



A next generation health
information company

Our Mission

Enable physicians and patients to more seamlessly engage the digital universe of data, knowledge and understanding to better diagnose, treat, and prevent disease

A Spinout of the Mount Sinai Health System

- Mount Sinai Health System is one of the largest and most respected academic medical centers and health systems in the United States, featuring 7 hospital campuses and Icahn School of Medicine. Mount Sinai has more than 6,000 physicians and 140 ambulatory practice locations throughout the Tri-State Area.
- Icahn School of Medicine is an international leader in medical and scientific training, biomedical research, and patient care, featuring more than 5,000 faculty
- Mount Sinai has made a substantial investment toward the future of genetic research and diagnostics and are committed to developing next generation treatments

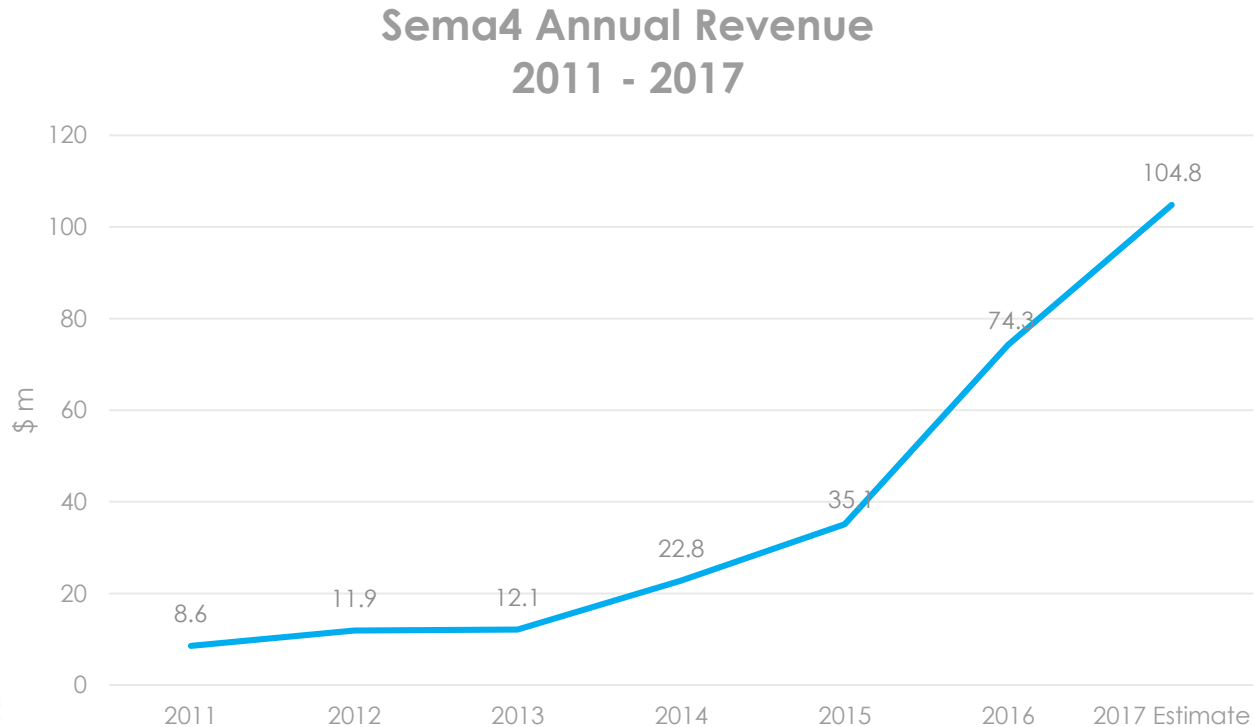


Our Science

Now Ranked 4th in the Country for Medical Research in Genetics

Institution by Rank	Change in Ranking from 2011
Baylor College of Medicine	1 → 1
Washington University	2 → 2
Stanford University	4 → 3
Icahn School of Medicine	31 → 4
North Carolina, Chapel Hill	6 → 5
Harvard Medical School	9 → 6
University of Washington	7 ← 3
University of Miami MS	20 → 8
Columbia University	13 → 9
Duke University	5 ← 10
Johns Hopkins University	24 → 11
University of Pittsburgh	12 ← 10
Yale University	26 → 13
University of Chicago	7 ← 14

Sema4 has grown rapidly over the past 5 years

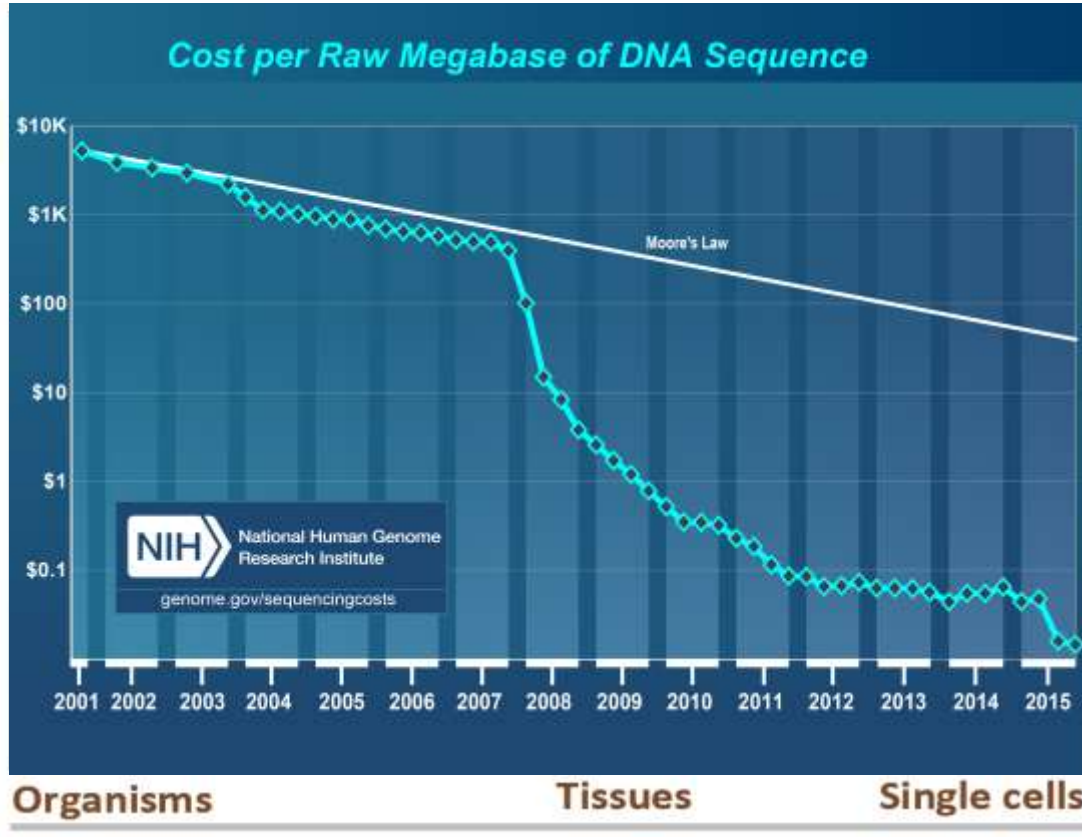


Advanced technologies have given rise to an explosion in the digital universe that is fundamentally transforming life as we know it. You know you're getting old and being left behind when...



27 million people watched the 'League of Legends' World Championship (more than the World Series or NBA Finals)

Advances in DNA sequencing technologies have now added to this explosion



These types of rapid technology advances are delivering massive scales of data around individuals, that if appropriately integrated, can dramatically alter our ability to predict disease risk or diagnose, treat, or even prevent disease



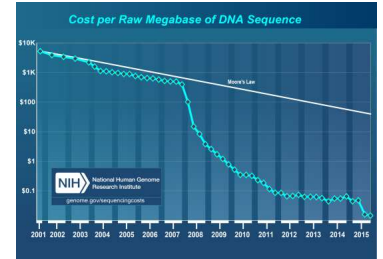
Mobile + Social Networks



The Cloud



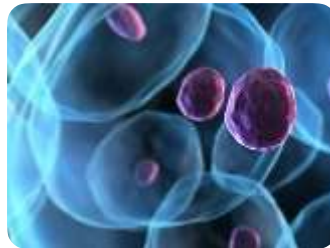
Big Data Analytics



Next-Gen Genomics



FUR YOUR EYES ONLY CIA 'implanted microphones into CATS' in a bizarre attempt to spy on Russia



