Clinical Cancer Genetics Program

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Research Geneticist

Genetics Branch, Center for Cancer Research

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1

Background and Program Aims

- Concept proposal requested by CCR Clinical Director
  - Submitted November 2017
  - Funded April 2018
- Build on the existing clinical resources, infrastructure and initiatives already in place in the Genomic Healthcare Section of the Genetics Branch of the Center for Cancer Research
- Fill academic and continuing education gaps, and
- Provide the infrastructure for performing genetic testing for patients in CCR
- Conduct research in clinical cancer genetics.

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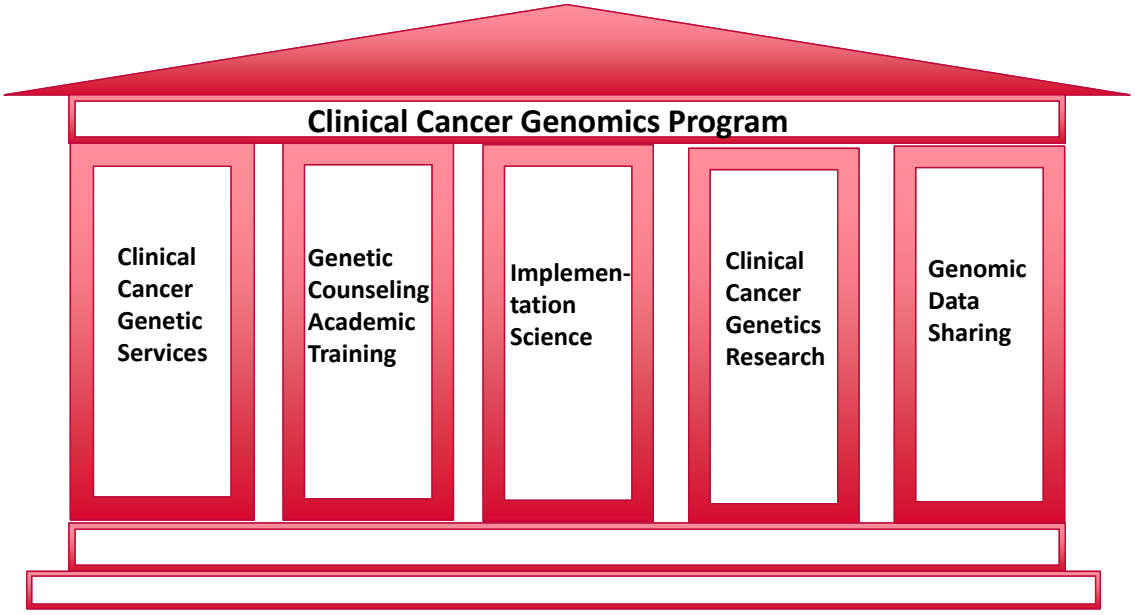
2

2

Program Components



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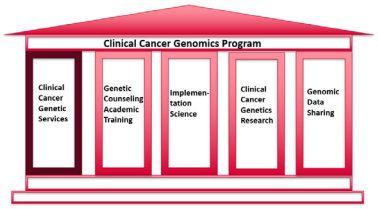

Clinical Cancer Genetic Services

Objectives:

- Expand capacity to provide a genetic clinical services for CCR and other NIH institutes as requested
- Provide support for ongoing NCI research studies that require genetic services or other genetic consultation i.e. during protocol development
- Provide long term follow-up services for individuals found to harbor a germline variant
- Provide clinical germline variant interpretations

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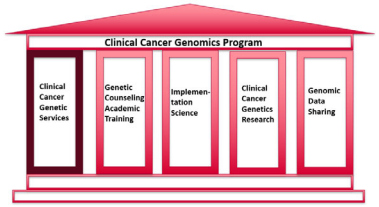


Clinical Cancer Genetic Services

- Staffed with 4 genetic counselors, one patient care coordinator, one cancer geneticist (vacant)

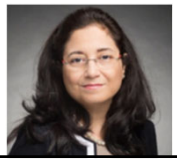
Grace-Ann Fasaye, ScM, CGC 

Michaela Taylor, MS, CGC 

Hermelat (“Hermi”) Mesfin, BS 

Alexandra Lebensohn, MS, CGC 

Yi Liu, MS, CGC 

Chimene Kesserwan, MD, FCAP, FACMG, Volunteer 

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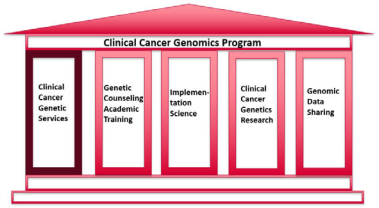

Clinical Cancer Genetic Services

- Perform genetic education and counseling
  - Collect and interpret personal and family medical histories, phenotyping
  - Determine if genetic testing is indicated, germline, somatic, or both
  - Select the optimal test to order and the laboratory to use
  - Provide education about the test, genes, somatic and/or germline, inheritance, risk management, resources, and research
  - Facilitate informed choices about genetic testing
  - Identify risks for family members, germline versus somatic testing
  - Help patients/families and their healthcare providers understand the medical, psychological, and familial implications of the genetic results

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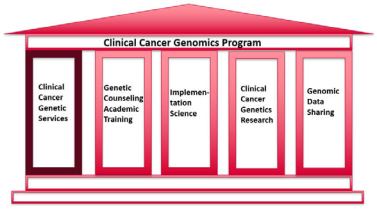
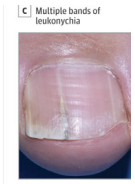


Clinical Cancer Genetic Services

- Phenotyping: Alex Lebensohn, MS, CGC
  - Patient phone encounter
    - Nail complaints, *BAP1* tumor predisposition syndrome
      - Assessed independently this finding in her patients
      - Presented the data to the team
      - Derm biopsied the nails N=47
      - N=41 (87.2%) with leukonychia, splinter hemorrhage, onychoschizia, and distal nail hyperkeratosis
      - Polydactylous involvement with onychopapillomas detected (38 of 39 patients [97.4%]).



