
Integrating Genomics into Primary Care
The NSGC Collaborative Services Summit

Framing the Integration Questions

- What does successful integration of genomics into primary care look like?

- How do you get there?

Successful Integration

- Recognition of the value of genomics in healthcare
- Effective identification of patients with high versus low genetic risks
- Appropriate, tailored interventions in high risk individuals that improve outcomes
- Appropriate use of genetic testing, including informed consent
- Excellent communication between healthcare providers/continuity of care

Steps Towards Integration

- Educating primary care
- Providing effective point of care tools to facilitate identification
- Enhancing communication between primary care and genetics providers
- Increasing access to genetics specialty services
  - Increasing the workforce
  - Increasing the efficiency of genetics services (without jeopardizing patient care)
  - Alternative service delivery models

NSGC Collaborative Genetics Services Summit

- Purpose: To bring together primary care providers, genetic specialists, and other stakeholders to develop competency-based collaborative models for integrating genomics into healthcare
- Impetus: Must understand the perspectives of primary care and genetics specialists to develop practical models
- Primary outcome: Consensus models for triaging genomics services that can be applied practically
- Secondary outcome: May identify gaps in genomics competencies/training

Summit Process

- Group of investigators representing genetic counseling, medical geneticists, nursing, physician assistants (PA's), family practice physicians, and genetics education convened to establish goals and project implementation
  - Angela Trepanier, MS, CGC PI
  - Jean Jenkins, PhD, RN -Michael Rackover, PA-C, MS
  - Frederick Chen, MD, MPH -Joan Bodurtha, MD, FACMG
  - Joseph McInerney, MS, CGC, NCHPEG
- Guided by an advisory committee (Chair, Cathy Wicklund, MS,CGC)and NSGC leadership

Planning Process

- Selected five cases for which to develop triage models
  - Reproductive risk assessment through population-based carrier screening
  - Cancer genetic risk assessment : Lynch universal screening
  - Risk assessment for medication selection and dosing (pharmacogenomics)
  - Risk assessment for complex disease: cardiovascular disease
  - Pediatric diagnostic evaluation for developmental delay
- Drafted major components of each using literature and professional experience

Developing Triage Models

- Multidisciplinary work groups convened to further develop triage models
- Charges:
  - Describe and reach consensus about all components of genomics services involved in case and the tasks and skill associated with each component
  - Map existing genomics competencies of each profession to the skills needed for each task
  - Reach consensus about how to triage genomics services based on outcomes of step 1 and 2
- Worked over course of 8 weeks by email and calls

Summit Meeting

- Held in November 2011 in Washington DC
- Facilitated meeting with multidisciplinary group of attendees (30)
- Goals:
  - Critically evaluate the components of each draft case
  - Assess validity and practicality of triage models
  - Provide additional input: who else should review? What is missing?
  - Discuss potential for implementation and identify barriers
  - Brainstorm next steps

Process

Planning

- Investigators, advisory committee and NSGC leadership develop goals, cases and plan of work

Pre-Summit Work

- Draft cases developed, work groups assigned to define tasks involved, skills needed and to map competencies of each profession

Summit

- Cases vetted by new group of participants, outcomes discussed, next steps identified

Cancer Risk Assessment and Testing (Lynch Syndrome)

Reasons for Case Selection

This case investigates Lynch syndrome from two pathways: tumor tissue testing (population-based screening) and assessment of risk through family history

- Life cycle: Growing component of adult clinical genetics services
- Key service providers: Opportunity to examine 1) which health care professionals need to be involved from a practical standpoint when all tumors are screened and 2) how this may be different from family-history based risk assessment and testing
- Informed consent: Role will vary depending on whether risk assessment done through tumor tissue screening or targeted genetic testing based on family history