Advanced Materials

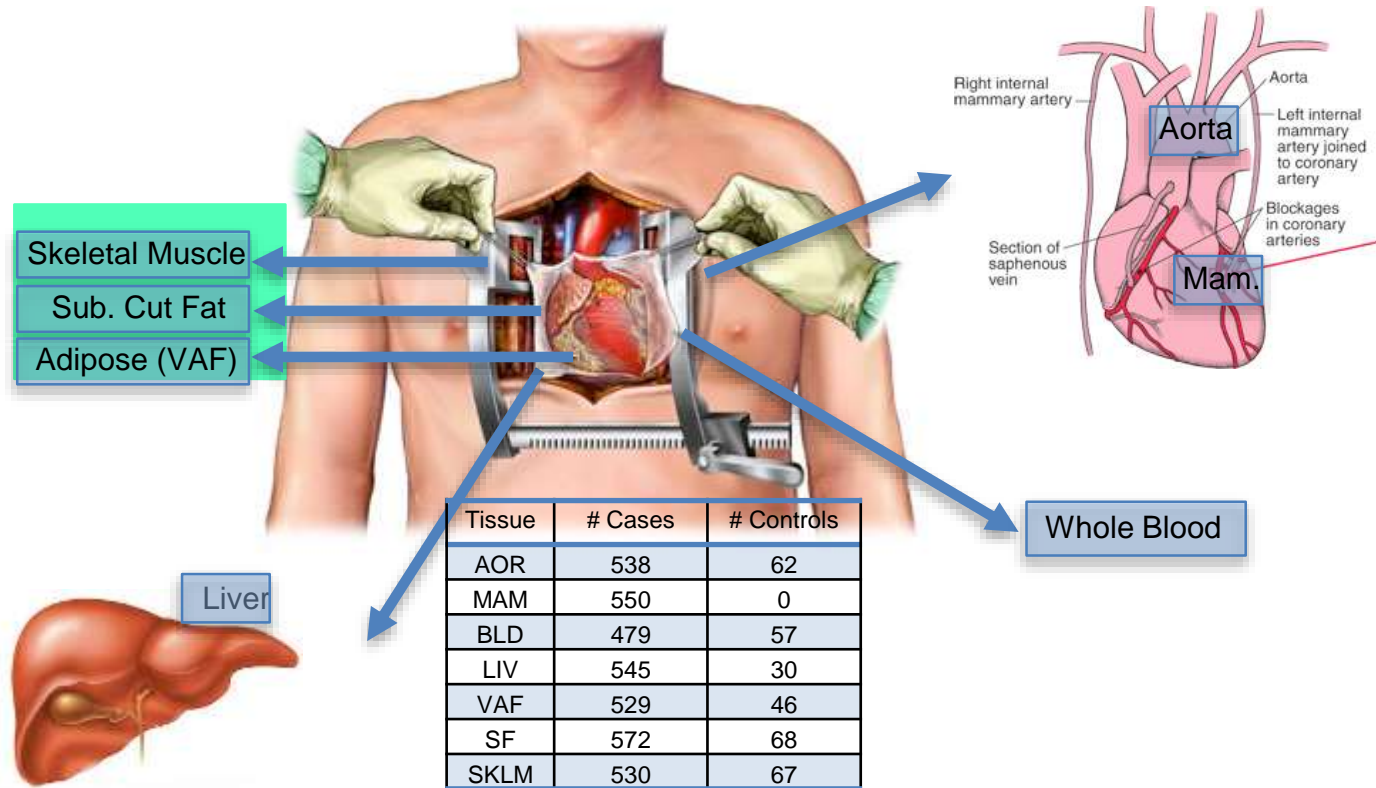


3D Printing



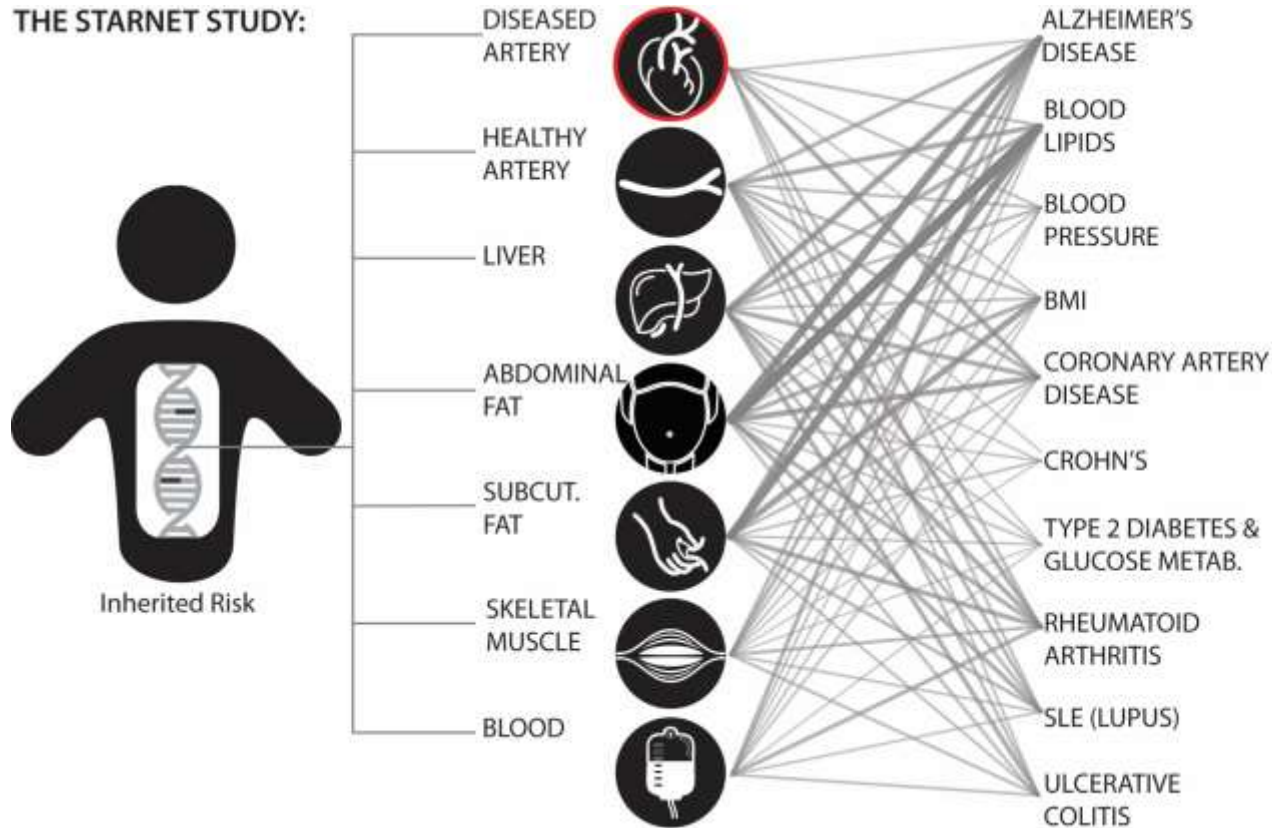
The "Internet of Things"

On the molecular side, significant cohorts profiled at an unprecedented depth



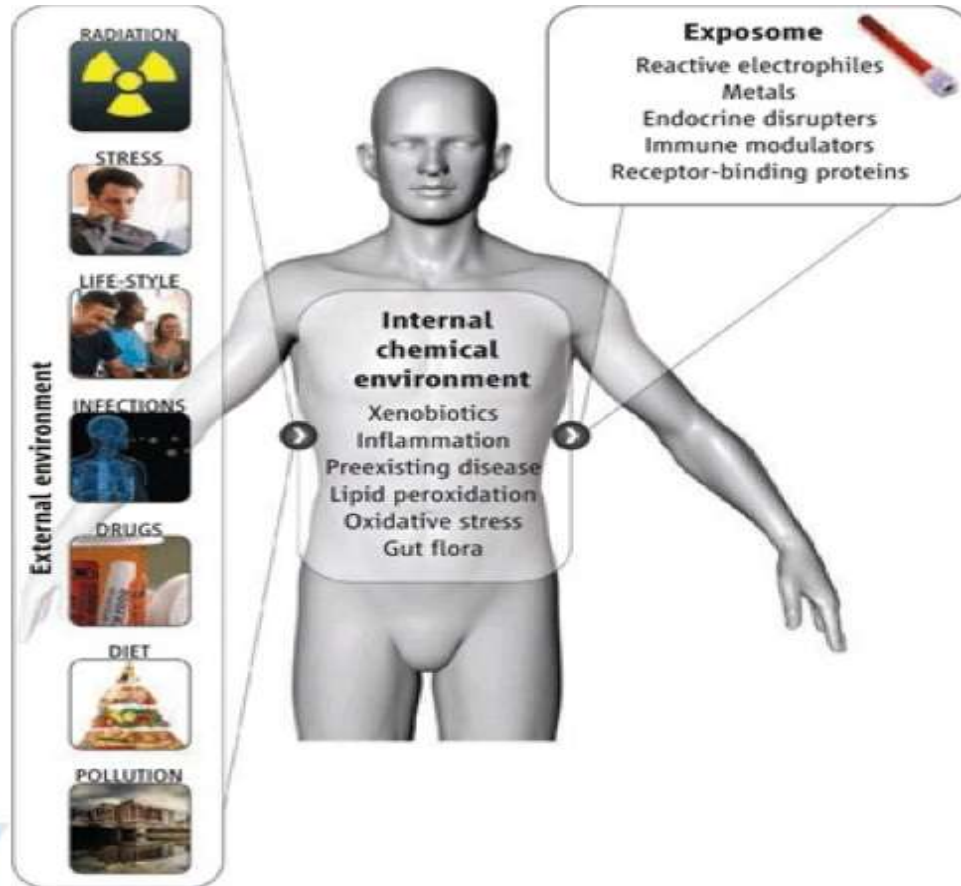
Providing for the first time extensive maps of disease

THE STARNET STUDY:



The thickness of the edges between the tissue and the disease reflect how frequently a disease-causing gene is shared between pairs of diseases and tissues.

