What Genetic Testing Issues Need to Be Addressed?-A Primary Care Perspective

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Urgent Genetic Testing Needs in Primary Care:

- DTC validation
- Hereditary Cancer
- Pharmacogenomics
- Prenatal Carrier Risk
- Newborn screening
- Chronic Disease and PRS
- Common Adult genetic disorders
  - Neuro
  - CV
  - Pulm
  - Endo

- Return of results from research studies
  - All of Us

How do we get genomics as another tool in the clinician toolbox?

Genetics as a Tool

- Polygenic risk scores
- GCRA
- PGx
- DTC validation
- Carrier screening
- Diagnostic testing

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

Cardiovascular as a Tool

- standardized diagnostic tests
- EKG, Echo, stress test

Radiology as a Subspecialty

- expert level interpretation
- interventional procedures

Cardiology as a Subspecialty

- expert level interpretation
- interventional procedures

Barriers to Implementation:

Testing:	Availability/cost/reimbursement of testing Slow data turnaround time for results
Clinical:	Few existing algorithms Lack of real-world/RCT data on outcomes and value Lack of patient access to services
IT:	Need for ongoing design for data management Inadequate decision support and point-of-care tools
Research:	Complicated consent and data sharing processes Bio-banking for genotype-phenotype discovery Systematic capture of results back to the patient (EHR)
Education:	Training current and future health professionals in genomics and precision medicine
ELSI:	Ethics (privacy, equity, incidental findings, decision making) Legal issues (advancing GINA)

Slide credit: Phil Empey

Electronic Health Record Needs:

Test orderables in the electronic health records

Discrete reporting of results

Clinical Decision Support to fire from discrete results

Cost/Reimbursement Needs:

Transparency in cost and reimbursement

Clear pathways for obtaining reimbursement

Reimbursement models that make sense

Out of pocket costs that are affordable

Guidelines and Education

Genomic Clinicians- clinicians not formally trained in medical genetics integrating genomics into routine practice and patient care

Need minimal competencies

- Ordering
- Pre and post test counseling
- Interpretation

Clear algorithms for who and when to test

- Unless eliminated by extreme low cost

Better integration into medical education

Scalability of tools

- More genetic counselors
- Reimbursable service providers
- New models for synchronous contribution
- New models for asynchronous contribution

Testing Needs:

Streamlined testing

- Cost of sequential testing almost as much as WES/WGS
- When do we eliminate step wise testing