Research

JAMA Dermatology | Original Investigation

Multiple Onychopapillomas and BAP1 Tumor Predisposition Syndrome

Alexandria Lebensohn, MS, CGC; Azam Ghafoor, MD; Luke Bloomquist, MD; Michael C. Royer, MD; Leslie Castelo-Soccio, MD, PhD; Kelli Kuraski, PA-C; Dzesta Hathaway, CRNP; Tenn Mugio, CRNP; Cathy Wagner, RN; Maria G. Agra, RN; Andrew M. Blakely, MD; David S. Schrump, MD; Ruffi Hassan, MD; Edward W. Cowen, MD, MHSc

IMPORTANCE *BRCA1*-associated protein (BAP1) tumor predisposition syndrome (TPDS) is a cancer genodermatosis associated with high risk of uveal and cutaneous melanoma, basal cell carcinoma, and multiple internal malignant neoplasms, including mesothelioma and renal cell carcinoma. Early detection of the syndrome is important for cancer surveillance and genetic counseling of family members who are at risk.

OBJECTIVE To determine the prevalence of nail abnormalities in individuals with pathogenic germline variants in *BAP1*.

DESIGN, SETTING, AND PARTICIPANTS In this prospective cohort study, individuals who were known carriers of pathogenic *BAP1* germline variants were consecutively enrolled between October 10, 2023, and March 15, 2024. Dermatologic evaluation for nail abnormalities was performed, including a history of nail abnormalities and associated symptoms, physical examination, medical photography, and nail biopsy for histopathology. This was a single-center study conducted at the National Institutes of Health Clinical Center.

MAIN OUTCOMES AND MEASURES Primary outcomes were the prevalence and spectrum of nail changes and histopathologic characterization.

RESULTS Among 47 participants (30 female [63.8%]; mean [SD] age, 46.4 [15.1] years) ranging in age from 13 to 72 years from 35 families, nail abnormalities were detected in 41 patients (87.2%) and included leukonychia, splinter hemorrhage, onychoschizia, and distal nail hyperkeratosis. Clinical findings consistent with onychopapilloma were detected in 39 patients (83.0%), including 35 of 40 individuals aged 30 years or older (87.5%). Nail bed biopsy was performed in 5 patients and was consistent with onychopapilloma. Polydactylous involvement with onychopapillomas was detected in nearly all patients who had nail involvement (38 of 39 patients [97.4%]).

CONCLUSIONS AND RELEVANCE This study found that BAP1 TPDS was associated with a high rate of nail abnormalities consistent with onychopapillomas in adult carriers of the disease. Findings suggest that this novel cutaneous sign may facilitate detection of the syndrome in family members who are at risk and patients with cancers associated with BAP1 given that multiple onychopapillomas are uncommon in the general population and may be a distinct clue to the presence of a pathogenic germline variant in the *BAP1* gene.

Supplemental content

Author Affiliations: Author affiliations are listed at the end of this article.

Corresponding Author: Edward W. Cowen, MD, MHSc, Dermatology Branch, National Institute of Arthritis and Musculoskeletal and Skin Diseases, National Institutes of Health, 10 Center Dr, MSC 1908, Bethesda, MD 20892 (cowenw@mail.nih.gov). Ruffi Hassan, MD, Thoracic and Gastrointestinal Malignancies Branch, Center for Cancer Research, National Cancer Institute, National Institutes of Health, Bldg 10, 10 Center Dr, Rm. 4E-5530, Bethesda, MD 20892 (hassanr@mail.nih.gov).

JAMA Dermatol. 2024;160(8):838-845. doi:10.1001/jamadermatol.2024.1804
Published online May 17, 2024.

838

jamadermatology.com

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Lebensohn et al. (2024). Multiple onychopapillomas and BAP1 tumor predisposition syndrome. JAMA Dermatology. PMID: 38759225

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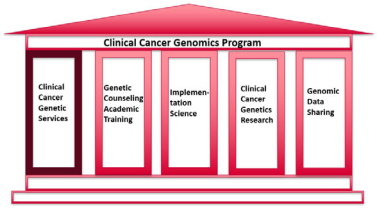

Clinical Cancer Genetic Services

- Established a centralized billing system, **refer to CCR SOP ADGC-6**
- Worked with Clinical Center DCRI to establish the CRIS Pedigree Tool
  - Worked with development team to design the application
  - Alpha and beta testing, assisted in initial orientation and launch
  - Worked with development team to design a patient portal for family history collection which would auto-populate the pedigree tool
    - DCRI never launched the program after testing

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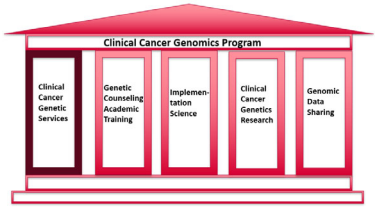
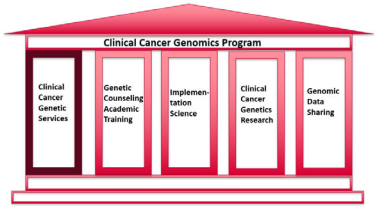


Clinical Cancer Genetic Services

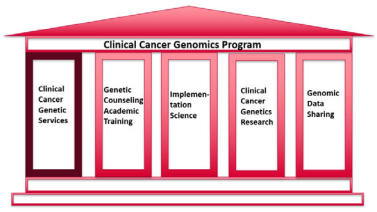
- From 7/10/2023-7/10/2024 the service saw 268 cancer genetic consults from across all CCR branches
- Genetic counselors are embedded as part of the team in five services: Inherited Gastric Cancer; Hematologic Malignancies; Kidney; Mesothelioma; Prostate
  - Pediatrics and Neuro Oncology-separate Genetic Counselor
- Support Tumor/Normal Whole Exome Sequencing (T/N WES) in Laboratory of Pathology
- From 7/10/2023-7/10/2024 the service saw 192 T/N WES consults from across all CCR branches



Clinical Cancer Genetic Services

- Requesting a consult-**DO NOT** use CRIS
- Email the NCI Cancer Genetics Consult Service at:

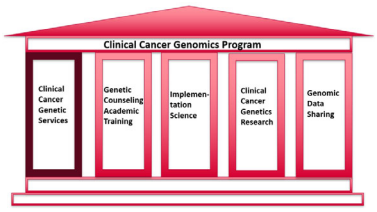
  NCI Genetics Consult Service <NCI_GeneticConsult@mail.nih.gov>
- The Clinical Cancer Genetics Program is **NOT** a Clinical Center recognized service
  - Clinical Center is concerned about confusion between our cancer genetics service and the NHGRI general genetics consult service
    - CCGP-529 patient notes, NHGRI-18 patient notes