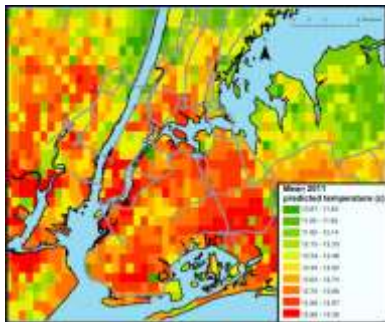
Complementing the molecular dimensions is the “Exposome”, the newest “Omic”



“The totality of environmental exposures throughout a lifetime”

-Analogous to the Genome

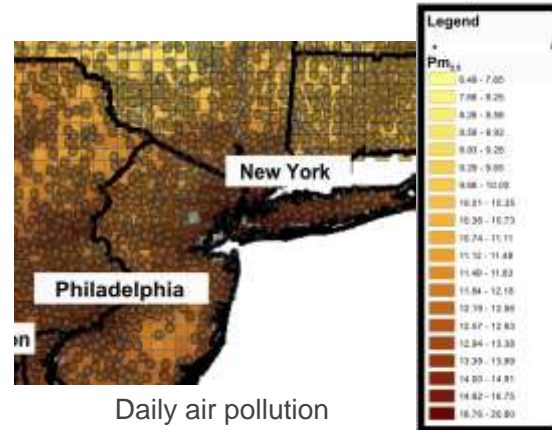
App enabled use of Geographical Information Systems for the “*External Exposome*”



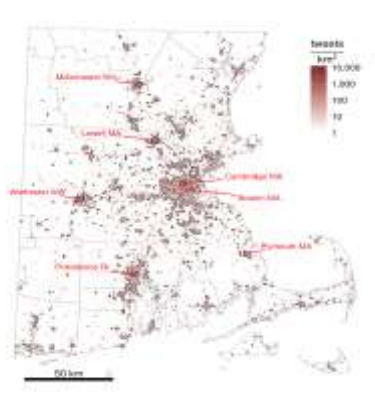
Air Temperature/Climate



Exposure to green, natural areas



Daily air pollution



Social Media Content

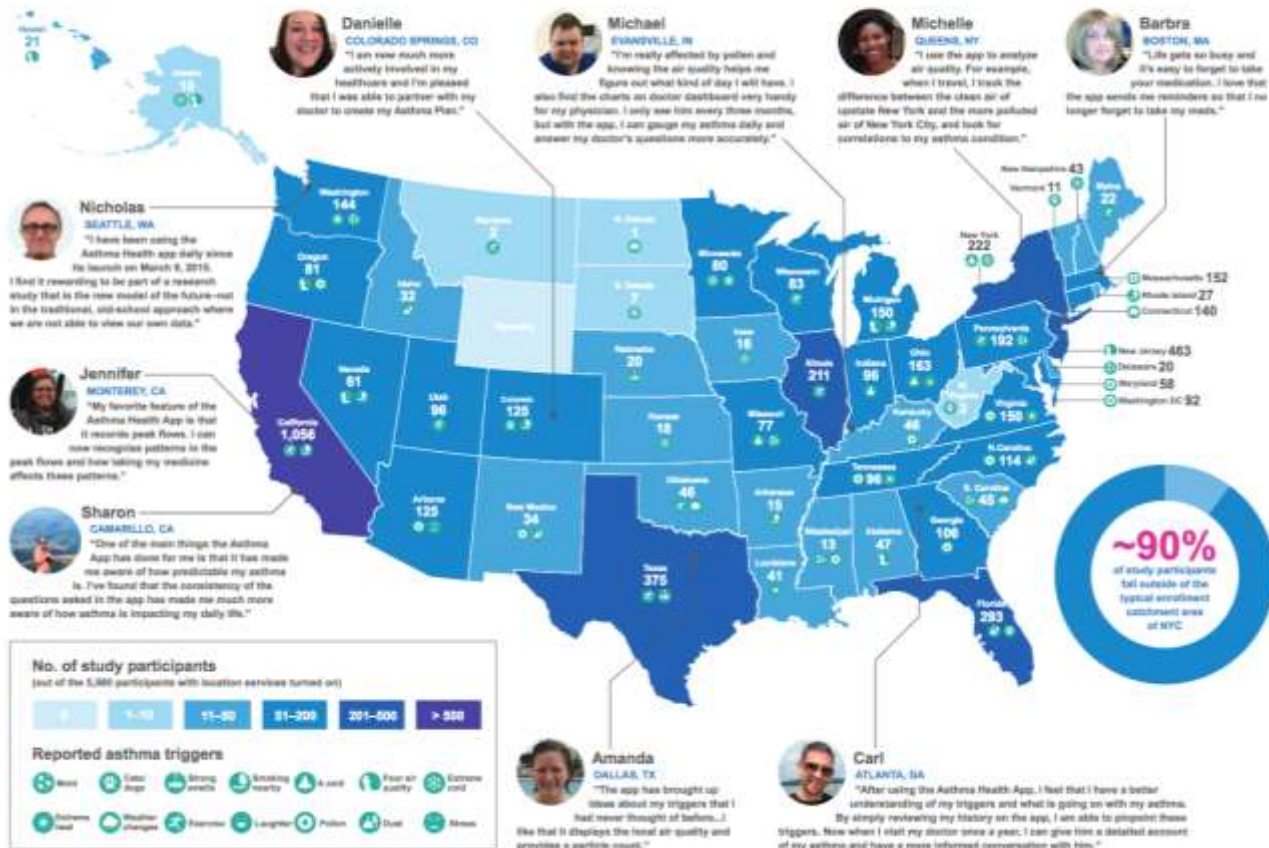


Access to Healthy Foods



Traffic patterns

Asthma Health App, U.S. Participants



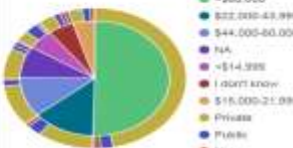
About Us

About Users

5486

Unique count of *WTTN*

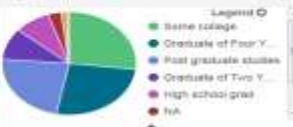
Hagopian & Himmelfarb



Erweiterung



12. <http://www.pearsoned.com>



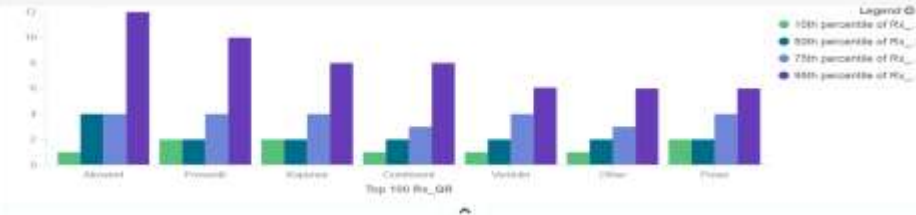
<http://www.elsevier.com/locate/jbiotec>

Medication

Rx_Quals_History

Top 25 Rx_QR #	Unique count of MMRN #
Proair	1894
Ventolin	1407
Other	777
Proventil	506
Xopenex	326
Combivent	111
Airvent	103