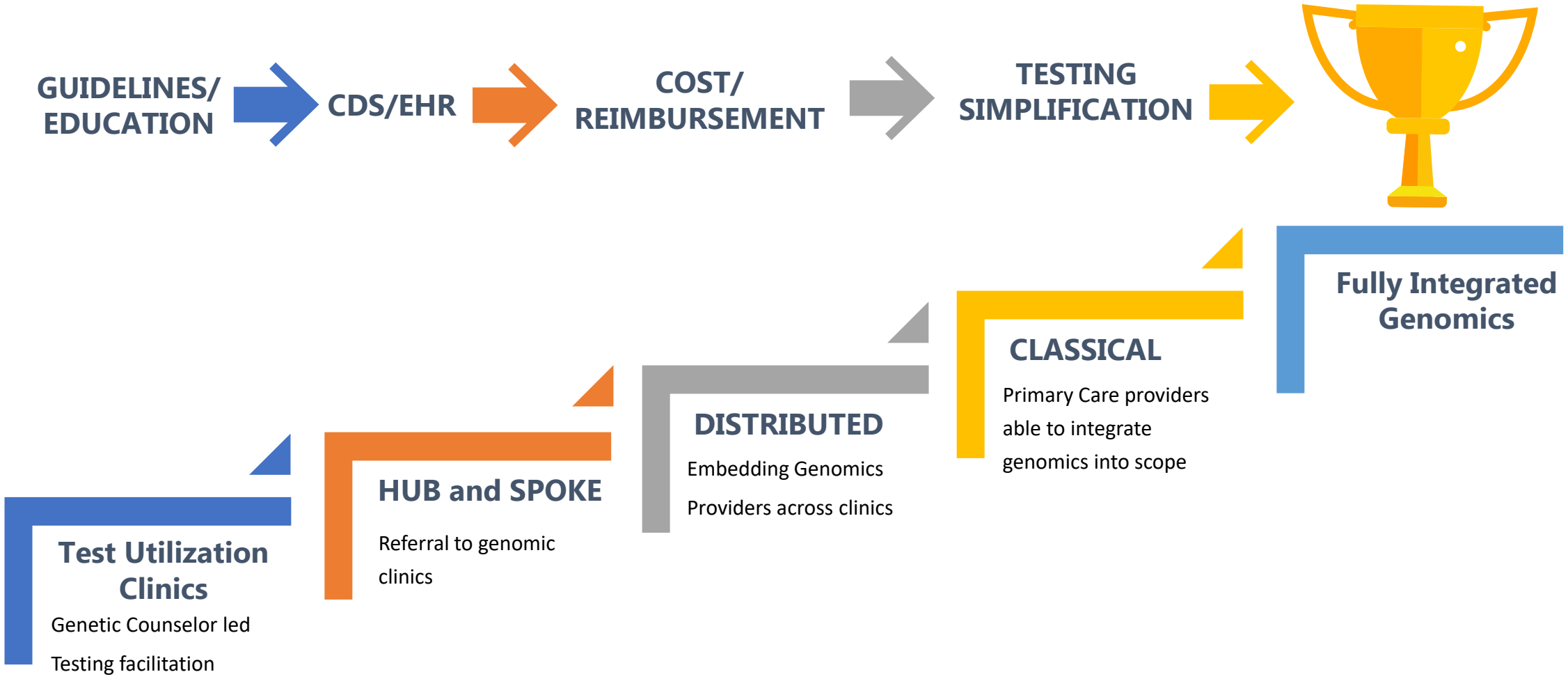
Exomes vs Genomes

Indication driven interpretation

Actionable secondary findings returned always

Ability to re query data for new indication or reinterpretation for initial indication

Coordinated step wise approach to the future



System Wide Solution-Early option solution:

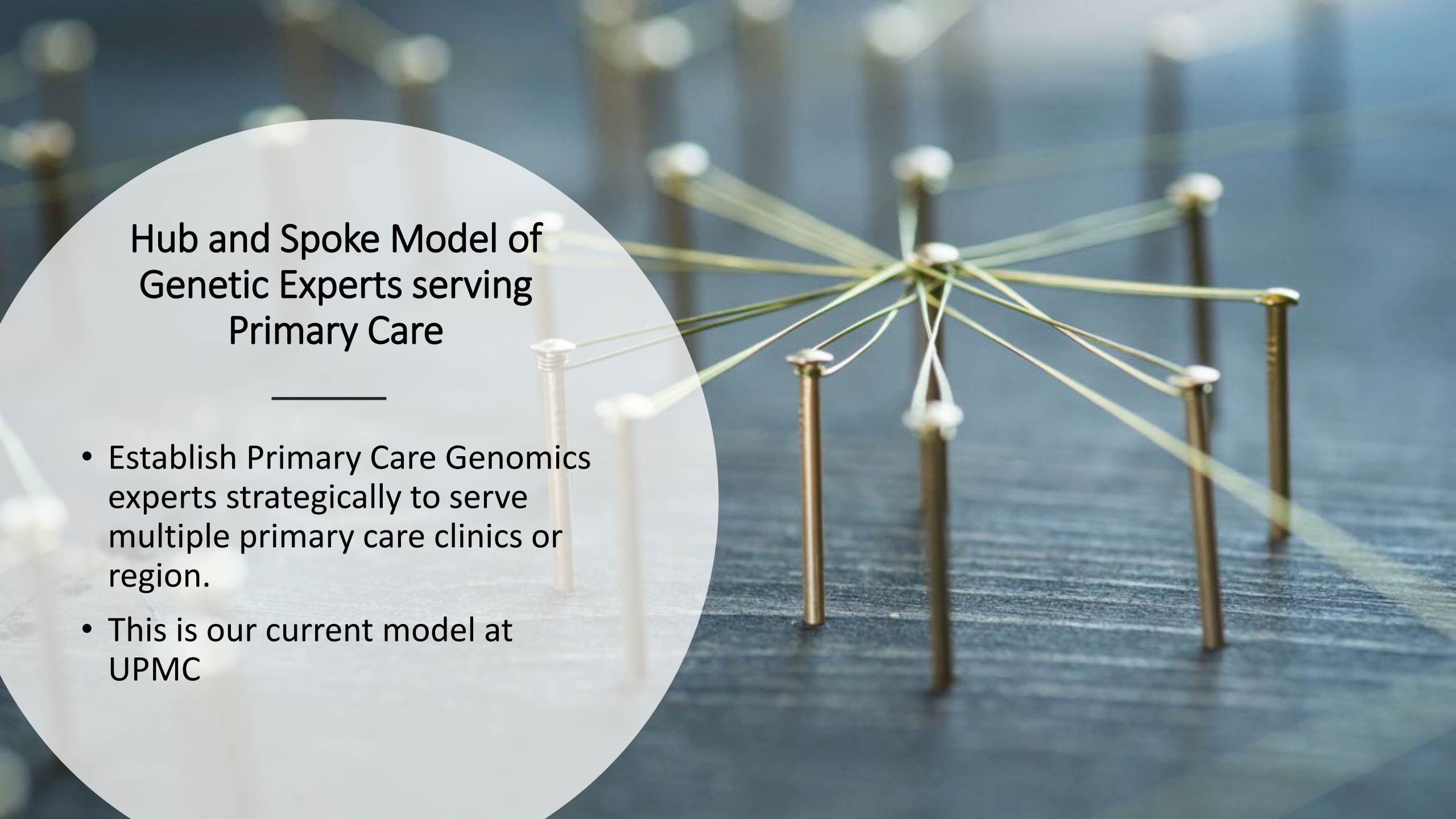
Genetic Testing Utilization Clinic Model in Partnership with the lab