Clinical Cancer Genetic Services

- Accept NHGRI clinical genetic fellows
- Accept genetic counseling students
  - Clinical rotations, Summer intensive
- Work with the Laboratory of Pathology tumor/normal whole exome sequencing (T/N WES)
  - Consent, pre/post genetic counseling
  - Germline variant interpretation
  - For details **refer to CCR SOP ADGC-5, includes checklist**
  - **For T/N WES consults email: TumorNormalWES@mail.nih.gov**

11

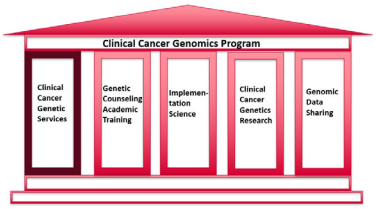


Clinical Cancer Genetic Services

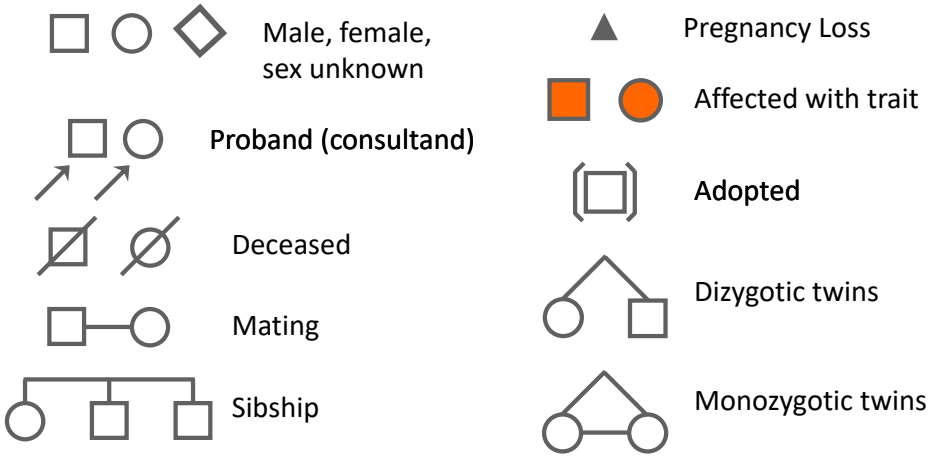
Pedigrees, what, why, how?

- Pedigrees are a graphic illustration of the family history
  - Includes ALL individuals in a family whether affected with disease or not
  - Maternal AND paternal lineages
  - Diseases (not just cancer) with age of diagnosis for each individual
    - Confirmation with medical records whenever possible
  - Benign conditions such as but not limited to colon polyps
  - Risk reducing surgeries
  - Current age and ages of death for deceased individuals
  - Minimum 3 generations
  - Race and ethnicity

12



Standard Pedigree Nomenclature



Bennett et al. 2023. Practice resource-focused revision: Standardized pedigree nomenclature update centered on sex and gender inclusivity: A practice resource of the National Society of Genetic Counselors. JGC, PMID: 36106433

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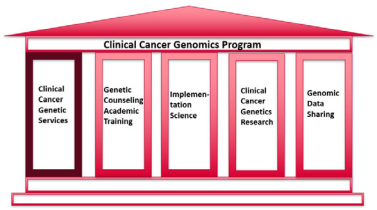

Clinical Cancer Genetic Services

Pedigrees, what, why, how?

- Facilitates the identification of genetic syndromes and whether a family is a candidate for genetic testing
- Aides the provider in establishing a presymptomatic diagnosis of a genetic disease
- Helps identify at risk individuals
- Helps to establish patterns of inheritance
- Illuminates social and biological relationships
- Helps inform the differential diagnosis
- Pedigrees are **NOT** stagnant, should be updated at each encounter

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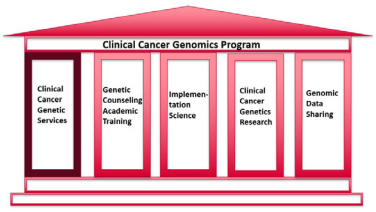
14

14

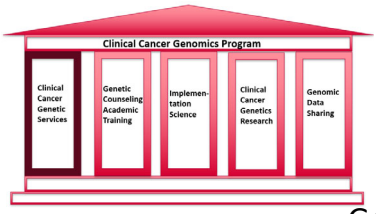


Clinical Cancer Genetic Services

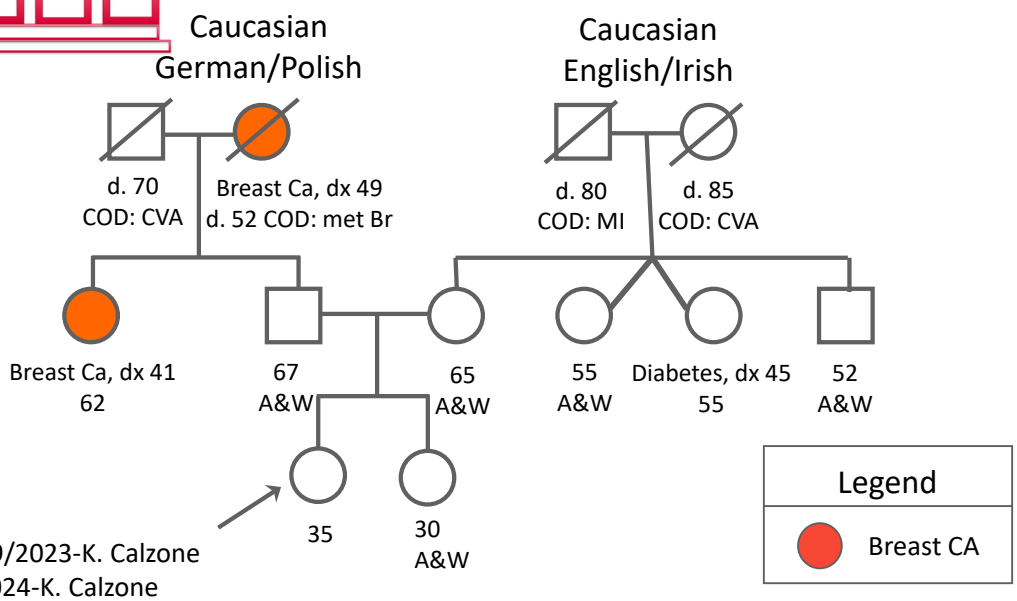
Pedigrees, what, why, how?