R12 OR R12F N100 OFF



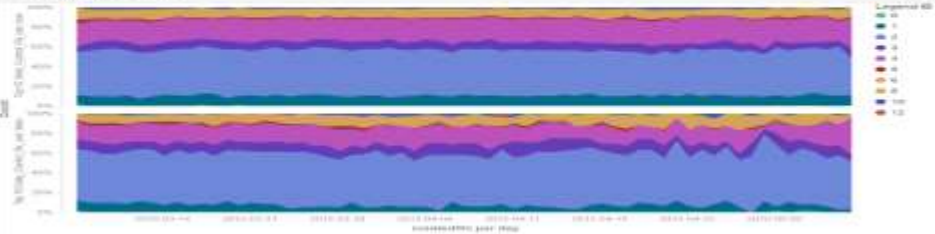
Library Acquisitions: Journals

Daily Asthma Trends

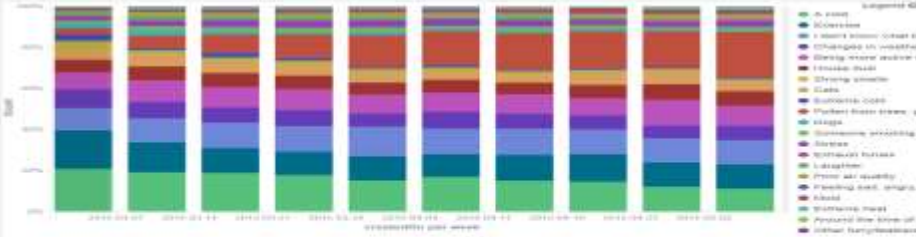
50896

Chen

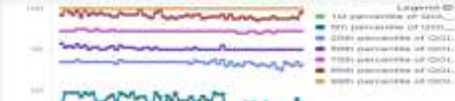
Black-CPR cannot be compared with



Shady, Tennessee



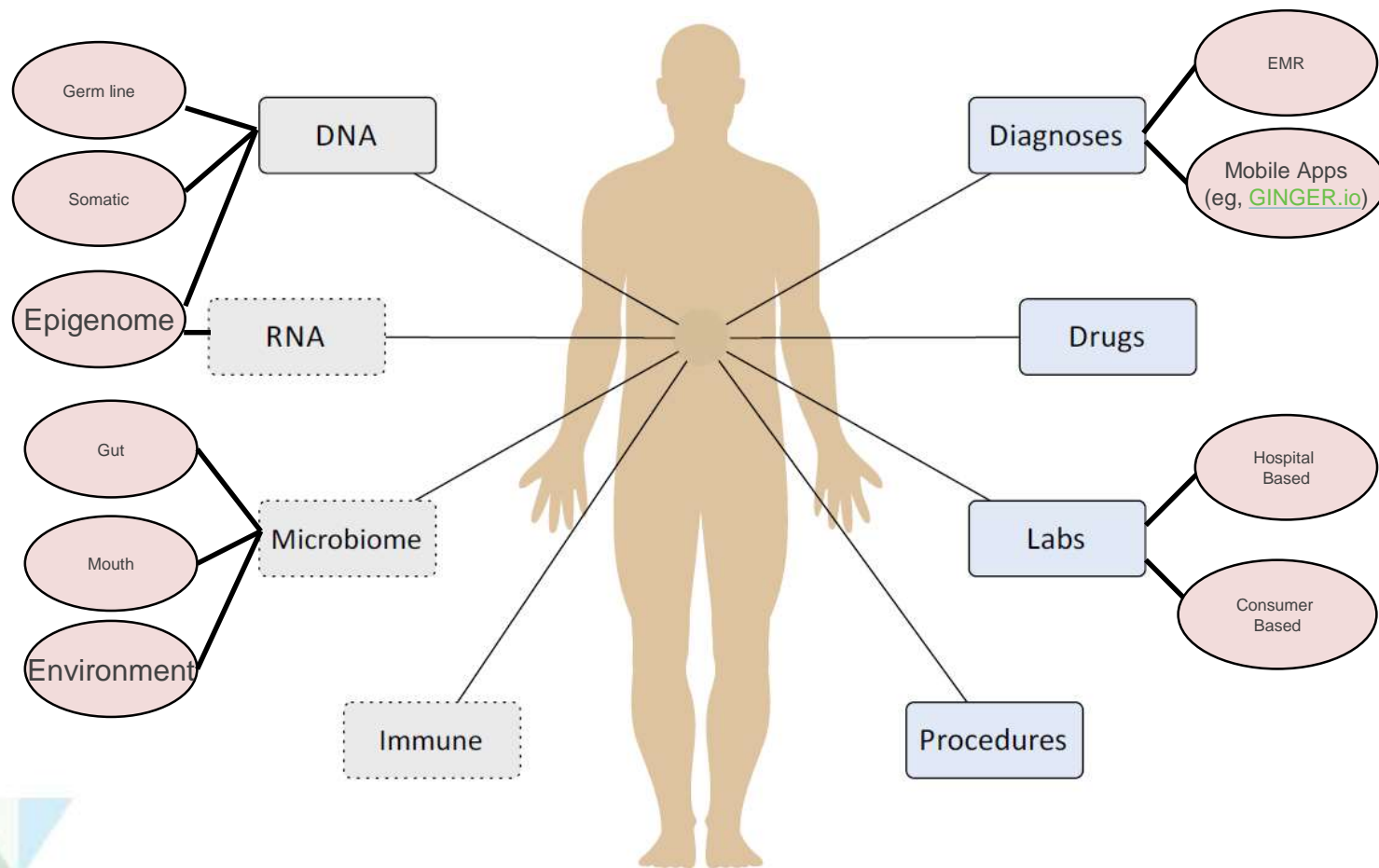
250K Monthly Working Hours



Keywords: gender inequality, social capital, trust, economic growth



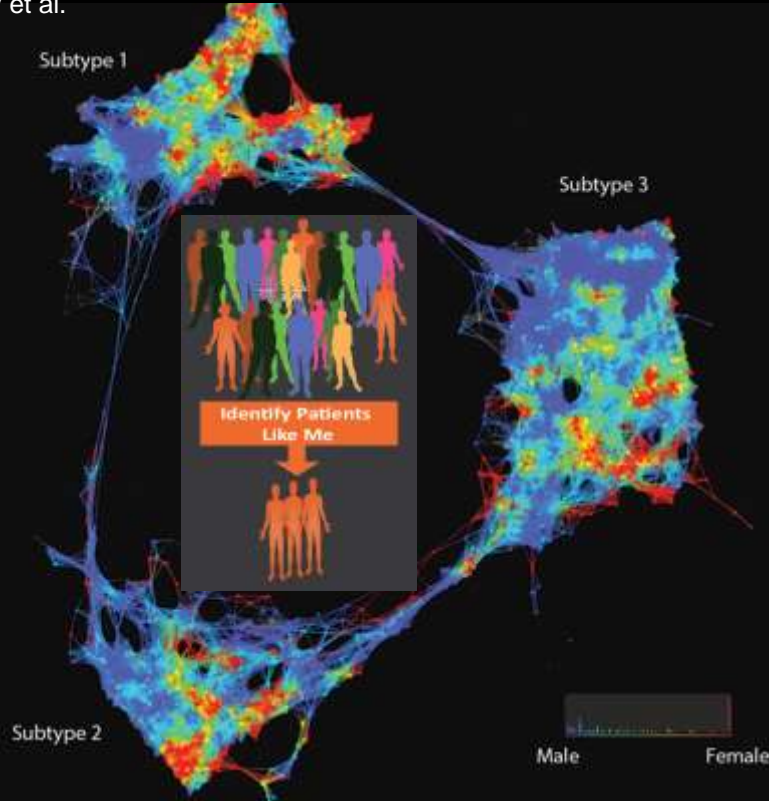
The value of a more completely phenotyped and molecularly profiled population



Using EHR data, diabetic patients organized into 3 distinct groups representing different severity, comorbidities and genetic components

Science Translational Medicine NIH/AAAS

Dudley et al.



Subtype 1:

- More likely to suffer from blindness and vision defects
- Grouping genetically supported

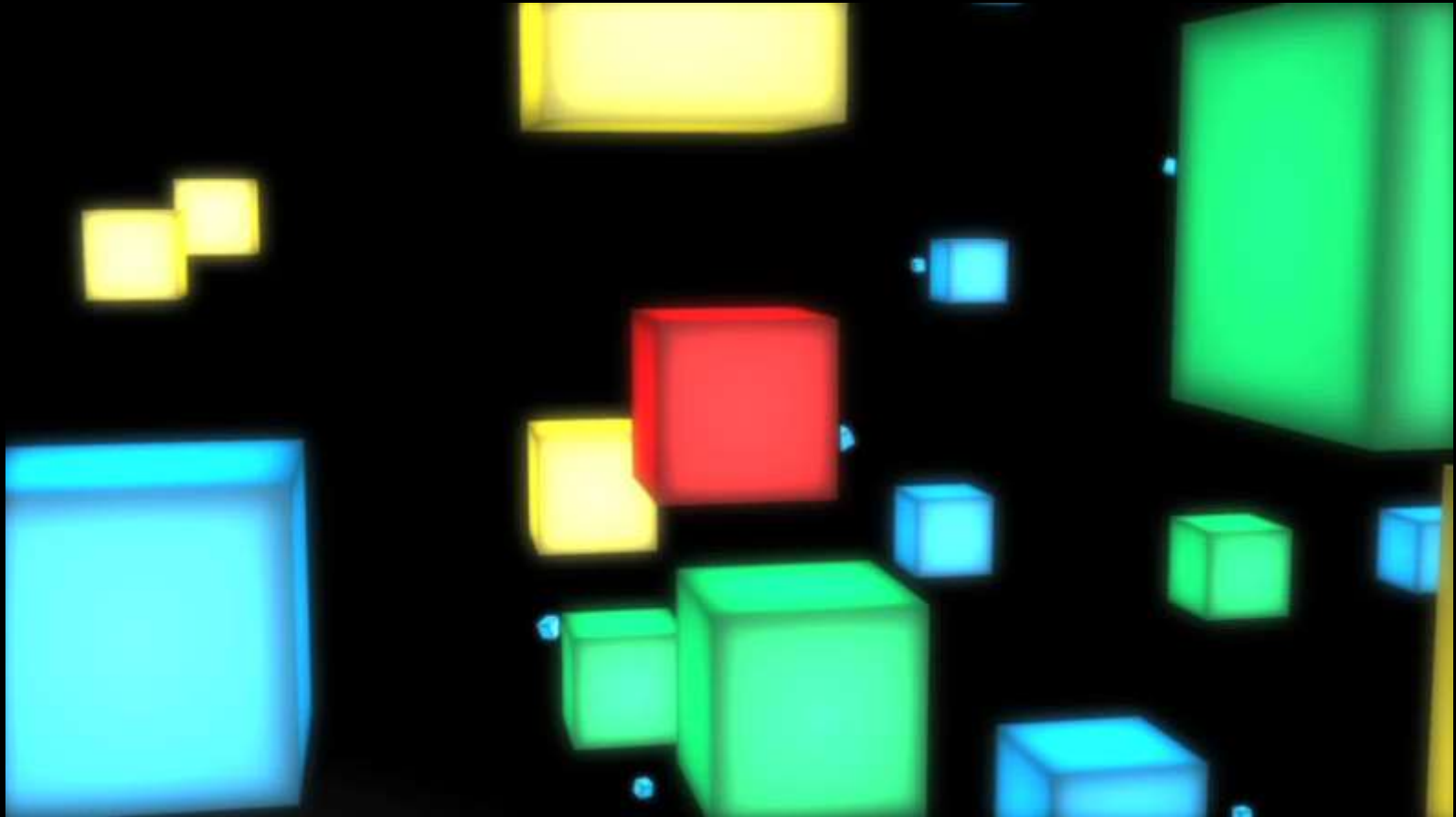
Subtype 2:

- Greater risk of infections and cancer
- More immune deficient

Subtype 3:

- Higher blood pressure, blood clots, more metabolic syndrome like
- Grouping genetically supported

We can go further to integrate these different dimensions of data to build models