- **Improper Test Choice**
- **Poor pre/post test counseling**
- **Bad Utilization Management**
- **Inaccurate test interpretation**
- **Testing not ordered due to intimidation factor**

Reimbursable
Genetic Counselor Facilitated
Testing Clinic

Utilization Management
Pre/Post Test Counseling
Best Test Selection

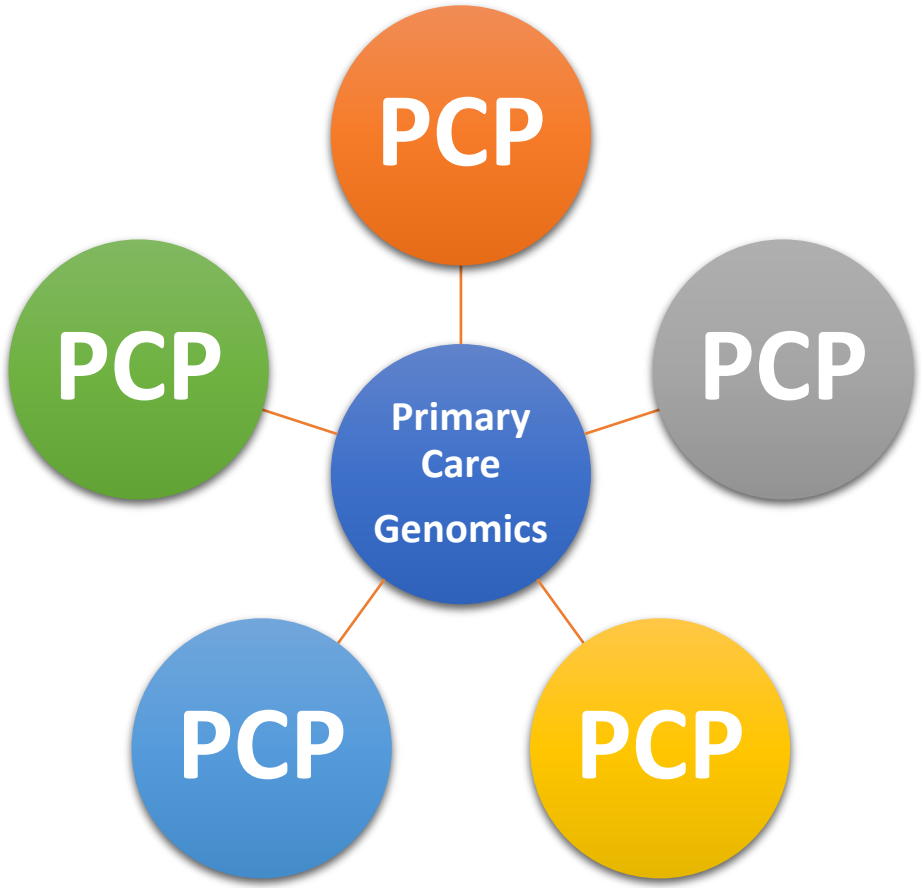
Support all clinicians and patients to get the best test, coverage, informed consent and interpretation.
Increase access to genetic testing as a tool in a facilitated manner.