- Can be collected via questionnaire prior to an appointment
  - Questionnaires can then be used to create the initial pedigree
  - Provides a mechanism for patients to ask biologic family members about their health prior to their appointment
- Can inform the differential diagnoses
- Helps identify the most informative person to test (may NOT be your patient)
  - Individual affected with disease
- An easy way to learn how to collect pedigrees…take your own

15



Three-Generation Pedigree



Completed: 4/29/2023-K. Calzone
Updated: 5/1/2024-K. Calzone

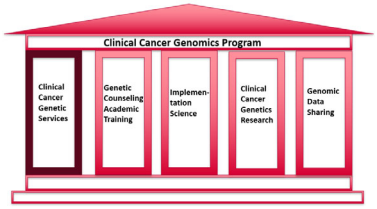
16

Poll Question

Do you update your patients' family history at each encounter (similar to medication list, problem list, etc)?

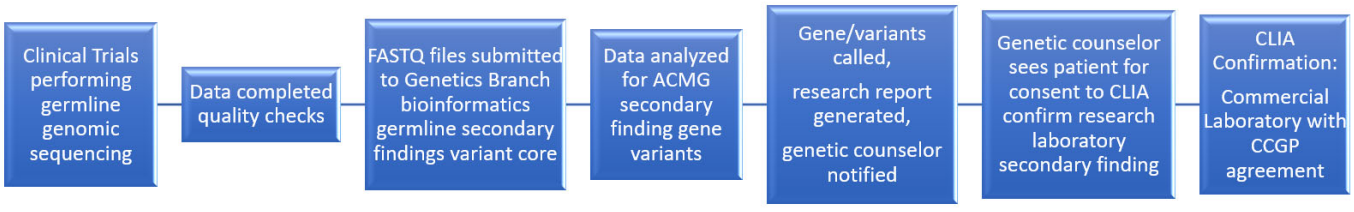
1. Yes
2. Sometimes
3. Never

17



Clinical Cancer Genetic Services

- IRB Secondary Findings guidance applies to new protocols, and existing protocols to which investigators add genomic sequencing by protocol modification, submitted to the IRB **after October 2022**.
- Secondary Findings Service-CCR proposal



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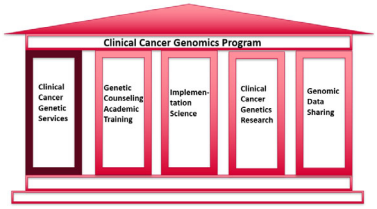

- Cancer Genes

Clinical Cancer Genetic Services

Phenotype	ACMG SF List Version	MIM Disorder	Gene	Inheritance	Variants to Report[a]
Genes related to cancer phenotypes					
Familial adenomatous polyposis	1.0	175100	*APC*	AD	All P and LP
Familial medullary thyroid cancer/multiple endocrine neoplasia 2	1.0	155240	*RET*	AD	All P and LP
		171400			
		162300			
Hereditary breast and/or ovarian cancer	1.0	604370	*BRCA1*	AD	All P and LP
	1.0	612555	*BRCA2*		
	3.0	114480	*PALB2*		
Hereditary paraganglioma-pheochromocytoma syndrome	1.0	168000	*SDHD*	AD	All P and LP
	1.0	601650	*SDHAF2*		
	1.0	605373	*SDHC*		
	1.0	115310	*SDHB*		
	3.0	171300	*MAX*		
	3.0	171300	*TMEM127*		
Juvenile polyposis syndrome	2.0	174900	*BMPR1A*	AD	All P and LP
Juvenile polyposis syndrome/hereditary hemorrhagic telangiectasia syndrome	2.0	175050	*SMAD4*	AD	All P and LP
Li-Fraumeni syndrome	1.0	151623	*TP53*	AD	All P and LP
Lynch syndrome (hereditary nonpolyposis colorectal cancer)	1.0	609310	*MLH1*	AD	All P and LP
		120435	*MSH2*		
		614350	*MSH6*		
		614337	*PMS2*		
Multiple endocrine neoplasia type 1	1.0	131100	*MEN1*	AD	All P and LP
MUTYH-associated polyposis	1.0	608456	*MUTYH*	AR	P and LP (2 variants)
NF2-related schwannomatosis	1.0	101000	*NF2*	AD	All P and LP
Peutz-Jeghers syndrome	1.0	175200	*STK11*	AD	All P and LP
PTEN hamartoma tumor syndrome	1.0	158350	*PTEN*	AD	All P and LP
Retinoblastoma	1.0	180200	*RB1*	AD	All P and LP
Tuberous sclerosis complex	1.0	191100	*TSC1*	AD	All P and LP
	1.0	613254	*TSC2*		
von Hippel-Lindau syndrome	1.0	193300	*VHL*	AD	All P and LP
WT1-related Wilms tumor	1.0	194070	*WT1*	AD	All P and LP

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Miller, D.T., et al. (2023). ACMG SF v3.2 PMID: 37347242.

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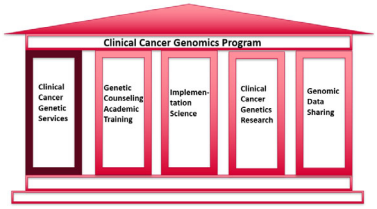

- Cardiac Genes

Clinical Cancer Genetic Services

Genes related to cardiovascular phenotypes					
Aortopathies	1.0	154700	*FBN1*	AD	All P and LP
	1.0	609192	*TGFBR1*		
	1.0	610168	*TGFBR2*		
	1.0	613795	*SMAD3*		
	1.0	611788	*ACTA2*		
	1.0	132900	*MYH11*		
Arrhythmogenic right ventricular cardiomyopathy (a subcategory of arrhythmogenic cardiomyopathy)	1.0	609040	*PKP2*	AD	All P and LP
	1.0	607450	*DSP*[b]		
	1.0	610476	*DSC2*		
	1.0	604400	*TMEM43*		
	1.0	610193	*DSG2*		
Catecholaminergic polymorphic ventricular tachycardia	1.0	604772	*RYR2*	AD	All P and LP
	3.0	611938	*CASQ2*	AR	P and LP (2 variants)
	3.0	615441	*TRDN*[c]	AR	
DCM	1.0	601494	*TNNT2*[d]	AD	All P and LP (See text)
	1.0	115200	*LMNA*[e]		
	3.0	617047	*FLNC*[d]		
	3.0	604145	*TTN*[f]		
	3.1	613881	*BAG3*		
	3.1	604765	*DES*		
	3.1	613172	*RBM20*		
	3.1	611879	*TNNC1*		
Ehlers-Danlos syndrome, vascular type	1.0	130050	*COL3A1*	AD	All P and LP