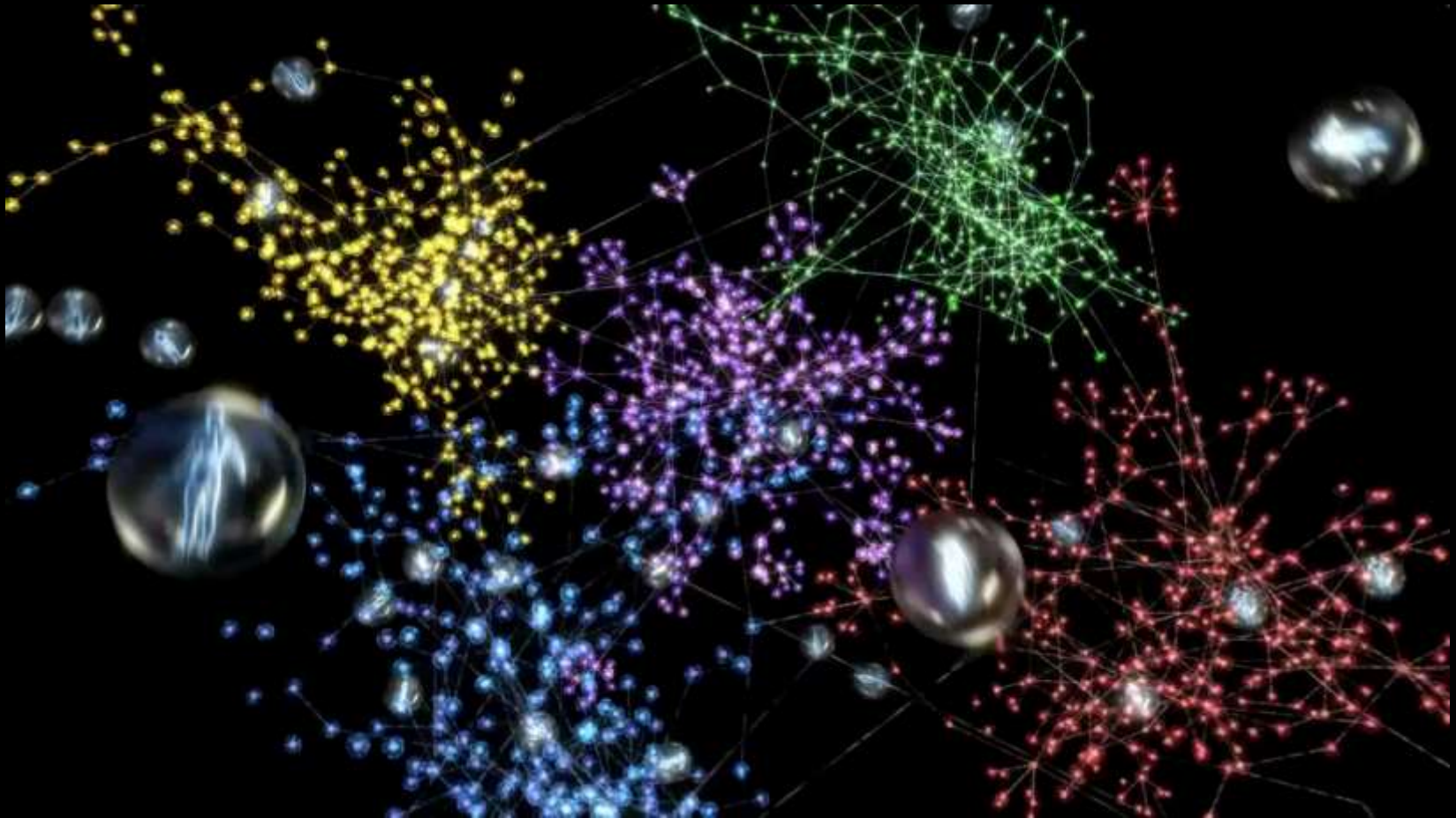


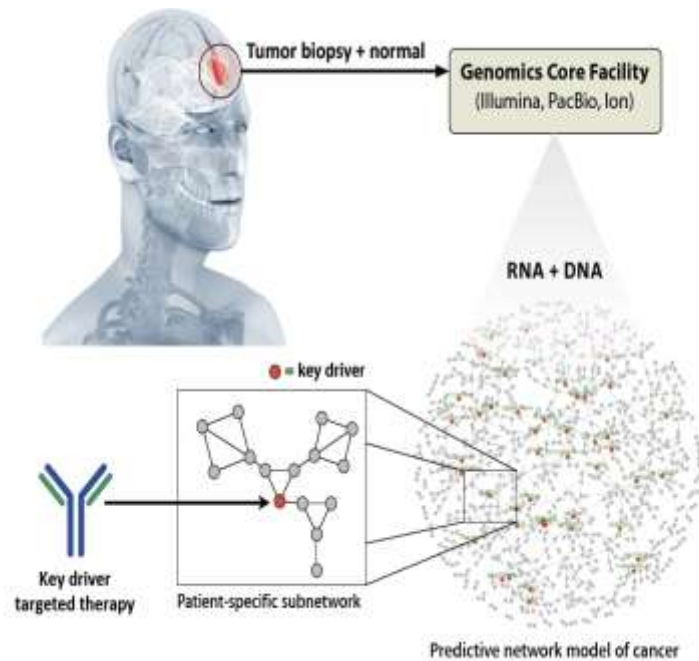
■ Host Molecular(DNA, RNA, Proteins) ■ Microbiome Molecular ■ Clinical (EMR) ■ Consumer Acquired

Simulation on these models enables in silico perturbations the elucidate information flow



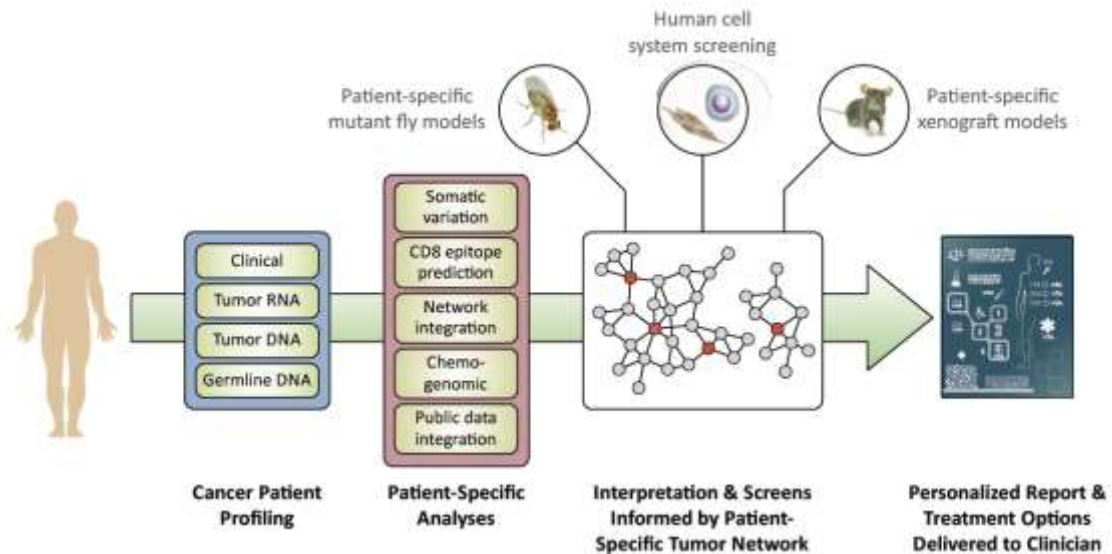
Sema4: New MS effort pushing an information driven approach to reinventing medicine





Pioneering today the application of these individual patient trajectories in cancer

Patient tumor and germline are profiled, key drivers identified, tumor constructed in avatar models, those models taken through HTS for identification of drug cocktail



Medical systems of the future...

Largest car company in the world
owns no cars (founded 2009)



64 COUNTRIES

> 200,000 drivers in US
> 1,000,000 drivers world wide

Largest hotel chain in the world owns
no hotels (founded in 2008)



