A strategic framework for an international system for genetic information to improve rare disease detection

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7,000 rare diseases have an impact on 350 million people worldwide

1 in 11 Americans will have a rare disease in their lifetimes

3 out of 4 rare disease patients are children, many affected for the duration of their lives

1 in 4 patients wait 4 years for a diagnosis, and average diagnosis delay is 1.5 years

50% of rare disease patients will not receive an accurate diagnosis

The goal is to establish an international rare disease detection platform to integrate:
- genetic information,
- biomarker information,
- phenotype and
- Geo/social information
In order to establish this platform:

**Incorporate appropriate technologies**, such as an artificial intelligence engine and advanced data storage systems.

**Utilize strategic partnerships**, bringing together science, computing, genomics, ethics, policy, social media, healthcare, pharmaceutical and medical communities.

**Involve patients and patient associations** in establishing this rare disease detection platform.

**Mutually establish time sequenced steps** by collaborative partners.

**Develop a cohesive strategy** that incorporates partners and addresses ethical and data safety concerns.

**Integrate therapy** as this cannot be ignored – detection must predicate improvement in people’s lives.

Linking the road map for rare disease detection to the broader understanding of individualized medicine will benefit patients with rare diseases, and the medical and scientific communities.
How to build the AI ecosystem:

Cross border partnership

1. **With:**
   - **Patients** – start with the needs of patients
   - **Health care system integration** – health care data needs to be in a common e-language
   - **Regulators** – bring regulators to the table
   - **Pharma** – by contributing to the AI health ecosystem, all companies will benefit

2. **Through:**
   - **Partnership:** private-public – the ‘win here’ is the health of patients
   - **Physical infrastructure** – the physical structures to support Big Data are critical
   - **Common international standards, methods and structures**
   - **Sustainable development**
     - Sustainable business development (e.g. public/private venture investing)
   - **Technical infrastructure**
     - AI engines need pure fuel
       - high quality, ethically compliant, health data
   - **Talent:** Build, retain and diversify talent in health
     - New sector invites **new training opportunities**
     - **Women in science** is fundamental
     - **Equity in science** across populations and people
     - Private sector needs to **support academia** to train and retain
       - E.g. private-academic cross hires
Conclusion

Capacity building agenda.

A multi sector outcomes based plan
• Patients and advocacy groups
• Health care
• Academia
• Artificial intelligence
• Policy
• Funders
• Biotech
• Pharma
• Social media

Phase 1 = Year 1 – the plan

Phase 2 = Year 2 – funds and infrastructure

Phase 3 = Year 3 – activation