Genetics in Primary Care Institute: Co-Management Between Primary Care and Genetic Service Providers

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Acknowledgments

• The Genetics in Primary Care Institute (GPCI) is a three-year cooperative agreement between the American Academy of Pediatrics (AAP) and the Health Resources & Services Administration (HRSA)/Maternal & Child Health Bureau (MCHB), Genetic Services Branch

• June 1, 2011—May 31, 2014
Impetus for the GPCI

• Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) - recommendations in 2009 workshop provided blueprint for GPCI
• Documented gaps exist in PCPs’ knowledge, attitudes, and practice
• Advances in genetics and genomics continue, prompting more demand, while workforce shortages among genetic professionals grow
• Strategies and tools are needed to improve PCPs’ role in providing genetic services
Vision:
To improve primary care providers’ knowledge and skills in the provision of genetic medicine
Goals of the GPCI

- **Quality Improvement**
  - Develop and test strategies to integrate genetics into the medical home

- **Technical Assistance Center**
  - Provide education and technical assistance to a wide audience of PCPs

- **Resident Education**
  - Embed genetic education into the future pediatric PCP workforce
Roles of the PCP

- **Evaluate** through screening and surveillance
- **Educate** patients and their families
- **Explain** the results
- Make **appropriate referrals**
- **Coordinate care** with a subspecialist
- **Counsel** patients and families
- Provide **long-term follow-up** and care
Integrating Genetics into Primary Care Practice
QUALITY IMPROVEMENT PROJECT
Project Oversight

- GPCI Project Advisory Committee
- Multidisciplinary Expert Group
  - Amy Driscoll, MD, FAAP
  - Beth Tarini, MD, MS, FAAP
  - Wendy Chung, MD, PhD, FACMG
  - Abdallah F. Elias, MD, FAAFP, FACMG
  - Kerry Jedele, MD, FACMG, FAAP
  - Ruth Gubernick, MPH
  - Ingrid Larson, MSN, RN, CPNP
  - Beth A. Pletcher, MD, FAAP, FACMG
  - Michael Rinke, MD, FAAP
  - Lisa M. Vasquez, MPA
Needs Assessment

2012 Survey of AAP QuIN members (n=88)

• Less than half of pediatricians reported feeling competent providing services related to genetics and genomics
• 60% did not have adequate resources or information to determine which genetic tests to order
• Lack of knowledge on existing resources (eg, NBS ACT sheets)
• 100% of pediatricians agree/strongly agree that taking a FH is important for assessment of predisposition to disease
• EHRs lack functionality for recording FH information
• Less than 1/3 gather comprehensive FH from their patients

Objective and Goals

Test tools and strategies related to the integration of genetics into pediatric primary care

Project Goals
1. Eliciting *family history* as part of the health supervision visit
2. Improve the delivery of care for *pediatric patients with genetic conditions*
3. Develop policies and *improve office systems* to meet the first two goals of the project
Project Participation

- 13 teams in 11 states serving 130,000 pediatric patients annually
- Practice teams (3 people total) include a lead physician, 1-2 other clinicians, nurse or assistant staff person
- Participated in 6 months of data collection regarding project goals

Practice demographics:
- All use an EHR
- One family physician
- Diverse range of practice type, geographic setting, and patient population
# QIP Timeline and Overview

<table>
<thead>
<tr>
<th>Application Process:</th>
<th>Prework:</th>
<th>Action Period:</th>
<th>Evaluation:</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Form core improvement team and determine roles of each member</td>
<td>- Participate in prework call on December 18 (8 am CT) or 20 (11 am CT)</td>
<td>- Participate on conference calls</td>
<td>- Conduct 6 month post-project evaluation on sustainability and continued improvement</td>
</tr>
<tr>
<td>- Attend informational call on October 1 or 11</td>
<td>- Sign Consent Form</td>
<td>- Collect monthly data (chart review, registry review, and narrative report)</td>
<td>- Qualitative phone interviews</td>
</tr>
<tr>
<td>- Complete project application</td>
<td>- Complete baseline data</td>
<td>- Test changes using PDSA cycles</td>
<td>- Submit final chart review and registry review data</td>
</tr>
<tr>
<td>- Assess current practice and systems</td>
<td>- Create a storyboard</td>
<td>- Provide feedback on tools</td>
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<tr>
<td>- Make travel arrangements for Learning Session 1</td>
<td>-</td>
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</table>

**Education and Mentorship Provided:**

- Change Package with resources, tools, and information
- Bi-monthly clinical team coaching calls
- Access to a multidisciplinary Expert Group and genetic professionals
- Two in-person educational sessions
- CME conference, webinars, handbook, and other ad hoc resources
Family History