Total Guests
60,000,000+



Cities
34,000+



Castles
1,400+

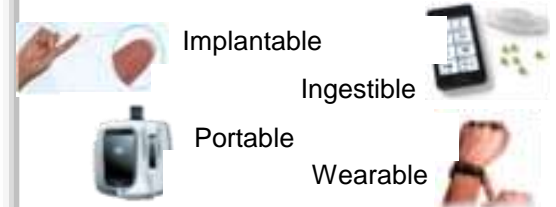


Countries
190+

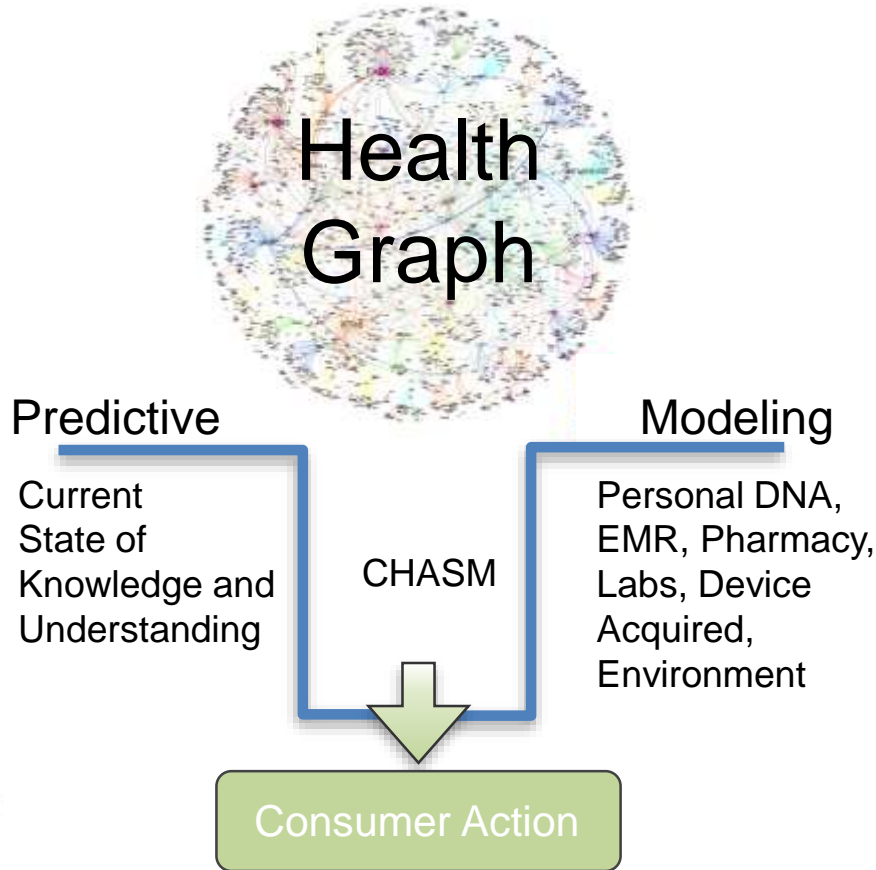
Listings Worldwide

2,000,000+

Soon the largest medical system in
the world will own no hospitals

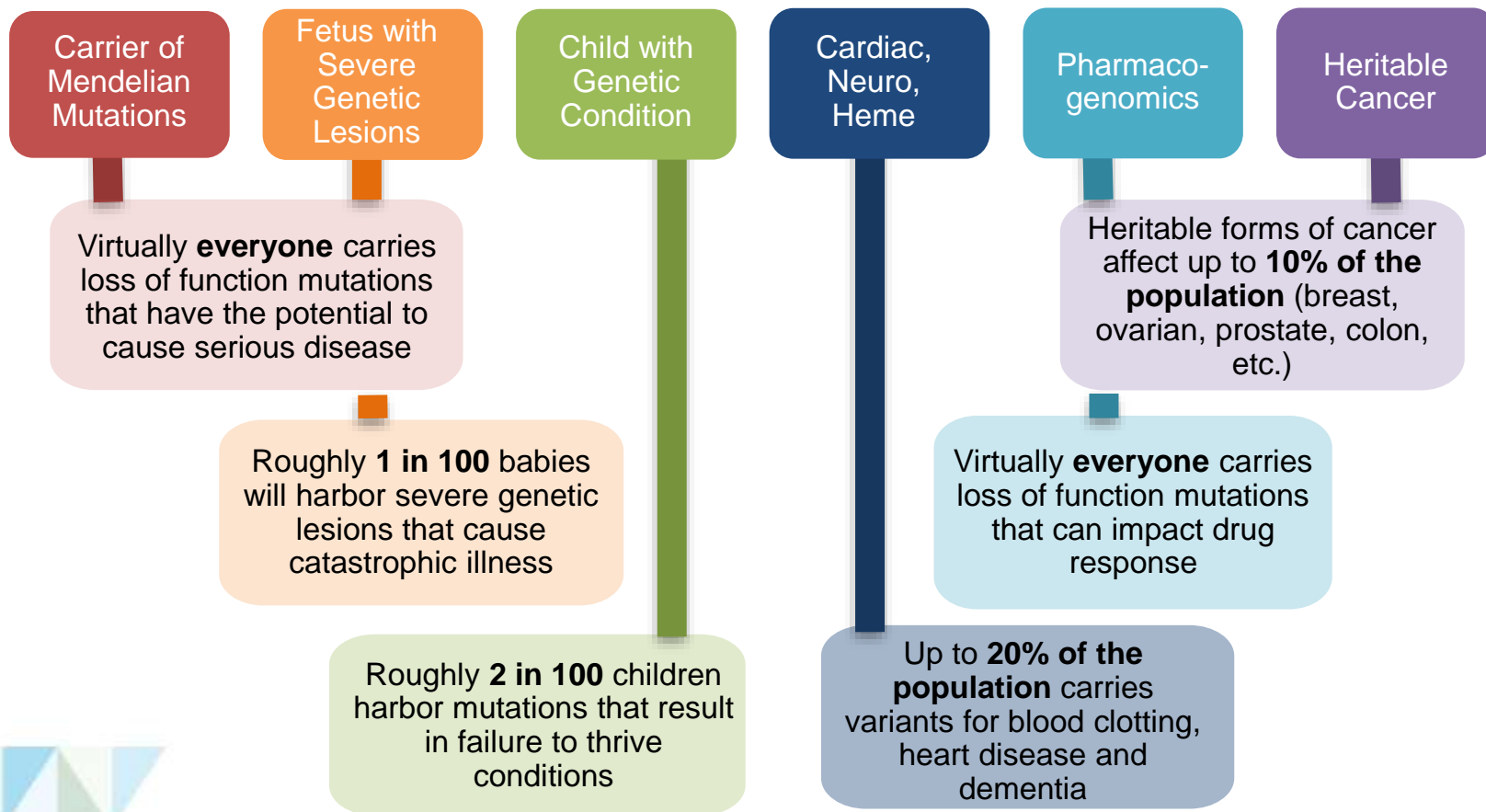


THE PROBLEM: We do not have the scale of content needed to build these models to realize this vision



Unique capabilities of coupling what people ***need*** to know with what they ***want*** to know for their future

Which translates into being able to see far more of the clinically actionable genome for much less cost



Our Mission: Enable physicians to more successfully manage the digital universe of data, knowledge and understanding to diagnose, treat, and prevent disease

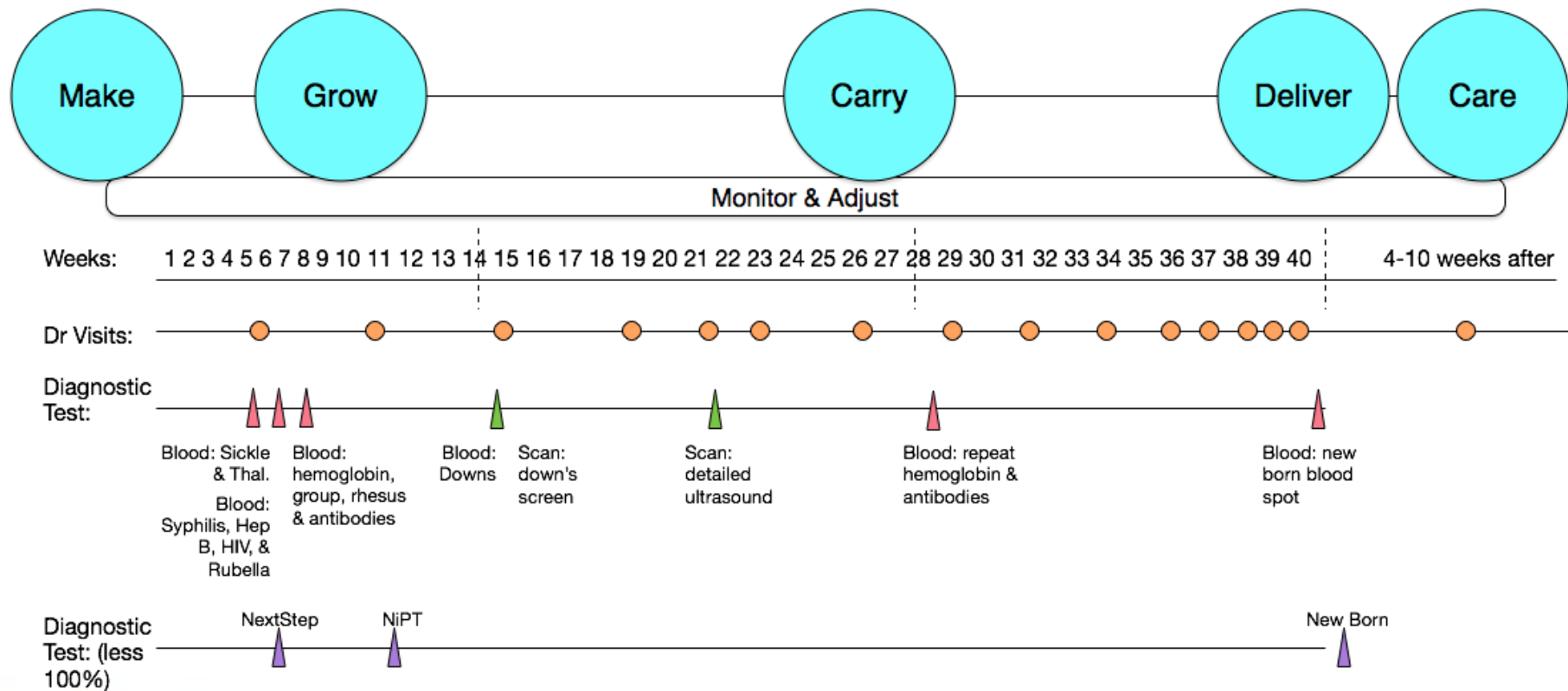
DATA ACQUISITION AND
MANAGEMENT
(taking into account more
complete information)



Not just a testing company, but an information partner to help physicians help families deliver children free of disease

- Aid physicians in maximizing patient outcomes
- Streamline physician workflow
- Partnering to enable learning healthcare systems

