Genomics adoption in the next 10-20 years

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Financial Disclosure

- Unified Patient Network, Inc. (employee)
- Consultant to:
  - MyOme, Inc.
  - Nest Genomics
  - Singular Genomics
  - X-Therma, Inc.

- Will not be discussing any products or services of these companies.

Obstacles removed over next 10-20 years

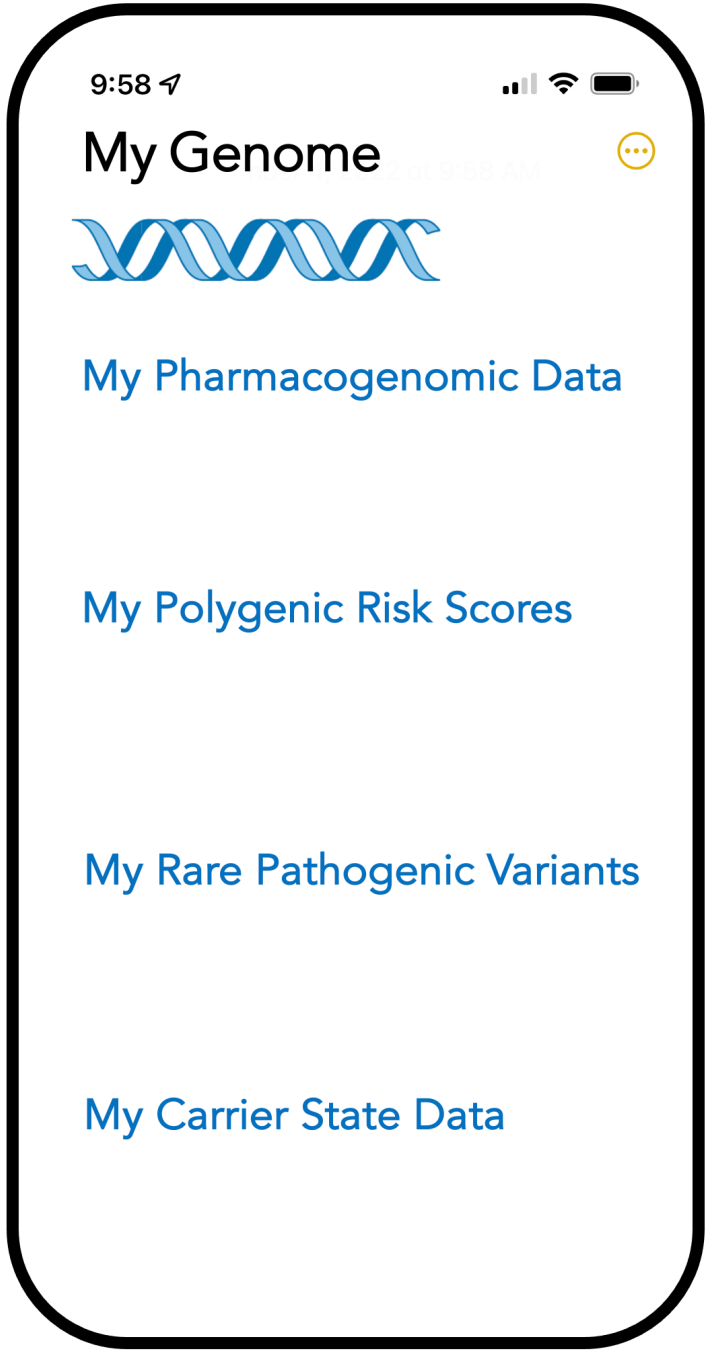
- Cost of genome sequencing & analysis will FINALLY get to affordable level with clinically useful turn-around times (TAT)
  - Ultima and other new competitors (“$100 genome”)
  - Illumina <$200/genome (list price reagents only)

- Genome sequencing (GS) will become the universal genetic test – (“Once and done”- sequence once, analyze repeatedly
  - GS diagnostic yield higher than gene panels, and VUS rate lower! (Rehm et al., medRxiv, Sep 2022)
  - Accurate detection of triplet repeat expansion diseases from GS (Ibanez et al., Lancet Neurology, March 2022)
  - TAT reduced to 24-72 hrs for all GS and analysis

When in lifespan will GS be done?

- Preconception (parents) – replace expanded carrier screening (+ ACMG SF & PGX)
- Prenatal – full GS from cfDNA or fetal cells
- NBS – many pilots ongoing now (GUARDIAN in US, 100K newborns; Genomics England, 100K newborns/yr)
- NICU/PICU (all undiagnosed disease patients)
- ASD and other neurodevelopmental disorders
- Germline + tumor GS in all cancers

- Annual “genome check-up” to coincide with annual physical exam



When will every person have instant access to their genome health reports via cell phone?

Eric Topol, Ground Truths, 9/11/22

Current missed opportunities

- Trio GS currently being done & reimbursed for NICU and some pediatric neurodevelopmental disorders, but parental genomes not being analyzed and used for future health benefit – ***"Lost genomes"***

- US far behind UK in genomics *implementation*
  - Genomics England (2013)- 100,000 GS rare disease & cancer
  - "Our Future Health" (2022) - 5 M volunteers to test role of polygenic risk scores (PRS) in preventing common adult diseases
  - *"This is unlike the National Health Service in the United Kingdom, the country leading the world in genomics, which has a major division known as Health Education England, that is responsible for educating clinicians about genomics".* E.Topol, 9/11/22