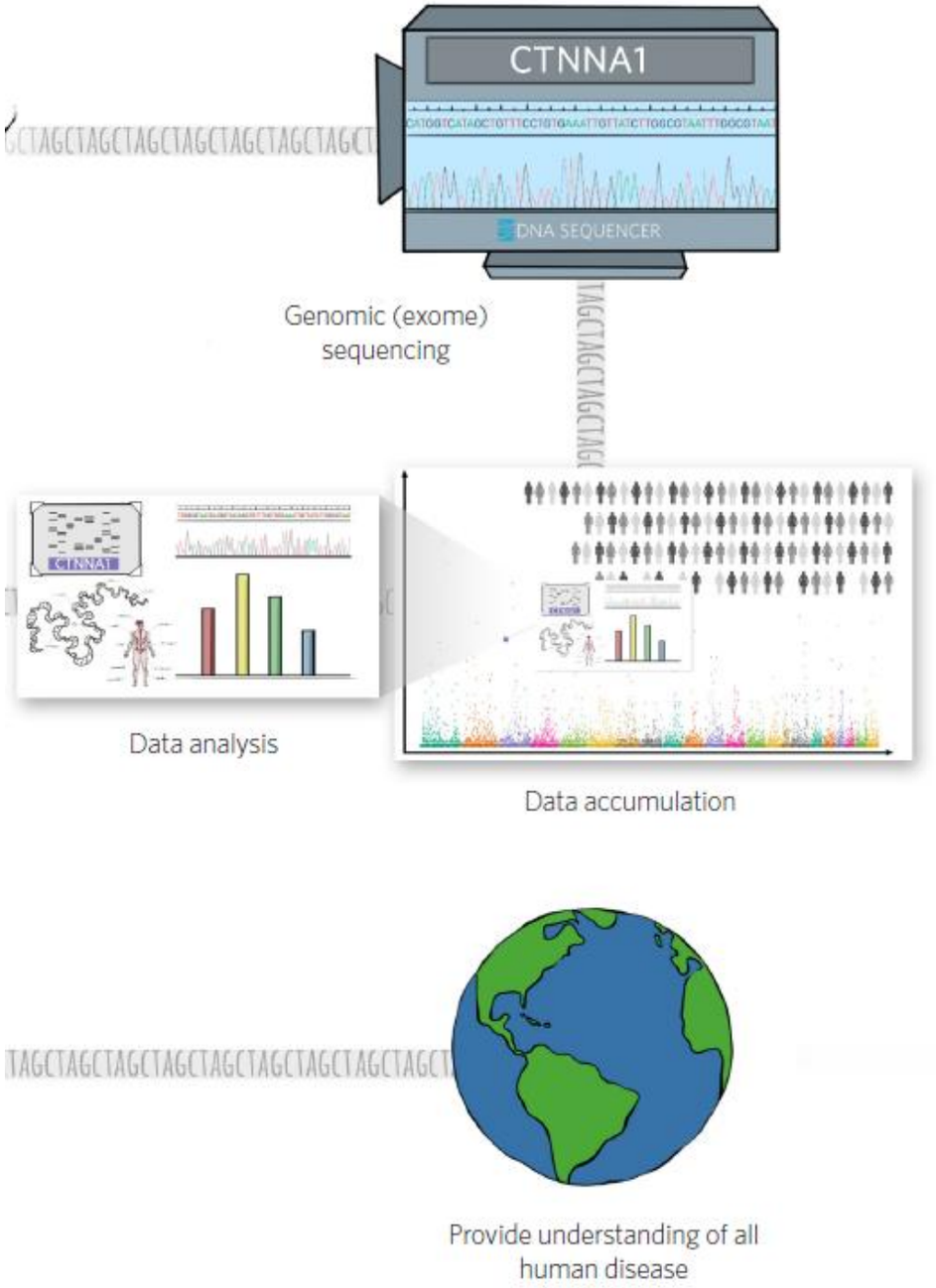


SCIENTIFIC COMMUNITY
Free access to all aggregated variant data from the sequenced cohort, by disease type, to continue to work on new breakthroughs, treatments and cures for disease.

PATIENTS
Patients can register with AmbryShare to receive updates, gain access to our family history tool, and take it to their healthcare provider to discuss if genetic counseling and/or testing is right for them.

What is AmbryShare?