Milestone connection between diagnostic testing and information system

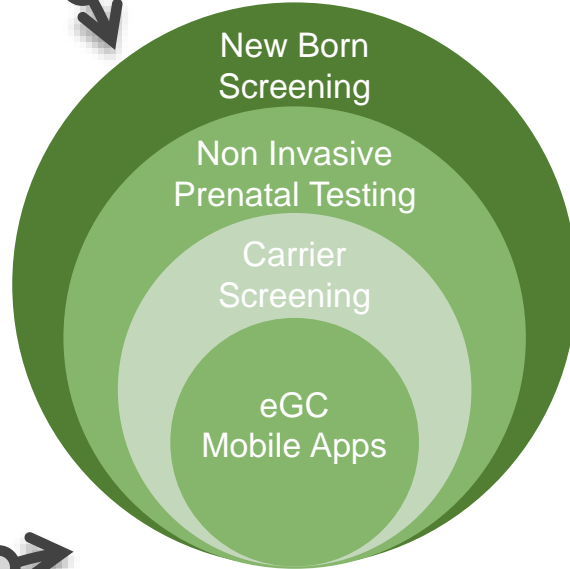


Direct to Consumer

Helix Carrier Screening

NBS from Dx pathways

Search for Resilience



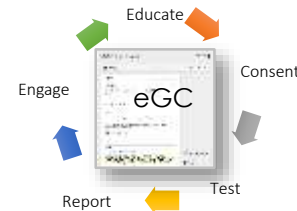
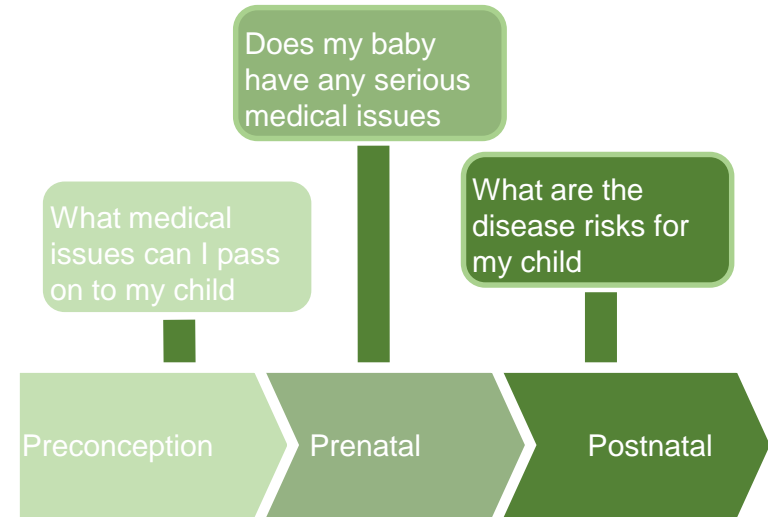
DT Medical Systems

NBS Dx Pathway for Pharma

Current Carrier and NiPT DTP

Clinical Genetic Testing

General surveillance and managing wellness

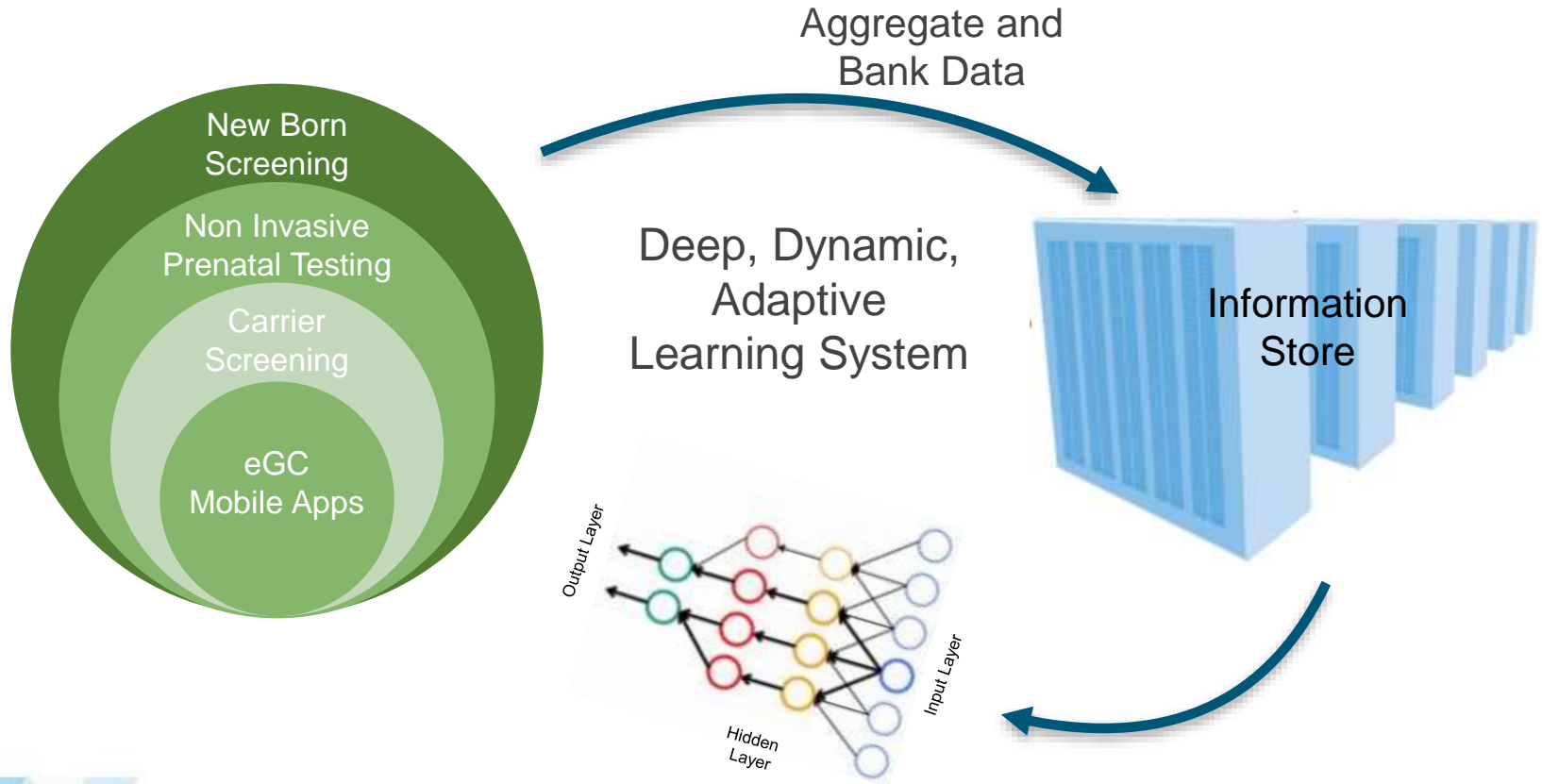


Web & Mobile Apps

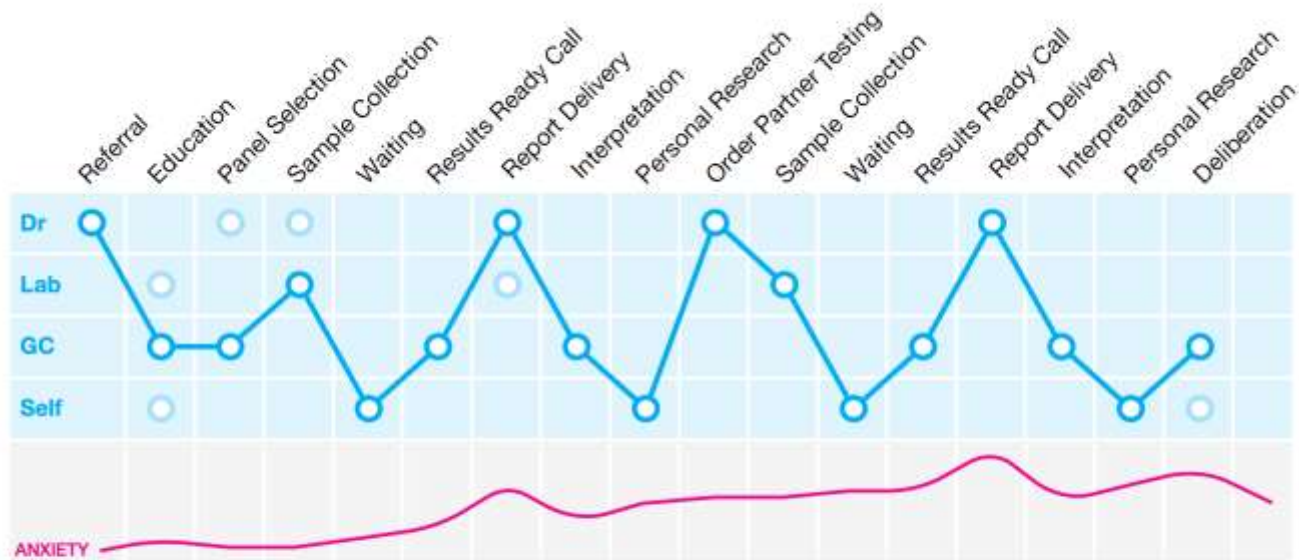


Managing pregnancy and postnatal journey

Creating a feedback loop to continually refine test interpretations and expand test utility



Existing Customer Journey



MAJOR TAKEAWAY

An anxiety-inducing rollercoaster experience with too many variations.

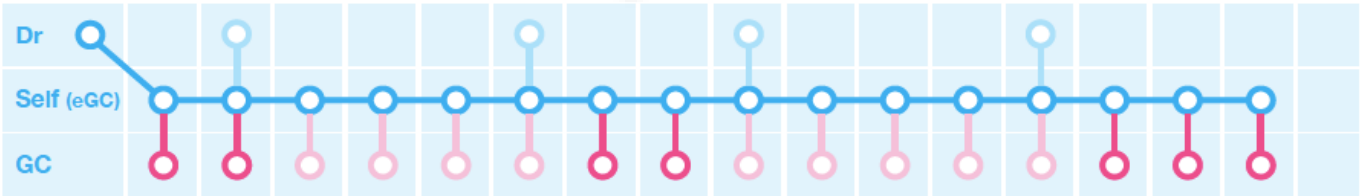
- Patients bounce back and forth across stakeholders almost at every step.
- Inconsistent service throughout, relying too much on individual interactions.
- GCs are overloaded with logistics and repetitive tasks that steal time from actual counseling.

