A Clinical Genomics Educational Opportunity:

# Monthly GenomeFIRST Case Conference

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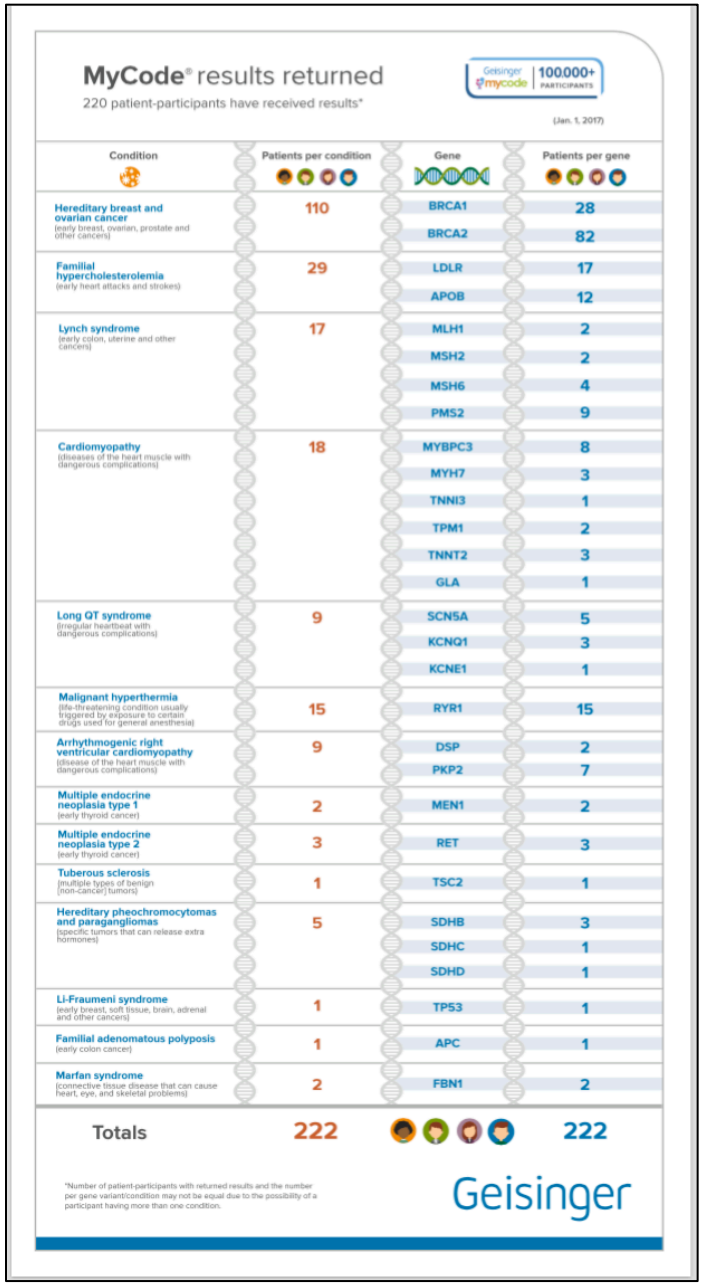
Geisinger Health System

# Geisinger Health System

- Academic/Clinical Translation
- Large health care provider for Central and Eastern Pennsylvania as well as New Jersey (Atlantic City)
- Use of the MyCode /GenomeFirst program to return clinically actionably results in a population-based study- over 125,000 enrolled



**MyCode® results returned**

220 patient-participants have received results*

Geisinger mycode 100,000+ PARTICIPANTS

(Jan. 1, 2017)