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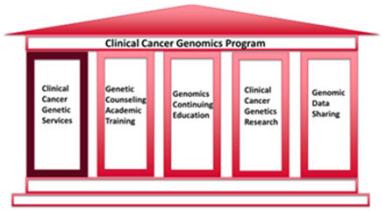

Clinical Cancer Genetic Services

- Cardiac Genes, cont

Phenotype	ACMG SF List Version	MIM Disorder	Gene	Inheritance	Variants to Report[a]
Familial hypercholesterolemia	1.0	143890	*LDLR*	SD	All P and LP
	1.0	144010	*APOB*	AD	
	1.0	603776	*PCSK9*	AD	
HCM[g]	1.0	192600	*MYH7*[b]	AD	All P and LP
	1.0	115197	*MYBPC3*		
	1.0	613690	*TNNI3*		
	1.0	115196	*TPM1*		
	1.0	608751	*MYL3*		
	1.0	612098	*ACTC1*		
	1.0	600858	*PRKAG2*		
	1.0	608758	*MYL2*		
LQTS types 1 and 2	1.0	192500	*KCNQ1*	AD	All P and LP
	1.0	613688	*KCNH2*		
LQTS3; Brugada syndrome	1.0	603830, 601144	*SCN5A*[b]	AD	All P and LP
LQTS types 14-16	3.2	616247	*CALM1*[g]	AD	All P and LP
		616249	*CALM2*[g]	AD	
		618782	*CALM3*[g]	AD	

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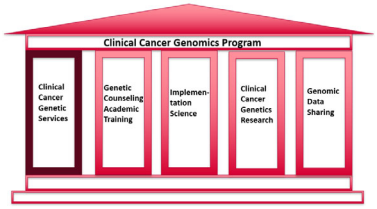

Clinical Cancer Genetic Services

- In-born Error of Metabolism Genes and Genes with MISC Phenotypes

Genes related to inborn errors of metabolism phenotypes					
Biotinidase deficiency	3.0	253260	*BTD*	AR	P and LP (2 variants)
Fabry disease	1.0	301500	*GLA*[h]	XL	All hemi, het, homozygous P and LP
Ornithine transcarbamylase deficiency	2.0	311250	*OTC*	XL	All hemi, het, homozygous P and LP
Pompe disease	3.0	232300	*GAA*	AR	P and LP (2 variants)
Genes related to miscellaneous phenotypes					
Hereditary hemochromatosis	3.0	235200	*HFE*	AR	*HFE* p.C282Y[i] homozygotes only
Hereditary hemorrhagic telangiectasia	3.0	600376	*ACVRL1*	AD	All P and LP
	3.0	187300	*ENG*		
Malignant hyperthermia	1.0	145600	*RYR1*[j]	AD	All P and LP
	1.0	601887	*CACNA1S*		
Maturity-onset of diabetes of the young	3.0	600496	*HNF1A*	AD	All P and LP
RPE65-related retinopathy	3.0	204100, 613794	*RPE65*	AR	P and LP (2 variants)
Wilson disease	2.0	277900	*ATP7B*	AR	P and LP (2 variants)
Hereditary TTR amyloidosis	3.1	105210	*TTR*	AD	All P and LP

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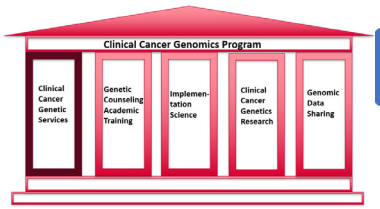
Miller, D.T., et al. (2023). ACMG SF v3.2 PMID: 37347242.

Clinical Cancer Genetic Services



Protocol
- IRB approved clinical protocol includes germline genomic correlatives that require adherence to IRB Secondary Findings Guidance

Research Laboratory
- Genomic analyses completed and data completed quality checks
- Research laboratory will submit FASTQ files to the Secondary Findings Bioinformatics Core for analysis for secondary findings

Bioinformatics Core
- The Secondary Findings Bioinformatics Core will analyze the data for the ACMG gene list current at the time of the analysis
- A research report will be generated indicated no gene variants detected OR the specific gene(s) and variant(s) identified.
- Research Report will be submitted to the Clinical Cancer Genetics Program (CCGP) for Genetic Counseling and CLIA confirmation

Genetic Counseling
- Research Report Received. The CCGP will notify the PI that X patient has been found to have one or more Genomic Secondary Findings
- The CCGP Patient Care Corrdinator will schedule the patient to be seen by a Genetic Counselor for consent for CLIA confirmation
- If consent is **NOT** given for CLIA confirmation the PI will be notified, no result will be disclosed and this will be documented in CRIS **without** specifying the gene or variant identified
- If consent **IS** obtained, a biospecimen will be collected and sent for CLIA confirmation to a commercial laboratory
- Once CLIA results are obtained the PI will be notified of the confirmation
- The patient will be notified of the CLIA results:
  -Cancer Genes via CCGP
  -Non-Cancer Genes via Genome Medical

- Utilize bioinformatician working with me on Genomic Data Sharing
- CLIA confirmation $250/gene category
- Deceased subjects, family members-04C0165
  - Connecting to deceased family members….TBD how??
- Negotiations ongoing with Dr. Khan, Genetics Branch Deputy Branch Chief and CCR leadership

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3

23

Poll Question

How do you submit a general cancer genetics referral?

1. Submit a genetics referral in CRIS
2. Email the Patient Care Coordinator Hermelat Mesfin
3. Email NCI_GeneticConsult@mail.nih.gov

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24

24

Poll Question

How do you submit a request for consent for Tumor/Normal Whole Exome Sequencing?