New Customer Journey

Current Journey



eGC Journey



sema4

Create your
account today.

Access to your results, records and health data.
All in one place.

Create Account



Live Consult Demo

Please select your role to launch the demo

PATIENT



Sarah Wagner
Patient

COUNSELOR



Janelle McCarthy
Genetic Counselor
Mount Sinai

SCENARIO Upon her Carrier Screening test, Sarah Wagner (patient) has scheduled a Live Consult session with Janelle McCarthy (genetic counselor).

This prototype demonstrates the key aspects of the Live Consult session.

Stephanie Arnold

Profile Settings

Tests

Tests

Expanded Carrier Screening Test

NEW

Test Date: 1/31/17

Referring Physician: Robert Smith, M.D.

- ✓ Sample Received 2/1/17
- ✓ Analysis Complete 2/15/17
- ✓ Consult 2/17/17, 10 am
- ✓ Results Available 2/17/17

[View Results](#)



Mount
Sinai

Live Consult



Sarah W. ▾



Janelle McCarthy
Genetic Counselor

Waiting for Janelle to start the session



NextStep™ Consulting Session

Begins at 12:30 EST, 02.12.2016

Call in Number [555-555-5555](tel:555-555-5555)



Sarah Wagner — [Resend Invite](#)

Patient Phone: 555-555-5555

Session has not started

[Start Consult](#)

NextStep™ Live Consult Session

Genetic Counselor: Janelle McCarthy

WHAT WILL BE COVERED

Summary

Result Interpretation

Risk Overview

Recommendations





Sarah Wagner — [Resend Invite](#)

Patient Phone: 555-555-5555

Session In Progress • 00:22

[End Consult](#)

NextStep™ Results Summary

↓ REDUCE RISK

No variants were detected in 279 Genes.

VARIANT FOUND

Condition 1 - Lorem Ipsum

Condition 2 - Lorem Ipsum



Sarah Wagner — [Resend Invite](#)

Patient Phone: 555-555-5555

Session In Progress • 00:31

[End Consult](#)

Condition 1 - Lorem Ipsum - ABCD Gene

Condition Description

Lung Airway Without Cystic Fibrosis



Lung Airway With Cystic Fibrosis



Cystic fibrosis (CF) is a genetic condition characterized by the production of abnormally thick, sticky mucus, particularly in the lungs and digestive system. While it is normal to have mucus lining the organs of the respiratory, digestive, and reproductive systems in order to lubricate and protect them, in people with CF this mucus is thick and sticky. This abnormal mucus results in the clogging and obstructing of various systems in the body. CF is a chronic condition that worsens over time.





Sarah Wagner — [Resend Invite](#)

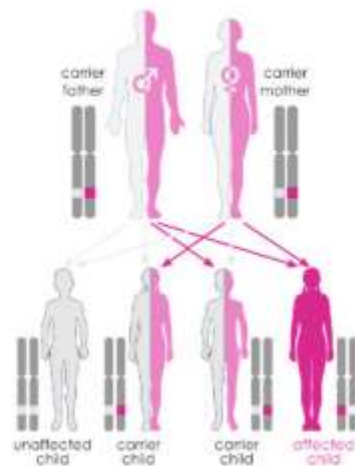
Patient Phone: 555-555-5555

Session in Progress • 00:40

End Consult

Condition 1 - Lorem Ipsum - HEXA Gene

Inheritance Diagram



Sarah Wagner — [Resend Invite](#)

Patient Phone: 555-555-5555

Session In Progress

00:48

[End Consult](#)

Reduced Risk for 279 Conditions

CONDITION	GENE	FREQUENCY	DETECTION RATE	VARIANT	RESIDUAL RISK
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced
Abetalipoproteinemia - AR	MTTP	1 in 186	>95%	NOT FOUND	1 in 3701 ↓ Reduced



Sarah Wagner — [Resend Invite](#)

Patient Phone: 555-555-5555

Session In Progress • 00:57

[End Consult](#)

Recommendations & Next Steps

NEXT

- **Test your partner** Testing your partner will check if he is also a carrier of [Tay-Sachs](#) and [Mediterranean Fever](#). Testing will give you a more conclusive understanding of your risk.

AFTER PARTNER TESTING

- **Consideration of residual risk** after a negative carrier screen is recommended, especially in the case of a positive family history for a specific disorder.

DURING PREGNANCY

- **Fetal DNA Test** Nemo enim ipsam voluptatem quia voluptas sit aspernatur aut odit aut fugit, sed quia consequuntur magni dolores eos qui ration.

AFTER BIRTH

- **Carrier Testing** if you and your partner carry a condition likely to affect your child we recommend testing your child after birth so they can.



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Your Sema4 account.

Access to your results, records and health.
All in one place.

Need to create
an account?

Create

Log In

Email

Dr.Smith@mssm.edu

Password

.....8

[Forgot Password?](#)

Log In



123 Broad Street
New York, NY 10029
123-456-7890

For Pregnancy

Register Sample
Diseases Screened
Information Sheet
Provider Info

For Oncology

Partnerships
Research
Test Catalog

For Providers

Test Catalog
Ordering Tests
Testing Labs
Customer Service

Our Story

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Dr. John Smith

Notifications



Abnormal Result Found Abigail Smith, Sample #23802830



Tests

**Order**

Name ▾	DOB ▾	Sample ▾	Test ▾	Date ▾	Physician ▾	Status ▾	Results Viewed ▾	Consult ▾
Abigail Smith	1/12/83	23802830	Next Step	1/12/16	Smith, John	Final	Y	Y
John Adams	1/8/80	83092803	Next Step	1/12/16	Smith, John	Final	N	Y
George Hoover	2/12/78	10838275	Next Step	1/12/16	Smith, John	Final	Y	Y



Welcome back, Jane

Your CarrierCheck results are ready.

This report will help you understand what your results mean and give you some suggestions on what to do next.

What We Found

We analyzed your DNA to see if you are a carrier of **gene variants (or changes) associated with 65 inherited conditions**. Carriers are usually healthy, but may have an increased risk of having a child with a genetic disease.

⊕ Carrier

1 Condition

You tested **positive** for one of the gene variants that we screened for.

⊖ Negative

64 Conditions

You tested **negative** for most of the gene variants that we screened for.



Dr. John Smith

Tests

[Order](#)

Name	DOB	Sample	Test	Date	Physician	Status	Results Viewed	Consult
Abigail Smith	1/12/83	23802830	Next Step	1/12/16	Smith, John	Final	Y	Y
John Adams	1/8/80	83092803	Next Step	1/12/16	Smith, John	Final	N	Y
George Hoover	2/12/78	10838275	Next Step	1/12/16	Smith, John	Final	Y	Y
Katherine Washington	1/12/82	66729300	Next Step	1/12/16	Smith, John	Final	Y	Y
Quincy Cleveland	3/24/84	20814423	Next Step	1/12/16	Smith, John	In Progress	N	N
Eleanor Smith	11/18/75	11390872	Next Step	1/12/16	Smith, John	Final	Y	Y

Patient	Sample	Referring Doctor
Patient Name: XXXX Date of Birth: 6/19/1980 Reference #: 3441385 Indication: Carrier Testing Test Type: Full Pan-Ethnic	Specimen Type: Blood Lab #: 1410970 Date Collected: 7/16/2016 Date Received: 7/16/2016 Final Report: 7/16/2016	Referring: XXXXZXXXX, M.D. Park Avenue New York, NY 10128 Fax#: XXX-XXX-XXXX

Results Summary

➔ **POSITIVE for Smith-Lemli-Opitz Syndrome**

One copy of the c.964-1G>C mutation

Negative for all other mutations tested in the other 280 diseases

Recommendations

Testing the partner for this condition and genetic counseling

Interpretation Summary

This patient carries one copy of the DHCR7 c.964-1G>C mutation and is at least a carrier for Smith-Lemli-Opitz syndrome. The negative results for the other gene mutations do not completely exclude the possibility that this patient is a carrier for other diseases. Please refer to the accompanying tables for residual carrier risks in individuals for the corresponding ethnic groups, assuming a negative family history and the patient is not affected with one of the diseases.

What is Smith-Lemli-Opitz Syndrome

Characteristic facial features, microcephaly, intellectual disability, and behavioral problems (e.g. autism). Abnormalities of the heart, lungs, kidneys, gastrointestinal tract, fingers/toes and genitalia are also common. Variable severity of symptoms.

Dr. John Smith

Notifications



Abnormal Result Found Abigail Smith. Sample #23802830



Tests



Order



Name	DOB	Sample	Test	Date	Physician	Status	Res	Vio
Abigail Smith	1/12/83	23802830	Next Step	1/12/16	Smith, John	Final	Y	
John Adams	1/5/80	83092803	Next Step	1/12/16	Smith, John	Final	N	Y
George Hoover	2/12/78	10838275	Next Step	1/12/16	Smith, John	Final	Y	Y
Katherine Washington	1/12/82	66799300	Next Step	1/12/16	Smith, John	Final	Y	Y
Quincy Cleveland	3/24/84	20814423	Next Step	1/12/16	Smith, John	In Progress	N	N

Requisition

Test Kit

Information

Peace of Mind

One test. 281 genetic disorders. Your peace of mind.

Introducing Next Step. Guiding you to insights about your baby's development.
The only screen backed with years of clinical research from Mount Sinai and researchers from around the world.

[More information](#)

Need to register a kit?

[Are you a provider?](#)

THE END

WE LOOK FORWARD TO
EXPLORING WAYS TO
PARTNER!!!