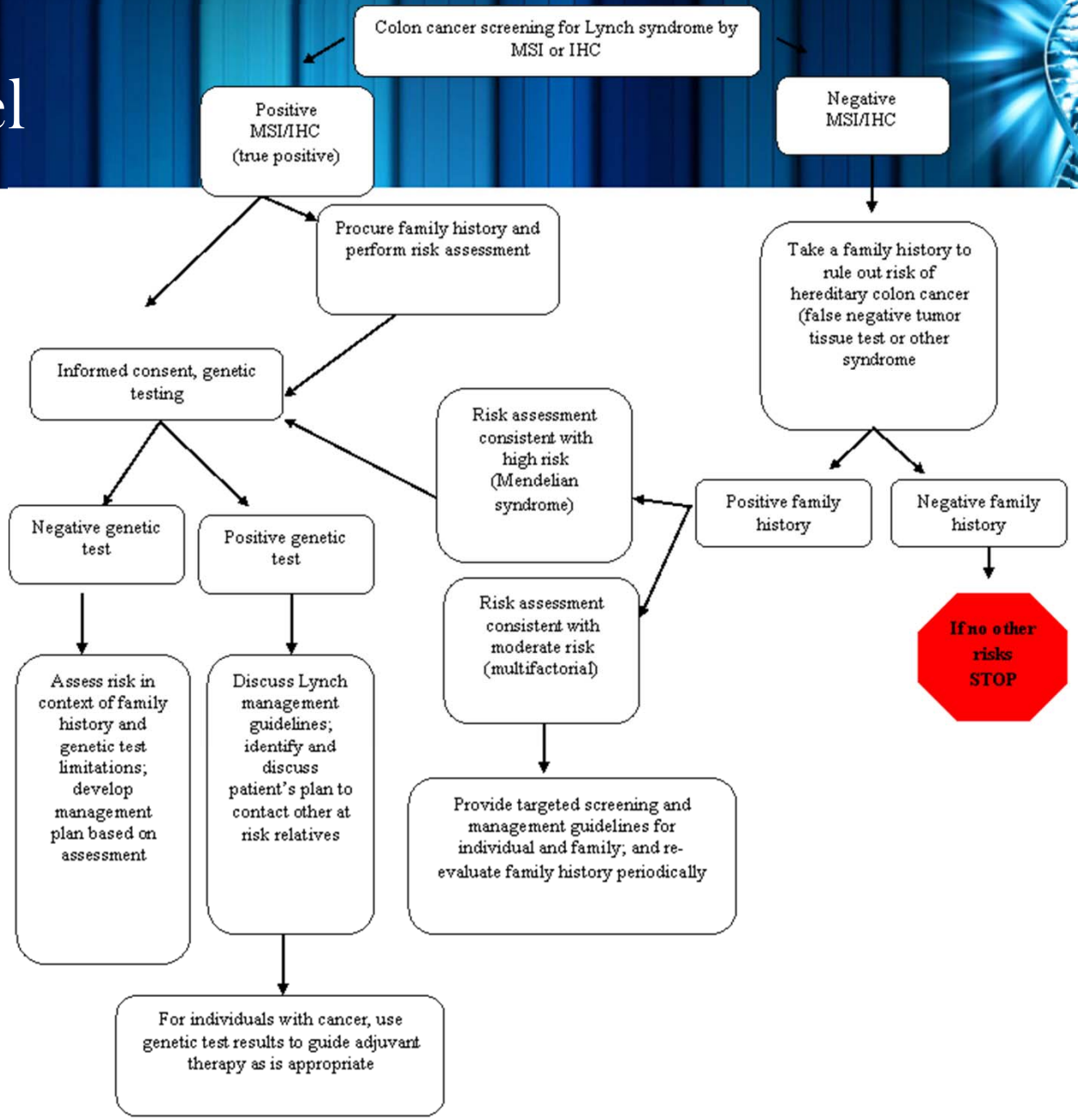
Cancer Risk Assessment and Testing (Lynch Syndrome)

Reasons for Case Selection, Cont'd

- Treatment/management: Lynch syndrome is an example of a condition where identification of at-risk individuals guides clinical management and reduces morbidity and mortality
- Other issues:
  - Allows for the examination of psychosocial variables associated with providing counseling to those recently diagnosed with a condition (cancer) versus those with a past history or no diagnoses
  - EGAPP guideline supporting Lynch screening for the benefit of identifying at risk family members makes this a strong candidate condition for describing the elements of cancer genetic risk assessment and counseling

Pre-Summit Triage Model

BT is a 45- year old man diagnosed with colon cancer. His tumor tissue is tested by IHC for mismatch repair proteins as part of an established screening protocol.