- A free online genomic database of anonymized aggregated variant allele frequencies from exome sequencing of 10,000+ women with breast and ovarian cancer.
- A plan to expand to many other disease groups and exome sequence hundreds of thousands of individuals with patient engagement in the process.
- AmbryShare is designed to help scientists understand the etiology of disease and identify targeted treatment and disease prevention strategies.
- We hope researchers and pharmaceutical companies worldwide will access this data to accelerate the pace of research.



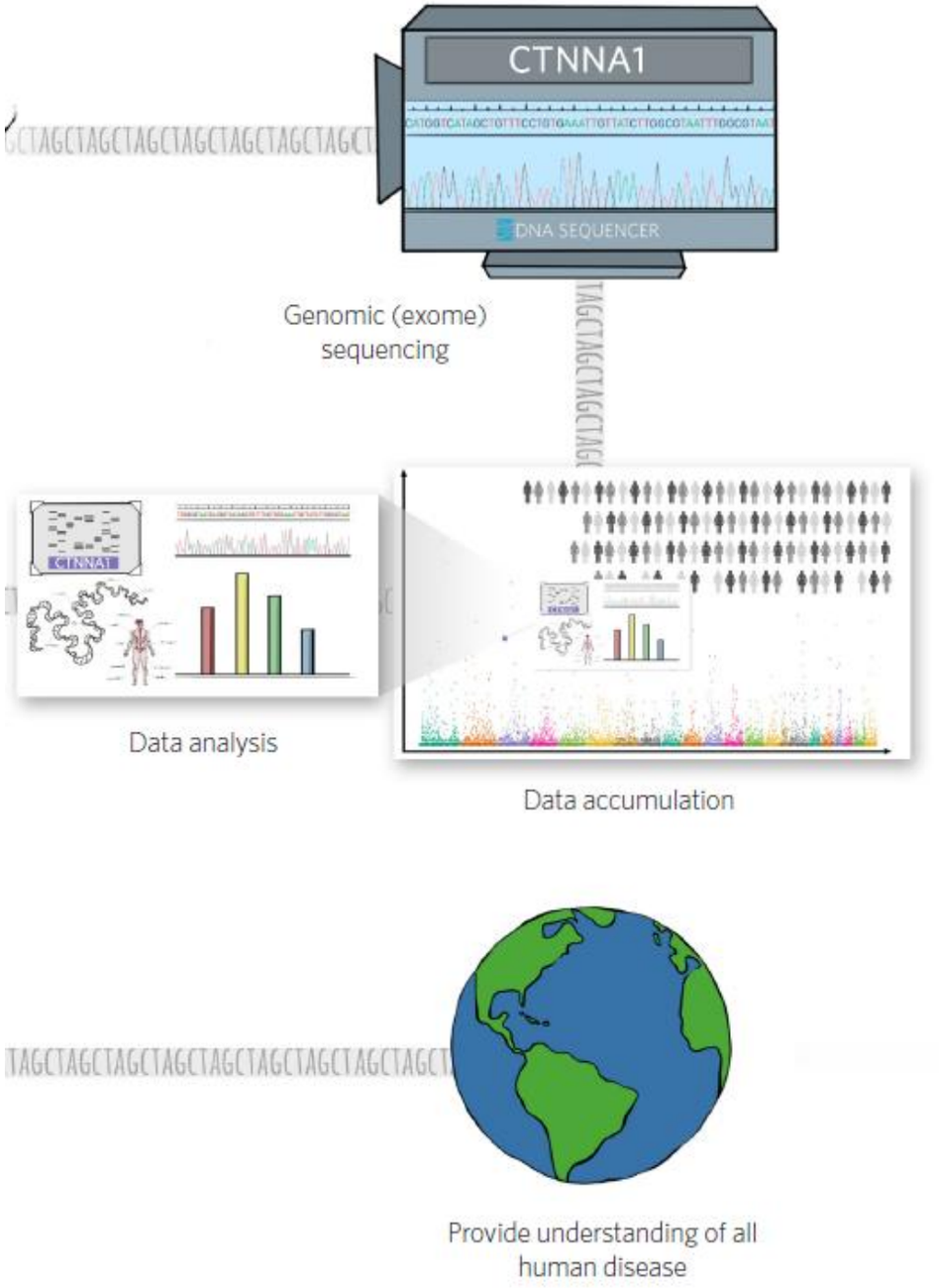
AmbryShare: How?