Steps Towards Collecting FH
1. Obtain buy-in
2. Test FH tools, strategies to integrate into workflow
3. Discuss FH with families
4. Identify red flags
5. Follow-up with identified risk
6. Evaluate your FH tool
7. Maintain and update FH annually
Results – Family History

Creation/Update of Multi-generational Family History (ALL project components included)

Percent

Cycle

Cycle 2
Cycle 4
Cycle 6
Cycle 8
Cycle 10
Cycle 12

Goal
All Subscribers

90.0
90.0
90.0
90.0
90.0
90.0
90.0
90.0
90.0
90.0
100.0
69.0
73.4

7.6
11.8
26.7
45.0
49.7
53.2
33.3
Best Practices – Family History

• Obtain buy-in from all providers and clinic staff
  • Reiterate value to all, even front desk staff handing out forms
• Test various tools and approaches to collecting FH
• Begin collecting FH at new patient or 2-month visit, goal to have comprehensive FH by 2 yrs.
  • Request information from family members; others accompanying child to sick and well-check visits
• Contact genetic professionals with concerns or suspicions
• Ask family members if they have any FH concerns
• Notice red flags, even negative (everyone is “healthy”)
• Work with EHR vendor or IT dept. to model FH screen in EHR from paper form to facilitate data collection and entry
Identifying Patients with Suspected Genetic Conditions

Steps Towards Improving Patient Care
1. Recognize Clinical Red Flags
2. Identify and Follow-Up with Patients Identified at Risk
3. Talk to Parents about an At-Risk Family Health History or Genetic Referral
4. Develop Relationships with Genetic Professionals
5. Review Indications for Possible Referral for a Genetic Evaluation
6. Order Appropriate Genetic Tests
Providing Appropriate Care for Patients with Genetic Conditions

Steps Towards Improving Patient Care
1. Implement Systems to Improve Genetic Services
2. Create a Patient Registry
3. Follow the Health Supervision Guidelines
4. Provide Family-Centered Care
Steps Towards Implementation

1. Identify patients with diagnosed genetic conditions; create a patient registry
2. Create aim statements / goals to improve procedures
3. Review patient registry data, re-evaluate aims
Size of Practice Registries for Children with Genetic Conditions

In January 2013:
- < 50 patients = 4 practices
- 51-150 patients = 6 practices
- > 1000 patients = 3 practices

In September 2013:
- < 50 patients = 3 practices
- 51-150 patients = 5 practices
- 151-999 patients = 4 practices
- > 1500 patients = 1 practice
Coordinating Care for Patients with Genetic Conditions

Steps Towards Improving Patient Care
1. Improve Processes to Co-Manage Care with Specialists
2. Obtain Current Emergency Plans (if applicable)
3. Obtain Current Emergency Letters (if applicable)
4. Plan for Transitions to Adult Care
5. Discuss Palliative Care
Results – 6 month post-project

![Diagram showing documented next steps and plan of care in registry over cycles. The graph indicates a trend where the percentage increases up to cycle 12, reaching a value of 79.8%. The data points are marked with percentages as follows: Cycle 2: 25.3%, Cycle 4: 25.8%, Cycle 6: 40.5%, Cycle 8: 59.4%, Cycle 10: 0.0%, Cycle 12: 79.8%. The goal line is represented by a red dashed line at 90.0%, while the fluctuating line represents all subscribers.]
Knowledge Pre vs. Post Comparison

All knowledge areas assessed in the pre-survey saw at least some improvement in the post-survey, with the importance of FH, role in providing genetic services, and communicating genetic info measures seeing the highest gains.
## Policies and Procedures for Patients with Genetic Conditions

<table>
<thead>
<tr>
<th>Practice has a standard process (written or not written) for...</th>
<th>Pre</th>
<th>Post</th>
<th>Δ from pre</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tracking genetic referrals made</td>
<td>38%</td>
<td>85%</td>
<td>47%</td>
</tr>
<tr>
<td>Identifying patients with a defined genetic condition</td>
<td>23%</td>
<td>77%</td>
<td>54%</td>
</tr>
<tr>
<td>Tracking children with a defined genetic condition</td>
<td>23%</td>
<td>77%</td>
<td>54%</td>
</tr>
<tr>
<td>Identifying genetic patients who are behind schedule for preventative services</td>
<td>23%</td>
<td>69%</td>
<td>46%</td>
</tr>
<tr>
<td>Contacting genetic patients who are behind schedule for preventative services</td>
<td>38%</td>
<td>69%</td>
<td>31%</td>
</tr>
<tr>
<td>Transition to adult care discussion with genetics patients by age 12</td>
<td>15%</td>
<td>62%</td>
<td>47%</td>
</tr>
<tr>
<td>Co-managing care genetics patients</td>
<td>8%</td>
<td>31%</td>
<td>23%</td>
</tr>
<tr>
<td>Obtaining emergency plans from specialists</td>
<td>15%</td>
<td>31%</td>
<td>16%</td>
</tr>
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Most practices established processes or written protocols around tracking and identification of patients with a defined genetic condition.