| Condition | Patients per condition | Gene | Patients per gene |
|---|---|---|---|
| Hereditary breast and ovarian cancer (early breast, ovarian, prostate and other cancers) | 110 | BRCA1 | 28 |
| | | BRCA2 | 82 |
| Familial hypercholesterolemia (early heart attacks and strokes) | 29 | LDLR | 17 |
| | | APOB | 12 |
| Lynch syndrome (early colon, uterine and other cancers) | 17 | MLH1 | 2 |
| | | MSH2 | 2 |
| | | MSH6 | 4 |
| | | PMS2 | 9 |
| Cardiomyopathy (diseases of the heart muscle with dangerous complications) | 18 | MYBPC3 | 8 |
| | | MYH7 | 3 |
| | | TNNI3 | 1 |
| | | TPM1 | 2 |
| | | TNNT2 | 3 |
| | | GLA | 1 |
| Long QT syndrome (irregular heartbeat with dangerous complications) | 9 | SCN5A | 5 |
| | | KCNQ1 | 3 |
| | | KCNE1 | 1 |
| Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia) | 15 | RYR1 | 15 |
| Arrhythmogenic right ventricular cardiomyopathy (disease of the heart muscle with dangerous complications) | 9 | DSP | 2 |
| | | PKP2 | 7 |
| Multiple endocrine neoplasia type 1 (early thyroid cancer) | 2 | MEN1 | 2 |
| Multiple endocrine neoplasia type 2 (early thyroid cancer) | 3 | RET | 3 |
| Tuberous sclerosis (multiple types of benign (non-cancer) tumors) | 1 | TSC2 | 1 |
| Hereditary pheochromocytomas and paragangliomas (specific tumors that can release extra hormones) | 5 | SDHB | 3 |
| | | SDHC | 1 |
| | | SDHD | 1 |
| Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers) | 1 | TP53 | 1 |
| Familial adenomatous polyposis (early colon cancer) | 1 | APC | 1 |
| Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems) | 2 | FBN1 | 2 |
| Totals | 222 | | 222 |

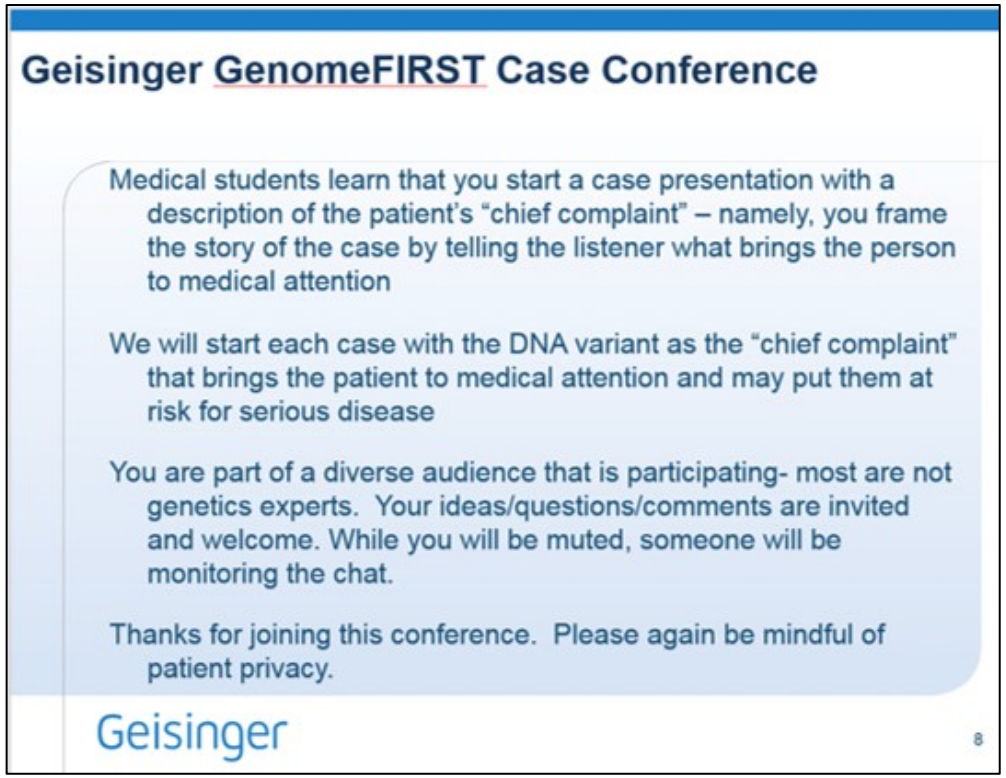
*Number of patient-participants with returned results and the number per gene variant/condition may not be equal due to the possibility of a participant having more than one condition.