Sample Task Analysis and Competency Mapping

Case Component	Tasks	Skills Needed	Registered Nurse	Advanced Practice (NP, CNS,CNM, CRNA)	Credentialed Genetics Nurses (GCN & APNG)	Genetic Counselors (including RN)	Primary Care Physicians	Medical Geneticists	PA	Other
1. Screening colon cancers for Lynch syndrome with MSI and/or IHC	1. Identify cases/generate order	1. Access to information about patients scheduled for colon cancer surgery 2. Knowledge of criteria for initiating testing 3. Ablitity to communicate with surgeons and pathologists	NO	YES	YES	YES	YES (relatives)	YES, at least 1 &3		Pathologist is required for this step
	2. Evaluate the quality and expertise of services for providing MSI and/or IHC testing	1. Ability to communicate with pathologists, particularly about expertise in interpreation of IHC staining 2. Knowledge of specificy and sensitivity of MSI/IHC 3. Determine if expertise are available to do this testing internally or if testing should be referred to a reference laboratory	NO	NO	NO	YES		YES		Pathologist is required for this step
	3. Perform test or send out tissue to a reference lab	1. If performing the testing in house, ability to do microdisecet tissue, interpret IHC staining, and access to molecular testing for performing MSI 2. If sending out samples, ability to prepare tissue and coordinate with reference laboratory	NO	NO	NO	NO	NO	NO		Pathologist is required for this step

Work Group's Summary Thoughts*

- The steps of receiving and interpreting results of tumor tissue testing and coordinating additional genetic testing would most typically be carried out by genetic counselors, nurses with specialization in oncology genetics, geneticists, oncologists, and surgeons.
- However, primary care physicians may need to be involved with all steps, especially in rural areas where access to genetics services may be unavailable

Members: Wendy Kohlmann, MS, CGC; Sarah Lewis, MHS, PA-C; Anne Ersig, PhD, PNP-BC; Katherine Hurst, MD; Wade Samowitz, MD; Helga Toriello, PhD, FACMG

Summary, continued

- Primary care physicians also have a key role in helping coordinate long term cancer screening and follow up for patients with Lynch syndrome
- However, continued support from specialists with expertise in Lynch syndrome would be important to help primary care providers identify the most up-to-date guidelines and to address family issues that develop over time

Summit Meeting Structure

- Overview and expectations, networking dinner
- Welcome, introductions, ground rules
- Triage model discussions
  - Presentation of model (10 minutes)
  - Small group discussions (5 group of 5-6 people; 30 minutes)
  - Full group discussion (20 minutes)
- Questions to consider
  - Are all essential components of genetics services captured?
  - How do competencies in your profession align with tasks?
  - How important is it for your profession to be involved?
  - Barriers to involvement or implementation?

Summary of Feedback- Lynch Case

- Model is worth pursuing but needs some work
- Struggle with where family history fits in (wheel versus linear model?)
- Reassess family history after MSI/IHC
- Need to see more detail about family history pathway
- Cascade screening- where does that fit in?
- Consider unequivocal MSI/IHC results
- Informed consent? Where?
- Get rid of stop sign
- How do you communicate with the unaffected?

Barriers to Implementing Lynch Model

- Cost, economic implications
- Provider resistance
- Responsibility of contacting relatives
- Dealing with all of this when patient is sick
- Positive cases sometimes never come in
- Ethics of duty to warn and barriers to warning
- Need to align competencies with algorithm. Lack of curricula to support needed competencies
- Workforce issues

Next Steps- Individual Models

- Will work by conference call with investigators and subset of Summit attendees to clean up models with goals of publishing task analysis +/- competency mapping
- Prioritizing Lynch model and possibly pharmacogenomic one but will potentially work on all of them

Summary Thoughts on Summit

- Need to educate primary care about vagaries of informed consent across genetic indications
- Family history should be at the top of any model, but not only way of identifying risk
- Reinterpretation, retest, reassess, recontact- 4 “R’s” must be part of every model
- Need to decrease siloing and encourage collaboration
- Summit validates broader movement in continuing education: team-based versus single provider-based
- Include EHR vendors in the conversation

More Summary Thoughts

- Modules that could be developed out of this work
- Another outcome: models could drive reimbursement
- Good activity as it gets people used to modeling/triage
- Need to consider patient experience and context
- Develop joint practice guidelines

What's Next

- Develop white paper on process and suggested outcomes
- Convene groups of attendees, investigators, and work group members to incorporate Summit feedback into triage models
- Publish task analyses for each indication +/- competency mapping
- Consider second summit to address identified barriers

Closing Thoughts

- Need to work together to come up with practical ways to integrate genomics into healthcare
- Models can help prioritize educational targets for primary care
- Still significant barriers to integration, though, and these need to be addressed as well

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