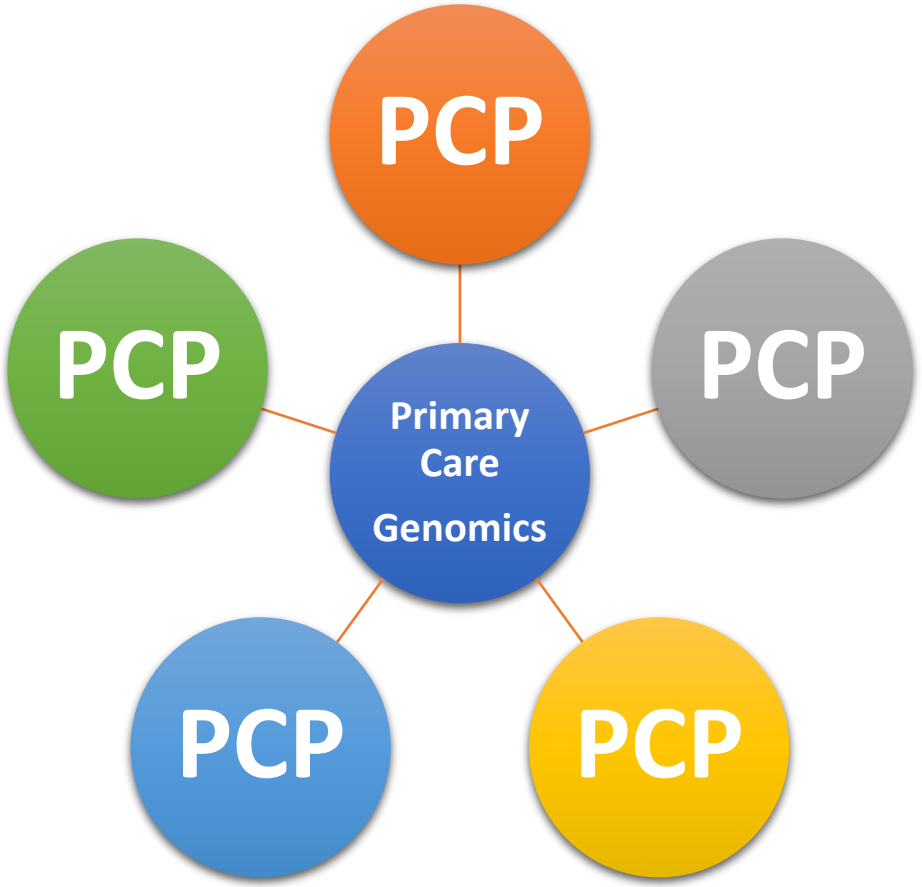


Hub and Spoke Model of Genetic Experts serving Primary Care

- Establish Primary Care Genomics experts strategically to serve multiple primary care clinics or region.
- This is our current model at UPMC

HUB and SPOKE Model





Strengths of Model

- Requires less total number of genomics in primary care experts.
- Provides a stop gap while education catches up.
- Opportunity to teach referring providers with each consult.

Weakness of Model

- External visit for patient
- Possible delay in access to service
- Unknown provider, outside of medical home
- Feels more like specialty consult as opposed to embedded in primary care.

