Moving Beyond Big Data and AI to Realize the Dream of Precision Medicine

January 23, 2020
Sema4: A patient-centered health intelligence company

Use data analytics to develop **predictive models** of human health

Define optimal individualized health course trajectories to **improve** patient outcomes.

Deliver information-driven insights to pharma/biotech companies to **accelerate drug development** of precision medicines.
Sema4: Patient-centered predictive health company

- Spun out of Mount Sinai, a premier research and patient care organization and one of largest healthcare systems in US, in 2017
- Interdisciplinary team of scientists, data engineers, and clinicians, transforming future of healthcare through data-driven insights (~850 Employees; ~150 PhD/MD level scientists)
- One of largest clinical genomics labs in the world
  - >150,000 advanced DNA tests run/year
  - Test volume more than doubling yearly
- Founded on idea that more information, deeper analysis, and increased engagement will improve diagnosis, treatment, and prevention of disease
Healthcare decisions are binary, ignoring that every individual is unique and therefore requires unique healthcare recommendations.

Characterizing Disease Status Over Time in Patient Population

Individual patient, tracked over time, defined as well or sick based on simple, binary biomarker.
With increasing amounts of data from advanced technologies generated on patients, we can do better.
Our AI-driven models make maximal use of individual patient data, in the context of the digital universe of data to enable more informed and accurate decisions.
Rapid technology advances → massive scales of data informing on individuals

Only technology I am aware of that is moving at super Moore’s law speed

Advanced Materials

3D Printing

The “Internet of Things”
What does it mean to move at Super Moore’s Law pace? Well consider moving at a Moore’s Law pace…
Technologies like NGS have led to enough new knowledge in the past decade that everyone would benefit from sequencing today.

- **Carrier of Mendelian Mutations**: Virtually everyone carries loss of function mutations that have the potential to cause serious disease.
- **Fetus with Severe Genetic Lesions**: Roughly 1 in 100 babies will harbor severe genetic lesions that cause catastrophic illness.
- **Child with Genetic Condition**: Roughly 2 in 100 children harbor mutations that result in failure to thrive conditions.
- **Cardiac, Neuro, Heme**: Virtually everyone carries loss of function mutations that can impact drug response.
- **Pharmacogenomics**: Up to 20% of the population carries variants for blood clotting, heart disease and dementia.
- **Heritable Cancer**: Heritable forms of cancer affect up to 10% of the population (breast, ovarian, prostate, colon, etc.).
Sema4 conducts advanced genetic testing during key stages in patients’ lives

**Focus of these materials**

**Pre-pregnancy & Pregnancy**

- **Carrier Screening:**
  - Enhanced reports
  - Personalized residual risk
- **NIPT:**
  - Targeted and expansive screening
- **Prenatal:**
  - Diagnosis for high risk pregnancies
  - Cytogenomic analysis
- **IVF:**
  - Reference agreement for PGT-A and PGT-M

**Pediatric**

- **Diagnostic panels:**
  - Targeted and expansive screening
- **NIPT:**
  - Targeted and expansive screening
- **Prenatal:**
  - Diagnosis for high risk pregnancies
  - Cytogenomic analysis
- **IVF:**
  - Reference agreement for PGT-A and PGT-M

**Adult/Oncology**

- **Hereditary Cancer:**
  - Multigene panels across cancer types
- **Molecular Oncology:**
  - Solid Tumor and Heme Onc
  - Somatic and Germline profiling
  - Whole transcriptome profiling
- **Pharmacogenomic Panels**
- **Genomic Health Screening**

**Expanded Carrier Screening (> 500 Genes)**

Our Expanded Carrier Screen provides insight into carrier status to help patients make informed family planning choices.

- Conditions covered span: cardiovascular, endocrine, hematologic, hepatic, immunodeficiency, metabolic, neurological, pulmonary, renal, and skeletal conditions

**Natalis:**

- Supplemental newborn screening

**Diagnostic panels:**

- Hearing and vision loss
- Cardiac defects
- Primary immunodeficiency/IBD
- Neurodevelopmental panels including Noonan syndrome and ASD
- Skeletal dysplasias/Limb defects
- Microcephaly
- Diagnostic Exome

**Hereditary Cancer (>100 Genes)**

Genetic testing for hereditary cancer can help determine if a patient carries a genetic change that increases their risk for certain cancers.

Testing panels analyze genes associated with brain, breast, colon, melanoma, ovarian, pancreatic, and prostate cancers

**Solid/Liquid Tumors (Whole Exome/Transcriptome)**

Designed to help identify appropriate targeted therapies and clinical trials for patients with solid tumors.

- 20+ genes found in
- 15+ guidelines
- 30+ targeted therapies that are approved by the FDA
- 15+ genes included in
- 800+ clinical trials

*Majority of testing volume; additional detail on reproductive health tests on following slide
Convenient At Home Genetic Testing, Education & Support

- Patient Online Education
- Video Pre-test
- Pre-test Genetic Counseling
- At-home Saliva Test Kit Shipped
- Patient Portal Tools (Test Tracking & Resources)
- Post-Test Genetic Counseling
- Video Post-Test

- Education
- Testing
- Support
In areas like cancer, sophisticated data matched with sophisticated AI and machine learning approaches to inform diagnosis and treatment.

Patient tumor and germline are profiled, key drivers identified, tumor constructed in avatar models, those models taken through HTS for identification of drug cocktail.
Sema4: driving differentiated insights

Track Samples

Review Results
- Clinical trials
- Therapy, prognosis
- VUS

Interactive Results & Analytical Updates
- FDA approved therapies
- Clinical trials eligibility
- Relevant publications
- Practice level statistics

Drill Deeper/Unique Sema4 Modeling†
- Pathway driven recommendations to further personalize treatment decisions

Patient Journey & Cohort Builder
- Compare to relevant cohorts
- Review your patient files longitudinally

Place Order*
Equity in Comprehensive Testing Starts With Affordable Access

Robust In-network Payer Contracts (covering > 200 million lives in the U.S.)

Align with our patient and provider focused goals

Support for providers
- Workflow integration
- Dedicated Customer Success Team
- Ethical billing practices
- Expansive pre-authorization services
- Digital tools to engage results easily and securely
- Proactive genetic counseling

Support for patients
- National payor network
- Patient-friendly billing policies
- Benefits investigation service
- Digital tools keeping data secure and patients in control
- Proactive genetic counseling
- Affordable discounted or free rates for those in need