Geisinger

**go.geisinger.org/results**

# GenomeFirst Case conference series

- ***The GenomeFIRST*** *Case Conference* is offered via Skype on the 2nd Thursday of each month at 8 AM Eastern.
- Cases are from Geisinger Health System's GenomeFIRST return of results program. What distinguishes this from other case conferences is that the "presenting complaint" is an incidental finding in the exome.
- This approach, i.e. the screening of existing genomic data for clinically important data, will become a common way for people everywhere to become aware of clinically important information in their genome in the years ahead.
- The discussion is decidedly aimed at a broad audience of non-experts. In 2017 we would like to open it up to even more interested practitioners (as well as other interested parties – for instance our external ethics advisory board and others).
- There is 1.0 hr of CME monthly through Geisinger Commonwealth School of Medicine (our newly affiliated medical school in Scranton, PA).
- Please contact **Chris Nicastro** at cmnicastro@geisinger.edu if you would like to be added to the monthly invite list



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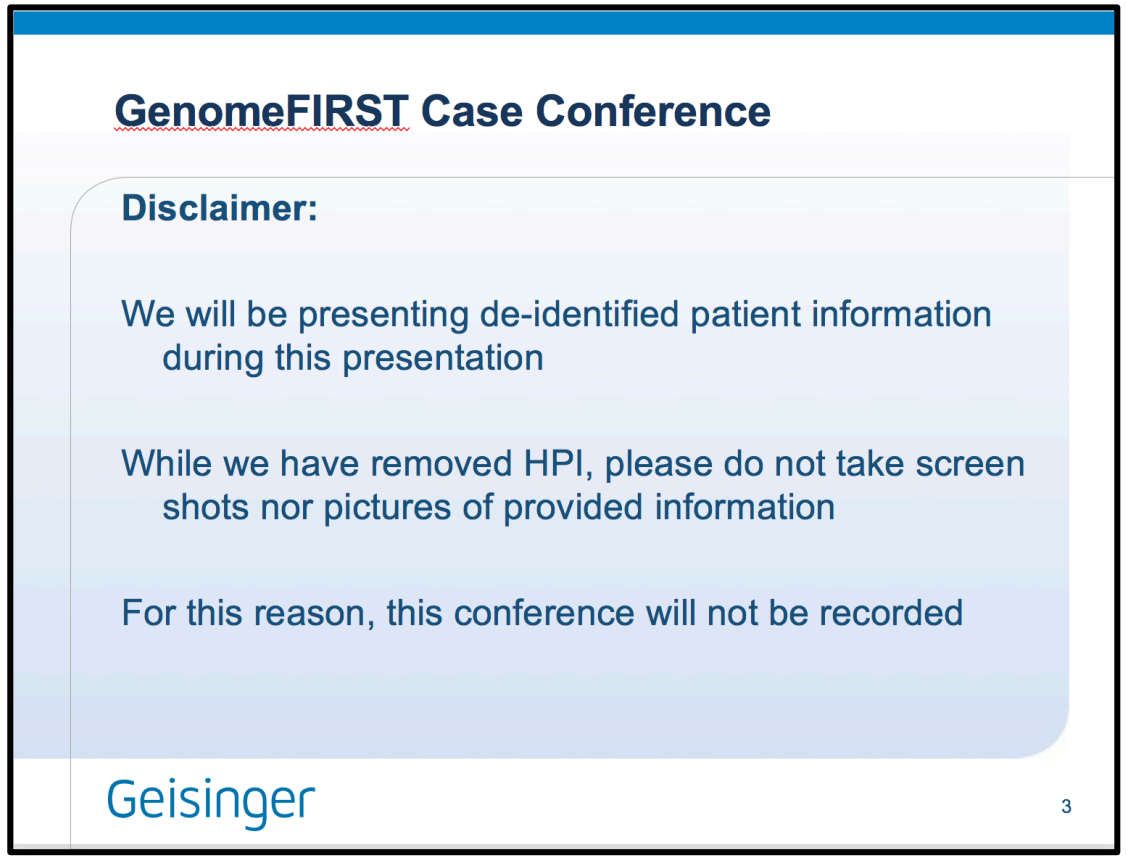

# GenomeFirst Case conference series

- Presentation of case as a medical student/resident/genetic counselor would
  - Highlight previous medical history, family history (more often to see no personal nor family history)
  - Review disorder management
  - Discuss teaching points – including asking students involved questions
  - Bigger picture view of how this fits into modern, integrated healthcare system
  - Have had trainees present when appropriate

- Themes from presentations
  - Value of screening- identification of early onset cancer disorders
  - Dual diagnoses
  - Cardiac cases with and without striking family histories
  - Psychosocial aspects of returning results to patients
  - Importance of cascade testing