IRB Submission

- Deemed EXEMPT from consent requirements under 45 CFR section 46.101(b)(4)
  - "Research involving the collection or study of existing data, documents, records, pathological specimens, or diagnostic specimens, if these sources are publicly available or if the information is recorded by the investigator in such a manner that subjects cannot be identified, directly or through identifiers linked to the subjects"

AmbryShare Cohort Selection

- Samples were selected from patient samples received at Ambry Genetics for clinical genetic testing.
- Patients who declined the use of their sample for research were automatically excluded.
- All patients included in the cohort had a disease and/or diagnosis. The first release was focused on patients diagnosed with breast and/or ovarian cancer.
- Samples were de-identified
- Whole exome sequencing was performed, aggregated, analyzed and deposited online

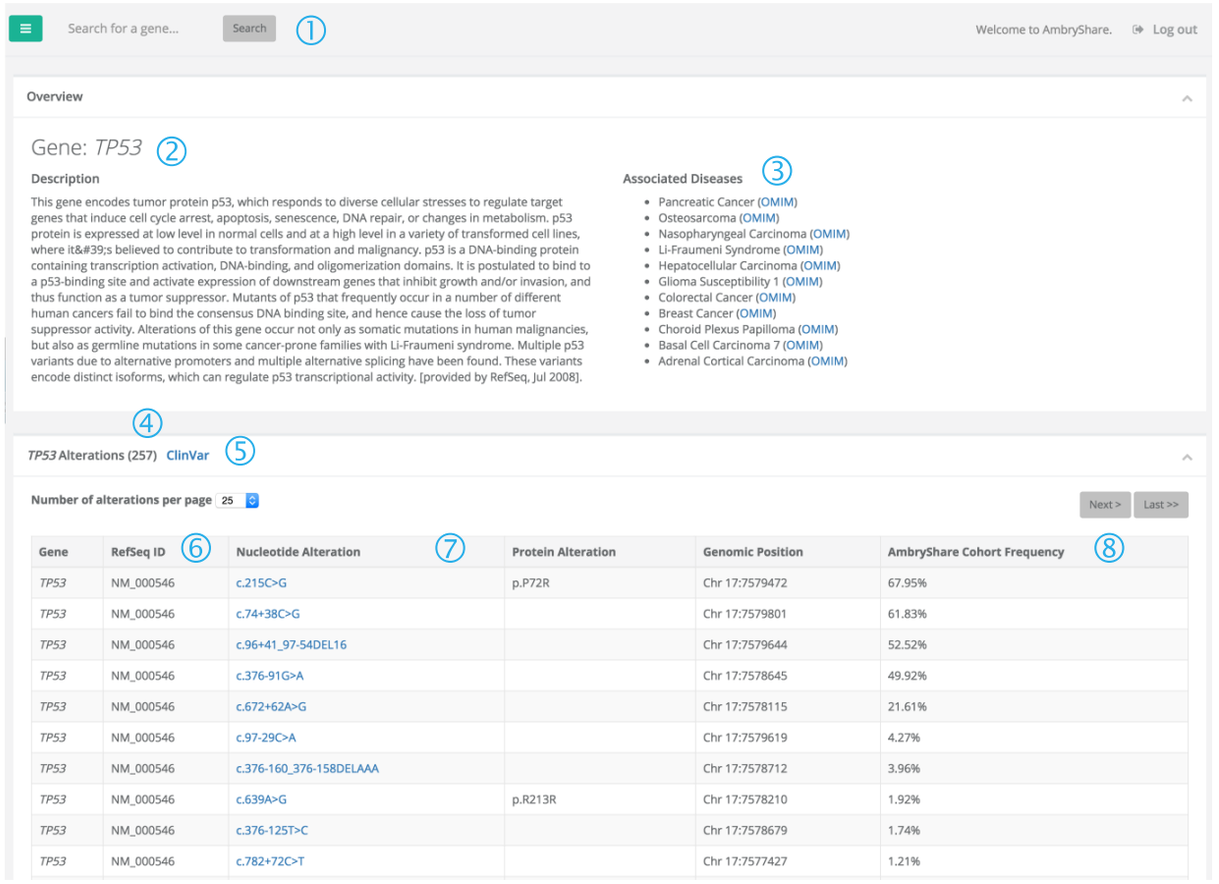


Ambry Genetics® | Data at a glimpse

What Type of Information is Available In AmbryShare?

The database provides:

- The disease type (*i.e.* breast cancer)
- Nucleotide alteration
- Protein alteration
- Genomic position
- AmbryShare cohort frequency for each variant observed.
- Information about protein structure and domain organization for each variant.