Fewer practices established processes or written protocols around emergency plans and co-managing care.
# Barriers to Implementation

<table>
<thead>
<tr>
<th></th>
<th>Barrier</th>
<th>% selecting 3 or 4 (significant barrier)</th>
<th>Average Rating</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Overall Project</strong></td>
<td>Time it takes to develop new policies and procedures</td>
<td>69.2%</td>
<td>3.0</td>
</tr>
<tr>
<td></td>
<td>Time it takes to implement new policies and procedures</td>
<td>69.2%</td>
<td>2.9</td>
</tr>
<tr>
<td></td>
<td>Time it takes to add these components to the health supervision visit</td>
<td>46.2%</td>
<td>2.4</td>
</tr>
<tr>
<td><strong>Family History Collection</strong></td>
<td>Limitations of the family history tools/screens in EHR</td>
<td>69.2%</td>
<td>2.7</td>
</tr>
<tr>
<td></td>
<td>Time required to enter the family history information into the EHR</td>
<td>46.2%</td>
<td>2.5</td>
</tr>
<tr>
<td><strong>Providing Care for Patients with Genetic Conditions</strong></td>
<td>Difficulty in managing patient registry</td>
<td>69.2%</td>
<td>2.7</td>
</tr>
<tr>
<td></td>
<td>Difficulty in establishing a recall system for patients</td>
<td>46.2%</td>
<td>2.2</td>
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Genetic Mentorship Program

Goal: To increase partnerships between PCP and genetic professionals at the local / regional level

Format:
- **Who:** A geneticist or genetic counselor paired with each practice from their region
- **What:** Meet monthly via phone, email, or in-person
- **Purpose:** Share information and resources, discuss patient cases, facilitate relationships
Case Example: Mentorship Program

Who:
• PCP - Nassim and Associates (Indiana)
• Genetic Counselor - Cecilia Rajakaruna (Louisville, Kentucky)
Case Example: Goals

1. Educating allied health staff on genetic conditions and FH
2. Tracking patients with genetic d/o
3. Integrating FH into the EHR
4. Working on care plan / creating care plans for patients
5. Developing policies and processes for genetic referrals
6. Connecting with local geneticists and allied health professionals
Educating Allied Health Staff

• Discussing genetics in the practice

• Talking with your genetics specialist

• Create small changes to incorporate into clinic workflow
Tracking Patients/EHR

- Electronic Health Record (EHR)
- Registry
Care Plans/Transition

• Co-Management

• Transition – the specifics related to this population

• Syndrome specific – health management guidelines
Connecting with Specialists

• Exposure to genetics

• Meeting your local team

• Utilizing your local specialists as resources
Genetics Referrals

Pediatric primary care provider should consider referral to a medical geneticist:

- Abnormal newborn screening results
- Developmental disorder or intellectual disability
- Blindness or deafness
- Presence of a known or suspected genetic condition or chromosomal abnormality
- Disorders of growth
- Structural or morphological variations
- Family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
Case Example: Results

• **Knowledge**
  • Appropriate utilization of services
  • Knowledge of genetic red flags

• **Attitude**
  • 100% of participants value a FH and will continue to improve their processes towards collecting, storing, and updating information

• **Behavior**
  • All practices have integrated a genetics family history screening into their practice
  • Practices’ patient registries had an average of 300 children with heritable disorders who are now receiving improved care
Case Example: Results

• **Systems**
  • “I had never heard of a genetic counselor prior to this project”
  • “Thanks to my mentor, I now have a relationship with my local genetics center and have resources to help me when considering a referral and improved relationship with specialists to co-manage care”

• **Patient Outcomes**
  • “If it hadn’t been for this project, I never would have considered a referral to genetics without a definitive diagnosis”
  • “Positive FH screen results in identification of genetic conditions or referral of patient or family member to genetics”
GeneticsInPrimaryCare.org

The clearinghouse for education and tools regarding genetics in primary care.

Informational destinations include:
- What is genetics?
- Resources for Your Practice
- Provider Education
- Expert Video Testimonials
- And More!
Additional AAP Resources

• Newborn Screening EQIPP Course
• “Dive into the Gene Pool” PediaLink CME Course
• Medical Genetics and Pediatric Practice: A Handbook
• PediaGene: AAP Genetics Screening Guide – Mobile Device Application
Contact Us

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