1. Submit a genetics referral in CRIS
2. Email the Patient Care Coordinator Hermelat Mesfin
3. Email TumorNormalWES@mail.nih.gov with the checklist

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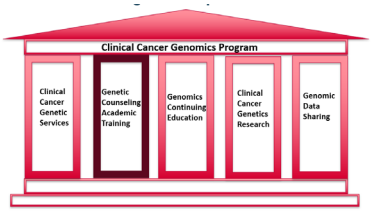
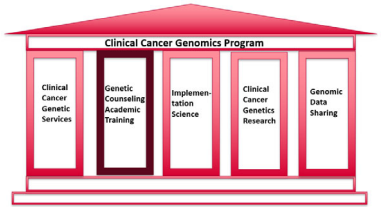


Genetic Counseling Academic Training

Objectives:

- Expand the existing training program for genetic counselors
  - Expand the cancer genetic content in the existing curriculum
  - Investigate outcomes of alternative mechanism(s) for training genetic counselors
    - Standardized patients/Simulation Center
  - Increase the number of genetic counselors trained annually

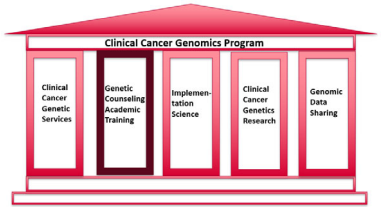
26



Genetic Counseling Academic Training

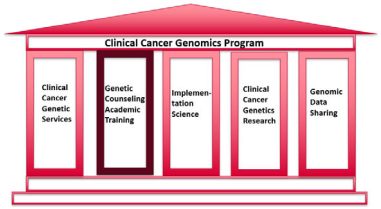
- Staffed by an Associate Program Director

  Leila Jamal, ScM, PhD, CGC
  - Joint appointment with the Department of Bioethics
  - Her research focuses on:
    - How patients and clinicians react to and use secondary finding information about germline cancer predisposition in their children
    - How patients react to and use inconclusive results from exome sequencing



Genetic Counseling Academic Training

- Collaboration with NHGRI/MOU fully executed in May 2019
  - Program rebranded to the NIH Genetic Counseling Training Program
  - Increase student cohort from 4 to 6 per academic year starting August 2019
  - Accreditation substantive change request submitted and approved
  - Develop, evaluate, and disseminate novel training methods
    - Simulated patient rotation for first year launched 2020
      - Protocol to evaluate the effectiveness, feasibility, and acceptability of this rotation



Genetic Counseling Academic Training

- Enhance the cancer genetics didactic and clinical training curricula for ALL students
  - Inferring the presence of germline variants from somatic test results
  - Hematologic malignancies
  - Pediatric cancer counseling
  - Environmental influences affecting germline cancer predisposition
  - Use and limitations of polygenic risk scores
  - Alternative/automated service delivery models for cancer risk assessment and counseling

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29

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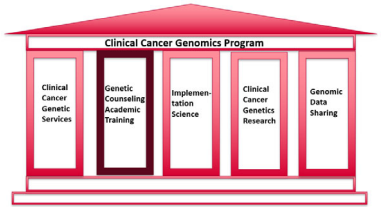

Genetic Counseling Academic Training

- Where do we go from here? Reimagining this program long term….

JHU/NIH Genetic Counseling Training Program

August 21, 2024 Update: The NIH-funded Genetic Counseling Training Program is temporarily pausing admissions to undergo strategic planning and development. NHGRI wants to hear from you on the current needs and challenges associated with genetic counseling while we plan to launch a new effort to develop leaders in the field. To get involved, or for questions, please contact Program Director Lori Erby at **lori.erby@nih.gov**.

Drawing on resources from three outstanding research institutions, the National Human Genome Research Institute (NHGRI) and National Cancer Institute (NCI) at the National Institutes of Health (NIH) and the Department of Health, Behavior and Society at the Johns Hopkins University (JHU) Bloomberg School of Public Health have collaborated to develop and support the JHU/NIH Genetic Counseling Training Program (GCTP), a competitive graduate program that addresses the growing need for genetic counseling services.

NIH NATIONAL CANCER INSTITUTE Center for Cancer Research https://www.genome.gov/careers-training/Professional-Development-Programs/Genetic-Counseling-Training

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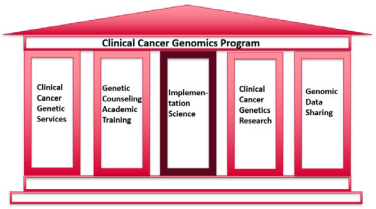

Genomic Implementation Science

Project Specific Objectives:

Project 1) Nursing Capacity in Pharmacogenomics:

- Determine the state of pharmacogenomic competency in nurses with prescriptive privileges.
- Determine the state of nursing curricular content in pharmacogenomics.
- Determine the state of pharmacogenomic competency in nursing faculty.
- Establish a pharmacogenomic practicing nurse education initiative addressing identified knowledge skills and ability deficits.
- Establish a pharmacogenomic nursing faculty education initiative addressing identified knowledge, skills, abilities, deficits and provide pharmacogenomic education curricular exemplars (i.e. Test to Learn) and model pharmacogenomic curricular content with associated resources.

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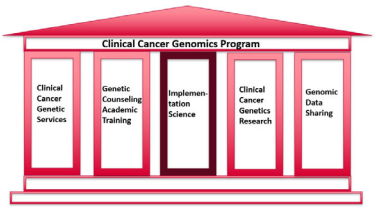
31

31



Genomic Implementation Science

Project Specific Objectives:

Project 2) Global Genomics Nursing Alliance:

- Accelerate the translation of genomic information and/or technology into nursing practice :
  - development of a roadmap for progress that recognizes real-world constraints;
  - sharing of existing resources to enable less developed countries to integrate genomic healthcare practices more rapidly into nursing; and
  - establishing an international partnership for ongoing research
- Create global minimal genomic competencies for nursing
- Generate a global genomic nursing science blueprint
- Establish global genomic communities of practice focused on: Workforce Development; Clinical Practice; and Overcoming Barriers.

