

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 actionable results in a population-based study- over 125,000 enrolled

MyCode® results returned
220 patient-participants have received results*
(Jan. 1, 2017)

Geisinger mycode 100,000+ PARTICIPANTS

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

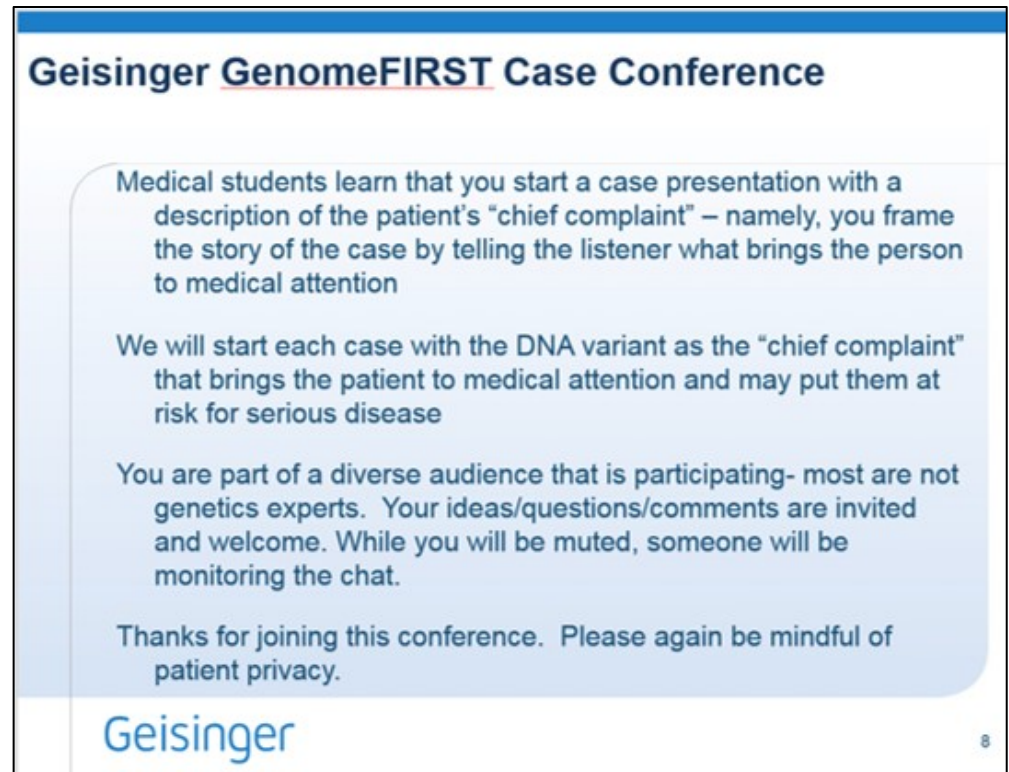
*Number of patient-participants with returned results and the number per gene variant/condition may not be equal due to the possibility of a participant having more than one condition.

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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



Geisinger GenomeFIRST Case Conference

Medical students learn that you start a case presentation with a description of the patient's "chief complaint" – namely, you frame the story of the case by telling the listener what brings the person to medical attention

We will start each case with the DNA variant as the "chief complaint" that brings the patient to medical attention and may put them at risk for serious disease

You are part of a diverse audience that is participating- most are not genetics experts. Your ideas/questions/comments are invited and welcome. While you will be muted, someone will be monitoring the chat.

Thanks for joining this conference. Please again be mindful of patient privacy.

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GenomeFirst Case conference series

Genetic
s January



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**GenomeFIRST Case Conference:
Dual Genetic Risk- Occam's razor
cutting both ways**

January 12, 2017



Please mute yourself

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GenomeFIRST Case Conference

Disclaimer:

We will be presenting de-identified patient information during this presentation

While we have removed HPI, please do not take screen shots nor pictures of provided information

For this reason, this conference will not be recorded

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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