

# OPENING THE FLOOD GATE OF RESULTS, ARE WE READY, AND HOW WILL WE HANDLE THIS IN HEALTH CARE?

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# POPULATION GENOMIC SCREENING AS A PREVENTATIVE HEALTH MEASURE



With the decline in cost of genomic testing, where is the tipping point for population screening vs risk-based testing?

# DEMOCRATIZING GENOMIC TESTING: GENETIC TESTING AS A TOOL

## Genetics as a Tool

- Polygenic risk scores
- GCRA
- PGx
- DTC validation
- Carrier Screening

## Genetics as a Clinical Subspecialty

- Classic Genetic syndromes
- Metabolic Genetics
- Rare disease Genetics

Clinical Exome/Genome  
As cost goes down will move to general tool

## Radiology as a Tool

- Standardized diagnostic tests
- X-ray, CT, MRI

## Radiology as a Subspecialty

- Expert level interpretation
- Interventional procedures

## Cardiovascular as a Tool

- Standardized diagnostic tests
- EKG, Echo, stress test

## Cardiology as a Subspecialty

- Expert level interpretation
- Interventional procedures

# LIFESPAN TESTING: ONE TEST MODEL

*Genomic Testing* → *Precision Medicine:*  
*A Path To A Lifetime Patient Relationship*

**Patient Retention: A single genomic test performed today is incorporated into a lifetime of healthcare management.**

**Precision Medicine: Genomic tumor profiling and residual disease monitoring guide therapy decisions and access to clinical trials.**

INTEGRATED LONGITUDINAL HEALTHCARE

Newborn/Pediatric

Prenatal Dx  
Newborn Screening  
NICU/PICU Dx  
Pediatric Rare Disease

Early Adult

Preventative Polygenic Risk Score  
ACMGG 78\*  
Carrier Screening

Later Adult

Pharmacogenomics  
Adult-Onset Disease Dx  
Epigenetic Modification Dx

\*American College of Medical Genetics & Genomics ( incidental findings e.g., hereditary cancer syndromes, cardiovascular disease)

# WHAT IS NEEDED TO ACHIEVE POPULATION SCALE GENOMIC SCREENING?

## Critical Ingredients:

- National buy-in
- Community informed processes
- Integrated clinical decision support for management
- Informatics infrastructure
- Educated workforce
- Simplified testing
- Data sharing mechanisms
- Patient empowerment
- Enhanced protections (GINA), privacy, security
- Funding



RECIPE  
— for —  
SUCCESS

# WHO AND HOW WOULD THE RESULTS BE HANDLED?



Would testing be centralized like newborn screening within state labs?

Would Health departments be responsible for notifying of positive results?

What would trigger the interpretation, at what stage of life, what would be considered actionable and when?

Specialty Care

This would require a chaotic network of referrals and often great delays in being seen

Primary Care

# WHY THE PRIMARY CARE WORKFORCE MAKES THE MOST SENSE- ORCHESTRA CONDUCTORS OF HEALTH



The Primary Care Workforce (Pediatrics, Family Medicine, Internal Medicine, Obstetrics and Gynecology)

First line medical care, lowest access barrier

Available across geography

Care provided across the continuum of life, age span, multigenerational with broad scope of practice

Home of Preventative Medicine

Multidisciplinary care models-pharmacists, nutritionists, therapists...genetic counselors

Patients have already expressed this preference for discussing genetic testing results

# PRIMARY CARE SCOPE OF PRACTICE ALIGNS WITH GENOMIC SCREENING

Topic:	Test:
Preventative Care	Risk Panels
Prescribing management	Pharmacogenomics
Routine cancer screening	Genetic Cancer Risk Assessment/MCED
Prenatal care	Prenatal Carrier Risk/NIPT
Newborn care	Newborn Screening
Chronic Disease Management	Polygenic Risk Scores/Environmental Influences



# SCREENING LIVES IN PRIMARY CARE

Screening	Who needs it?	Doctor
Clinical skin examination	Everyone 19+	Primary care or dermatologist
Pap test	Women 21+	Primary care or OB-GYN
Breast cancer screening	Women 40+	Primary care referral to diagnostic radiologist
Colon cancer screening	Everyone 45+	Primary care referral to gastroenterologist
Low-dose CT scan	Smokers who are 55+	Primary care referral to interventional radiologist
Sexually transmitted infection	Everyone 19+	Primary care or OB-GYN
HPV	Women 21+	Primary care or OB-GYN
Hepatitis C	People born between 1/1/1945 and 12/31/1965	Primary care
Osteoporosis	Women 65+	Primary care referral to endocrinology
Immunizations	Everyone	Primary care
Blood pressure	Everyone 19+	Primary care
Lipid panel	Everyone 19+	Primary care
Diabetes	Everyone with a body mass index (BMI) > 25	Primary care

Adapted from <https://www.nebraskamed.com/primary-care/13-preventive-screenings-why-theyre-important-and-who-needs-them-most>

## WORKFORCE ASSESSMENT READINESS?

Grade **D**

not ready

CAN PRIMARY CARE DO THIS?

YES

# PREPARING THE WORKFORCE

## Critical Needs

Time saving/efficiency measures

Knowledge

Confidence (both patient and provider)

Robust informatics infrastructure to support data integration, reanalysis, curated updates and portability

## Possible Solutions

Clear and concise “just in time” clinical decision support to manage results across the lifespan

Algorithms for all screened conditions integrated with CDS

Minimum viable product for supportive management and counseling

Enhanced referral system for management beyond primary care

Better EMR systems, ready for the needs of genomic medicine

# COMMUNITY INFORMED MODELS AND PREPARING THE PUBLIC

Who are the key interested parties?

Community/public

Clinicians

How do the key interested parties want this to happen?

What should the models look like-federal/state/local?

What models will be acceptable to the public and readily adopted (universal, population specific)?

How do we ensure diverse and equitable uptake of the models across populations with the US?



# WHO DECIDES?

## Box 1. **Wilson and Jungner classic screening criteria**<sup>1</sup>

1. The condition sought should be an important health problem. ?
2. There should be an accepted treatment for patients with recognized disease.
3. Facilities for diagnosis and treatment should be available. ?
4. There should be a recognizable latent or early symptomatic stage.
5. There should be a suitable test or examination.
6. The test should be acceptable to the population. ?
7. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
8. There should be an agreed policy on whom to treat as patients. ?
- ? 9. The cost of case-finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
10. Case-finding should be a continuing process and not a "once and for all" project.

## **Fire Hydrant Model**

Prep everything as possible in advance

Roll out in one massive event



## **Trickling Faucet Model**

Pick one high value, high evidence screen

A/B test roll out informed by the community (patients and clinicians)

Layer on additional screening tests when pilot phase is deemed successful.



HOW DO WE  
ROLL THIS OUT?