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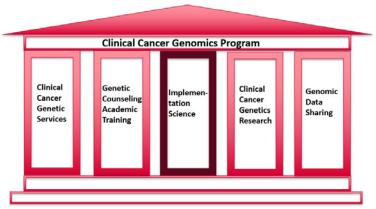
32

32



Genomic Implementation Science

Project Specific Objectives:

Project 3) Genomic Nursing Competency:

- Review and revise The Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcomes Indicators, 2nd Edition (completed PMID: 38797885)
- Conduct a national nursing workforce study to assess current precision health and genomic nursing knowledge, skills and abilities. (completed, publication in process)
- Review and revise the Essentials of Genetic and Genomic Nursing for Nurses with Graduate Degrees
  - Revision drafted by steering group, Delphi panel being invited to participate
  - Delphi online project being drafted for review; hoping to launch on or before 10/14/2024
- Conduct a hospital conglomerate wide precision health and genomics nursing competency implementation study compared to usual hospital planned education.

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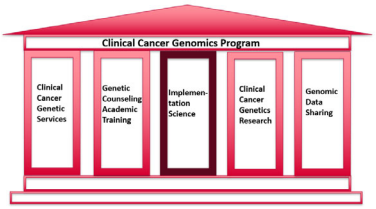
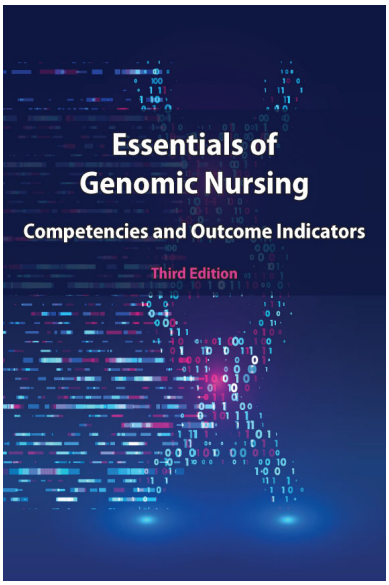


Genomic Implementation Science

Project Specific Objectives:

Project 3) Genomic Nursing Competency, continued:

- Integrate the Genomic Competencies into general AND specialty nursing scope and standards of practice.



https://www.nursingworld.org/nurses-books/ana-books/ebook-essentials-of-genomic-nursing-competencies-/

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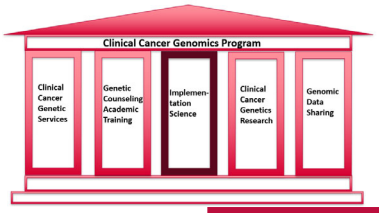

Genomic Implementation Science

National Nursing Workforce Study

2010	2024
N=619	N=1065
Education Level	
Diploma 3% Associates 17% Baccalaureate 41% Masters 31% Doctorate 8%	Diploma 2% Associates 8% Baccalaureate 25% **Masters 41%** **Doctorate 24%**
Actively seeing patients	
54%	51%

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Calzone, K., et al., (2013). National nursing workforce survey of nursing attitudes, knowledge and practice in genomics. Personalized Medicine, PMID: 24363765. Calzone, K., et al. (2024). Evaluation of the Integration of Genetics/Genomics into Nursing Practice. ISONG Abstract, accepted.

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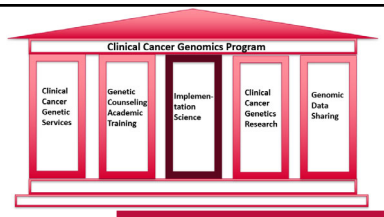

Genomic Implementation Science

2010	2024
Reported their genomic knowledgebase was poor/fair	
57%	55%* *24% reported a patient initiated a genetic discussion with them in the past 3 months
Would attend a course on their own time	
73%	79%
Considered it very important to learn about genomics	
67%	66%

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Calzone, K., et al., (2013). National nursing workforce survey of nursing attitudes, knowledge and practice in genomics. Personalized Medicine, PMID: 24363765. Calzone, K., et al. (2014). Evaluation of the Integration of Genetics/Genomics into Nursing Practice. ISONG Abstract, accepted.

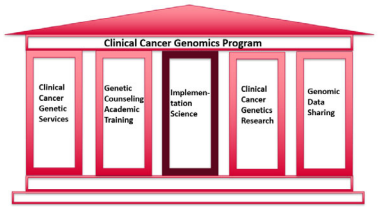
36

Poll Question

Please rate your understanding of the genomics of common diseases.

1. Excellent
2. Good
3. Poor

37

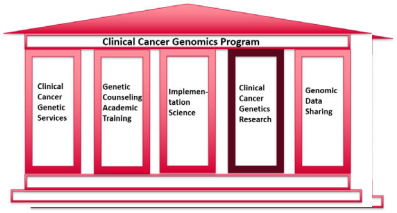


Genomic Implementation Science

Addressing the Deficit

- American Nurses Association has agreed to convene a panel of nursing leadership from academic, clinical, regulatory, and research entities to discuss the state of genomic capacity in nursing and how to overcome ongoing education and clinical deficits.
- Agreed to integrate into Scope and Standards of Practice

38

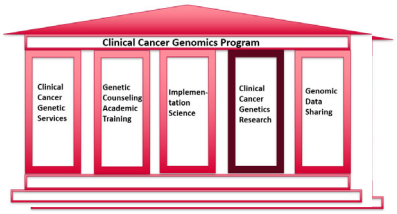


Clinical Cancer Genetics Research

Objectives:

- Establish education and practice resources for oncology nurses in genomics
- Conduct cancer specific precision health and genomics nursing competency implementation studies in both the academic and clinical settings.

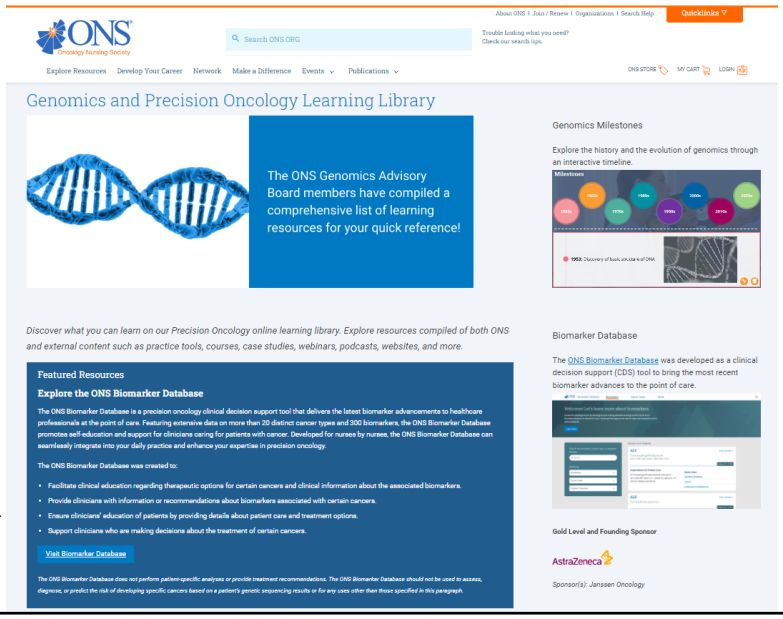
39



Clinical Cancer Genetics Research

- **Oncology Nursing Society**
  - Genomics Advisory Board
    - Establish and maintain the Genomics and Precision Oncology Learning Library
    - Conduct periodic programs at the national and chapter level in genomics and precision oncology
    - Evaluate outcomes