Distributive Model of Seeding Genetic Counselors Across Primary Care

Strengths of Model

- Can scale CGC's quickly
- Offloads nuanced pre-test, post-test counseling and test selection/interpretation from physicians.
- Existing multidisciplinary model in primary care with Pharmacists, Social Workers, Nutritionists.
- Opportunity for real time genomics education to the providers by the CGC's.

Weakness of Model

- CGC's cannot yet bill independently to support their practice nor order testing independently.

Integrate Genetics into the Classical Family Medicine Model

- Family Medicine physicians are trained in a broad scope of practice
- FM trains and is comfortable managing 70% of all areas of medicine
- FM recognizes when to refer to specialty care

- Can we get FM to the 70% of genomics?
- How do we educate current trainees to achieve this goal?
- How do we support current practicing physicians to achieve this goal?

Strengths of Model

- Many genetic issues align well with primary care topics including prenatal carrier risk, genetic cancer risk and screening, pharmacogenomic impact on prescribing, complex chronic diseases (diabetes, cv, etc)
- Integrates genetics like all other subspecialty care in FM

Weakness of Model

- Current medical education lacks adequate genomics training
- Residencies do not have access to genomics curriculum
- Practicing physicians are overwhelmed and cannot conceive of adding expertise on one more topic
- FM still does not recognize that genomics is now, not 5 years from now.

Primary Care Precision Medicine Clinic

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UPMC Precision Medicine

Advancing best practices for the clinical delivery of Precision Medicine services across the health system.

Primary Care Genetics

Determining best practices and algorithms for the integration and advancement of genetics into primary care.

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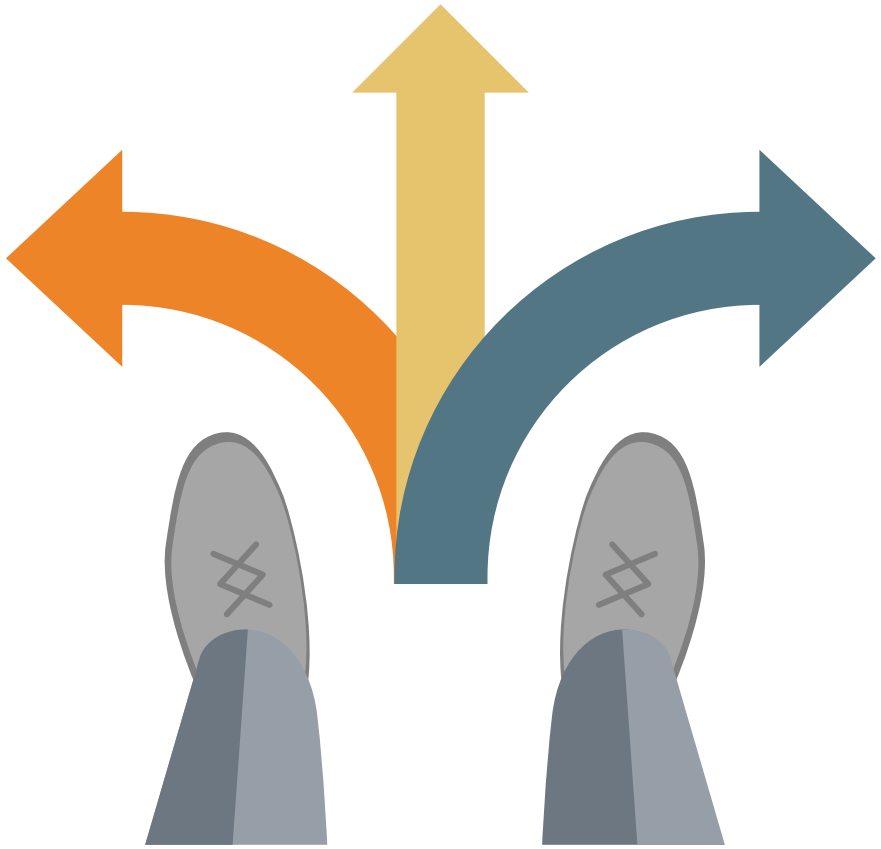

Genomics Education

Advancing genomics education across the system through the education of learners, providers and stakeholders.

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MEET THE NEEDS of CURRENT PATIENTS

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