Primary Care Modernization Design Group Process

- Goal: Make recommendation to the Practice Transformation Task Force as to whether this capability should be considered in the payment model.

Genomic Screen for CDC Priority Conditions

- Leading killers in the CT mirror those of the US, of particular concern are heart disease, cancer, and stroke, which screenings discussed here have the potential to affect.
- Screening is important when the disease being screened for is:
  - An important health problem
  - Not otherwise apparent
  - Screened via an approach that has good tools for finding it
  - Managed well after screening
- **BRCA 1/2** genomic screening of 50,000 patients in Pennsylvania was conducted as a pilot for the program:
  - 40% of women meet the criteria for testing based on family history, but do not receive it.
  - 40% of women do not meet criteria for testing, but were screened resulting in early detection.
- CDC Public Health Genomics office considers familial hypercholesterolemia, hereditary breast and ovarian cancer, and Lynch syndrome as tier 1 disease to screen for:
  - There are 10 genes to be screened for these diseases, therefore these are the screenings being examined by the model considered for PCM.
- Budget would be less than the cost for two preventive medicine visits:
  - Range of $300-$400 per test.
- These risks can be identified, interpreted, and managed now.
- The focus is on 10 genes because they drive these conditions, which there is sufficient data on.
- CT ethnic and racial diversity generally reflects that of the US and therefore this model may set the stage for understanding the impact these screenings would have nationally.

Comments and Questions

- Provider/Consumer: Is there data from a population health point of view that the screening can make a difference? What would be the estimated number of needed screenings in a defined period of time to prevent one death of adverse event?
  - Response – Provider: In the short run, avoidance of a heart attack under the age of 55, ovarian cancer at any age, breast cancer under the age of 55. The impact should be seen.
in a 5-10 year time period based on what is known of these conditions and management of impact in other settings are important to understand the impact.

- **Provider/Consumer:** 5% of people with breast cancer and 10% of people with ovarian cancer have this gene, but of these people how many go on to develop breast or ovarian cancer?
  - There is an 85-90% lifetime risk for cancer in women with BRCA 1 or 2
- **Provider:** Important to understand impact for patients, particularly management of screenings to manage patient expectations and understanding of risk. Providers need to be given the correct tools for this type of management. There would need to be a thoughtful approach on data.
- **FHC:** If the proportion of breast cancer related to these genes is only 3-5% of all disease cases can we only anticipate a drop in 3-5% of cases?
  - **Response – Provider:** There is an amount of misattribution associated with BRCA genes. An estimate considers as much as 6-10% attribution to BRCA 1 or 2 genes. It is only a slice of population burden, but focuses on those with risk of morbidity and mortality earlier in life.
  - **State:** For the population tested this has significant impact. Rare diseases collectively are a big percentage of the population (~10%). Genomic/precision medicine offers better care for that 10%. Important to create a pathway for precision medicine early as these screenings are further developed.
- **FHC:** Is cost for the test priced in the market place or through a particular center? Would providers need to seek suppliers of this test?
  - **Response – Provider:** Would want consistency through one supplier. Would be important to look at competition to compare prices. Color Genomics in Silicon Valley conducts these screenings for roughly $300-$400, Yale could deliver similarly.
- **Genomics Expert:** Reflex screening after the fact can identify hereditary origin and be an indicator for familial screenings. Will this occur?
  - **Response – Provider:** There is no reason why this cannot occur and there may be significant benefits from this. Those with the disease could get the screening from either their oncologist or PCP. For each case identified, there are potentially three family members who may be at risk for the gene.
- **Genomics Expert:** Is there support in the form of counselling only or would there be testing provided for at risk members?
  - **Response – Provider:** This depends on the details of the program, screening could be provided through the same mechanism for family members. Details would be dependent on the way the payments are structured.
- **Genomics Expert:** HIT requirements – is this going to be a technology agnostic EHR framework or one that will be used predominantly?
  - **Response – Provider:** Details would need to be worked out, but this could be done by mailing results to the provider and interoperability of EHR platforms.
  - **State:** If the design group recommends to the task force for adoption, we would work on detailing information exchange.
- **Genomics Expert:** Would be important to ensure consent is received upfront for additional screening programs, particularly important as additional genes are considered for testing.
- **State:** Is there data on whether confirmatory testing is conducted after the first test?
Response – Provider: There is still debate on whether secondary analysis needs to be done. Most labs consider a first test and secondary test to confirm the result is correct and this is included in the cost mentioned. Setting a high threshold to avoid false positives would be important for the screening program. The number of confirmable true positives is growing and this program could include ongoing analysis.

Consumer: Regarding education – one of the problems for newborn screening is for pediatrics physicians to understand that this is a screening test. Is there something that would be done to help PCPs understand this?

Response – Provider: Yes, would like to ensure that materials are clear and reviewed by a diverse set of audiences. Materials for patients and families tend to be helpful for PCPs as well. Education and support has to go out to any providers engaged, as well as patients and families.

Consumer: What were challenges in the Pennsylvania pilot screening program and how would these apply to CT?

Response – Provider: People were initially not convinced in the value of screening and there were concerns regarding implementation being burdensome. Geisinger providers concluded that the risk being identified for patients may not have ordinarily been otherwise. There was early resistance, but later on there was positive feedback on the program.

Next Steps

Recommendation to task force:

Provider: Worthy to consider, should be prioritized based on other recommendations.

Genomics Expert: Support for moving forward

Researcher: In support and questions helped clarify.

Consumer: In support

Provider/Consumer: In support, but support for those tested needs to be focused on as well. Emotional support post diagnosis should be considered.

Consumer: In support