https://www.ons.org/learning-libraries/precision-oncology



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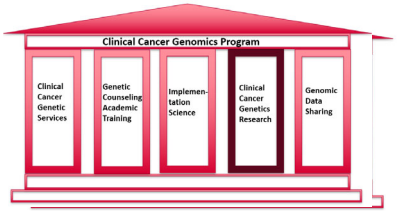


Clinical Cancer Genetics Research

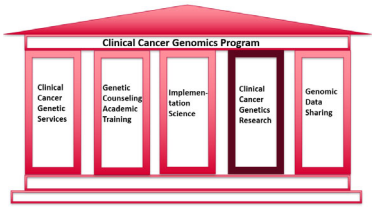
Extramural

- Genetic counseling summer students
- Global Genomic Nursing Alliance

Future Plans-Grant submitted

- Intramural/Extramural education program in cancer genomics, funding proposal All Ireland NCI Cancer Consortium Research & Innovation Grant Scheme under the Genomics and Precision category.
  - **GEN**omics **E**nabled **ONC**ology Education programme (GENE-ONC) takes a tripartite approach of educating faculty, clinical assessors, and students within one overall program.
  - Funding decision pending

41



Clinical Cancer Genetics Research

- Pharmacogenomic Faculty APRN Model Curriculum and Education
- Genomic Implementation
  - National (GGNCI) and International (Global Genomic Nursing Alliance)
    - Competencies, Roadmap, Maturity Matrix
    - GGNPS refinement/translation into other languages-open access for modification for local context
  - National Nursing Workforce Competency Initiative
- Summer Genetics Institute*, OMICS Nursing Science and Education Network* and Genomic Nursing Science Blueprint* (*discontinued by NINR new leadership, moving to G2NA, NHGRI, and possibly NCI)

Tonkin, E., et al. (2020). A Roadmap for Global Acceleration of Genomics Integration Across Nursing. JNS, PMID: 32301236
Tonkin, E. et al. (2020). A Maturity Matrix for Nurse Leaders to Facilitate and Benchmark Progress in Genomic Healthcare Policy, Infrastructure, Education, and Delivery. JNS, PMID: 32592453

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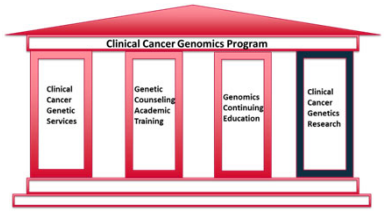
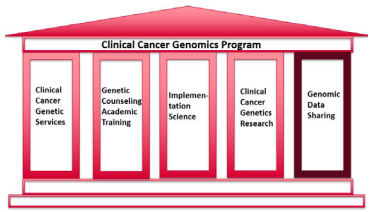


Clinical Cancer Genetics Research

Future Plans

- Cascade Testing
- Exome sequencing initiative with Laboratory of Pathology
  - Models of consent/genetic counseling
  - Increasing role of genetic counselors in germline variant curation
    - Addressing scope of practice and establishing training mechanisms
- Genetic counseling workload assessment
  - Develop robust mechanisms to measure patient complexity

43



Genomic Data Sharing

Objectives:

- Establish a platform for the expansion of clinical genetic and genomic research
- Establish mechanisms for translation of scientific discoveries into clinical practice

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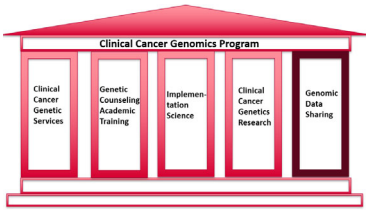


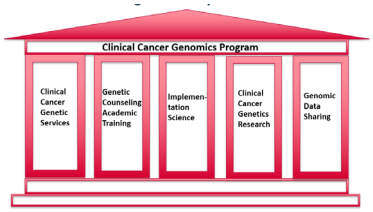
Genomic Data Sharing

- Genomic Data Sharing
  - Staffed by Abid Al Reza, PhD
- Work with CBIIT on the development and maintenance of the GDS online portal (projects, data sharing plans, institutional certifications)
- Established an online mechanism for GSR Sensitivity Determinations
- Interface with all CCR intramural investigators (clinical and laboratory including human, animal etc studies)
- Work closely with the CCR Protocol Support Office
- There are 360 CCR studies registered in dbGaP

Poll Question

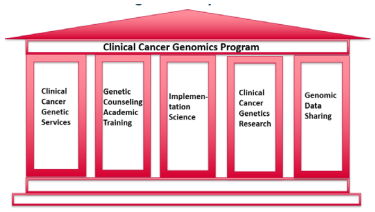
How important do you think it is for the nurse to become more educated about the genomics of common diseases?

1. Very Important
2. Somewhat Important
3. Not Very Important
4. Not At All Important



It Takes a Village

Clinical Cancer Genetics Program
Grace-Ann Fasaye, ScM, CGC; Leila Jamal, ScM, PhD, CGC; Alexandra Lebensohn, MS, CGC; Yi Liu, MS, CGC; Hermelat Mesfin, BS; Abid Al-Reza, PhD; Michaela Taylor, MS, CGC
Center for Cancer Research
James Gulley, MD, PhD, Fatima Karzai, MD, Mel Bronez, MPA
*Bill Dahut, MD
Genetics Branch
Paul Meltzer, MD, PhD, Javed Khan, MD
Melissa Shue, Kandie Webb, Claire Simmons



Questions

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