① Search by gene/alteration

② Gene description

③ Associated diseases

④ Total number of alterations identified

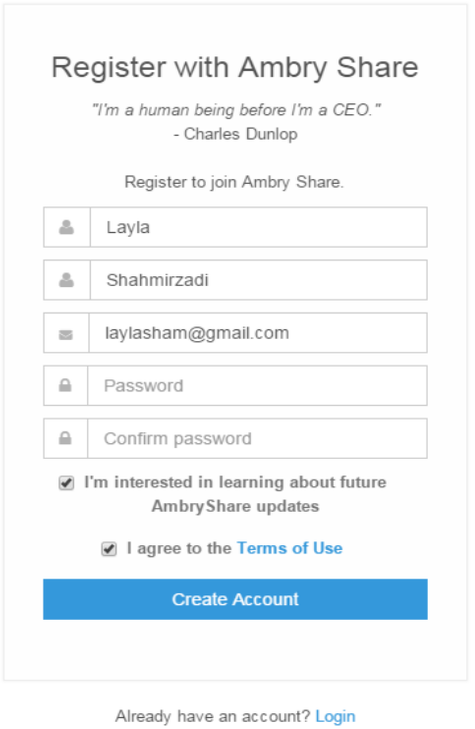
⑤ Link out to ClinVar

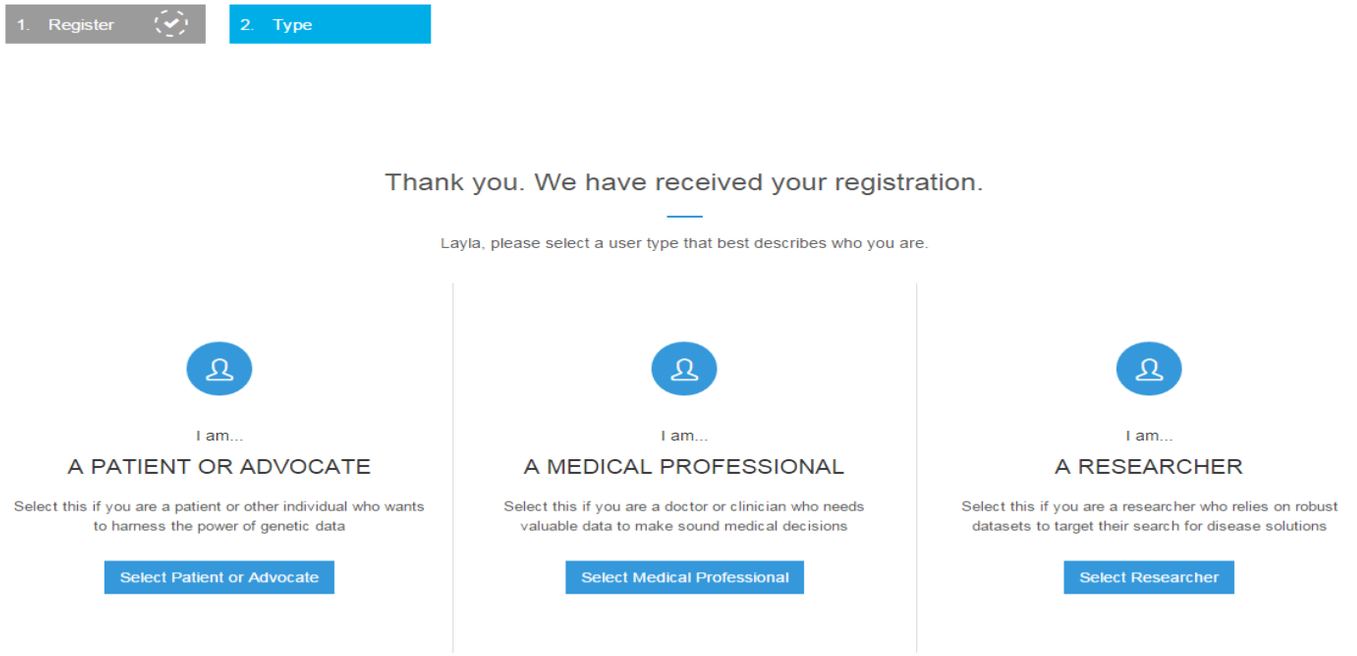
⑥ RefSeq ID

⑦ Nucleotide and protein alterations

⑧ Allele frequency among AmbryShare cohort

More about the Database: Register with AmbryShare

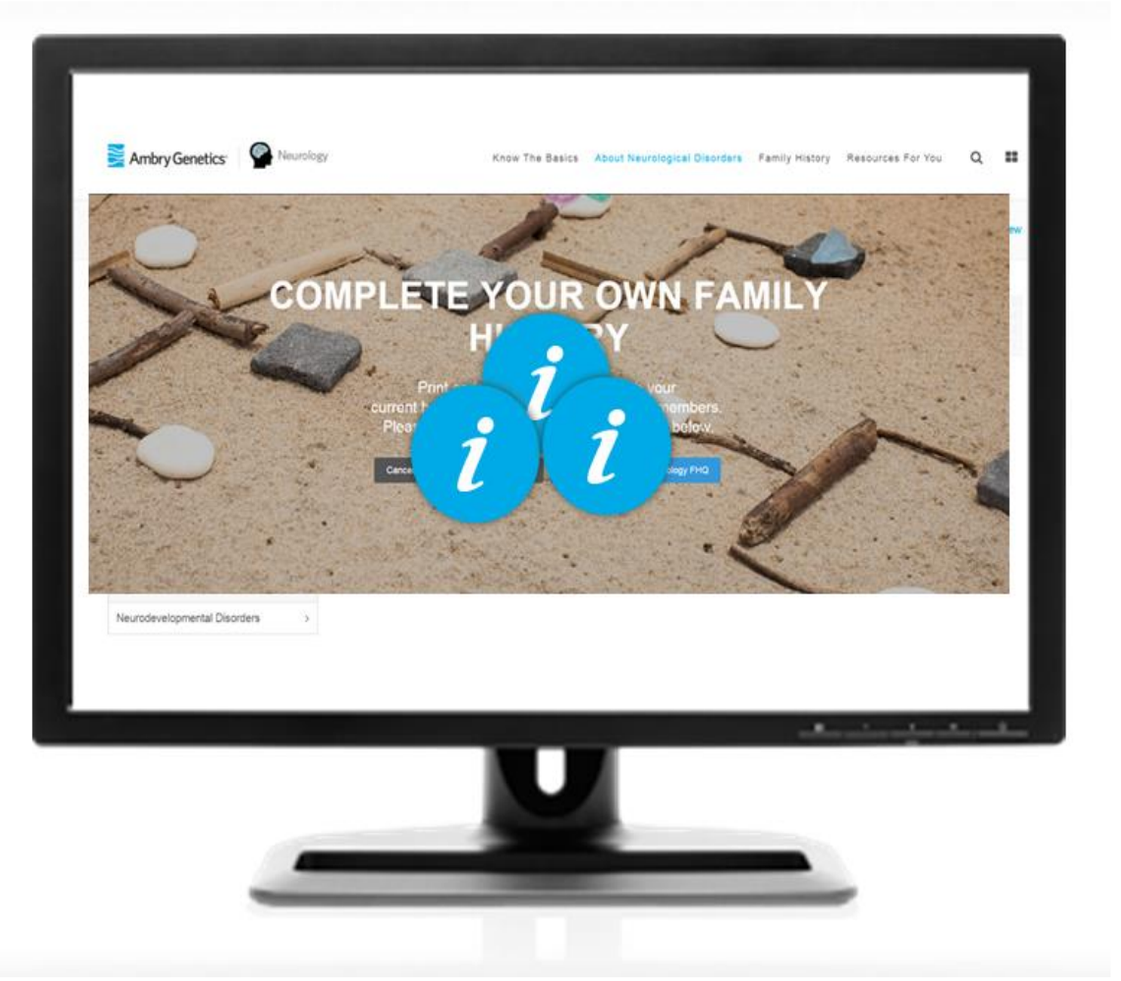




https://share.ambrygen.com/terms-of-use

What Are the Future Plans for AmbryShare?

- Continue to add patients and many diseases to the database
- IRB approval for permission to re-contact
- Expanded patient Involvement
- Worldwide research collaborations



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Get to Know Ambry Genetics

Ambry Genetics is a privately-held healthcare company with the most comprehensive suite of genetic testing solutions for inherited and non-inherited diseases. Since 1999, Ambry has tested nearly one million patient samples benefiting 94% of all U.S. patients covered by public and private insurers. Ambry is dedicated to scientific collaboration by offering its rapidly growing database of anonymized genomic data (variant frequencies) free to the global medical research community to fulfill the promise of the human genome to cure or manage all human disease. Ambry is dedicated to the belief that human health should not be patented or owned, and genomic data should be freely shared so we can try to understand all human disease.

For more information on collaboration with AmbryShare, contact us at share@ambrygen.com