

Step by Step Instructions

A Toolkit to Improve Care for Pediatric Patients with Genetic Conditions in Primary Care



GENETICS 101

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Acknowledgements

A Toolkit to Improve Care for Pediatric Patients with Genetic Conditions in Primary Care

American Academy of Pediatrics
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GENETICS IN PRIMARY CARE INSTITUTE
geneticsinprimarycare.org

A cooperative agreement between the American Academy of Pediatrics and the Health Resources and Services Administration, Maternal and Child Health Bureau



THANKS

This toolkit is based on the learnings from the Genetics in Primary Care Institute (GPCI) Quality Improvement Project, a project supported by the Maternal and Child Health Bureau and managed by the Practice Improvement Network, a network of the Quality Improvement Innovation Networks at the American Academy of Pediatrics (AAP).

The GPCI is a three-year cooperative agreement between the AAP and the Health Resources and Services Administration Maternal and Child Health Bureau (federal award number UC7MC21713). The GPCI has been established with an overarching vision to improve primary care provider knowledge and skills in providing genetics-based services. A major goal of this initiative is to increase understanding amongst the public and medical home providers regarding how genetics and family history influence the health of the individual.

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Practices

- All About Children Pediatric Partners PC, West Reading, PA
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- Iron Horse Pediatrics, Colorado Springs, CO
- Nassim and Associates, New Albany, IN
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Reviewers

Thank you to the individuals who reviewed the toolkit for their suggestions and insight.

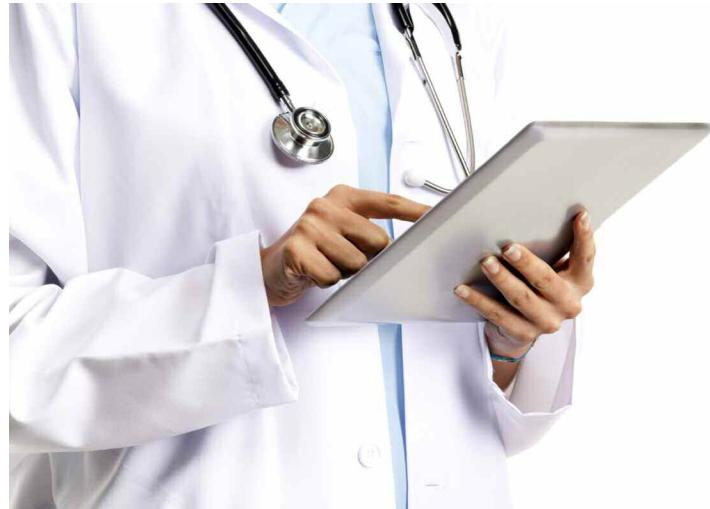
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Dedication

This Toolkit is dedicated in grateful appreciation for the life of Thomas J. Sullivan, MD, FAAP. Dr Sullivan was a former president of the Virginia AAP chapter, a devoted and engaged member of QuINN, and a leader in the Genetics in Primary Care Institute Quality Improvement Project. In his words, "Quality improvement engages the entire team; doctors, nurse practitioners, office managers, nurses, administrator and family advisory panel." The impact of Dr Sullivan's compassion and commitment to the best care for all children will continue on through the many pediatricians and clinicians whose lives he touched and the policies and resources he contributed to.

Section 1

Getting Started



One step at a time

Getting Started

Background

The Genetics in Primary Care Institute (GPCI) seeks to take the advances in genetics over the last fifty years, especially those in the last 10 years since the completion of the Human Genome Project, and make them relevant to the practice of primary care pediatrics. Infectious disease revolutionized how medical care was delivered to children in the 20th century with the advent of immunizations, antibiotics, advanced diagnostic tools and a thorough integration of infectious disease into primary care. We think genetics has a similar potential in the practice of pediatrics in the 21st century.

We now realize that the medical specialty of “genetics” really represents the coalescence of genetics and genomics and epigenetics. The challenge and task of the GPCI has been to enhance the translation of this information into material that is relevant for the practice of primary care medicine. The potential for the prevention, diagnosis, and treatment of many genetic and genetic-related disorders is now at hand. As a result, the ability to address the triad of prevention, diagnosis, and treatment has been our goal and are key factors in making genetics relevant to primary care practice. Specifically, the GPCI has focused on helping guide the primary care provider (PCP) in maximizing evaluation and diagnostic processes related to genetic risk, becoming familiar with therapeutic modalities, and assisting with the delivery of routine care for children with suspected and diagnosed genetic conditions.

About this Toolkit

This toolkit was developed by the GPCI to help PCPs integrate genetics into their practice. The information in this package was developed in conjunction with a team of providers and refined during a 10 month quality improvement project through the American Academy of Pediatrics.

The toolkit uses quality improvement practices as the foundation for integrating genetics into the primary care practice.

The toolkit is broken up into 5 sections: Getting Started, Collecting Family Health History, Identifying Patients with Suspected Genetic Conditions, Providing Appropriate Care for Patients with Genetic Conditions, and Coordinating Care for Patients with Genetic Conditions.

Each section contains steps for your practice to take in identifying and caring for patients with genetic conditions. The focus areas contain action steps, tools, communication tips, and additional resources to help you implement this toolkit. There are also examples from the practices who participated in the quality improvement project. We hope their experiences will show that you too can apply “genetic thinking” and test and implement tools and resources to integrate genetics into your practice.

“All of the clinicians are extremely dedicated to making sure that our patients receive the best care possible, and we desperately want to make sure that our patients will have access to all of the benefits of personalized, genomic medicine.”

Even though there is a lot of information here, don’t be overwhelmed. You can take things step-by-step. Each step helps improve care for your patients. Using this toolkit will also help your clinic improve systems of care that can be applied to other patient populations in your practice.

Getting Started

Genetics 101

Why improve your understanding and treatment of patients with genetic conditions? Genetics impacts all of us. It influences our hair color, our height, and the health conditions we'll face during our lifetime. Some of us may be predisposed to allergies, or heart problems, or cancer. By understanding the health conditions your patients are more likely to have, you can help prevent or treat these conditions more effectively.

Genetic and genomic technology has improved significantly over the last decade. More than 5 new genetic tests emerge each month, the cost of new technologies is becoming more affordable, and gene therapies are being developed where there were previously none.

Did You Know?

Genetics refers to the study of genes and their role in inheritance – the way certain traits are passed down from one generation to another.

Genomics is the study of all of a person's genetic material, the interactions of that genetic material, the interactions of that genetic material with the environment, and the subsequent phenotypic changes.

Genetics uses information from one or two genes to explain a disease or condition, whereas genomics examines all of the genetic information to determine biological markers predisposing an individual to disease.

Chances are that most PCPs' familiarity with both "new" and "old" genetics is outdated, and we encourage you to explore some of the following resources to update your knowledge and skill base.

Tools & Resources

- [Genetics in Primary Care Institute](#) provides tools, resources, and education to increase PCP knowledge and skills in providing genetic-based services
- [Genetic Science Learning Center](#) is an online education program that translates science and health for non-experts
- [National Coordinating Center and Regional Genetic Services Collaboratives](#) provides tools and resources for providers about newborn screening and genetics
- [Medical Genetics in Pediatric Practice \(AAP Policy Manual\)](#)
- [GPCI Webinars](#) provide on demand webinars through the Time Out for Genetics and Integrating Genetics into Your Practice webinar series. The webinars provide PCPs and specialists with information for better understanding the relevance of genetic medicine to primary care by highlighting the important skills that primary care providers already have and build upon them to provide a comprehensive approach to primary care in which genetic thinking is integrated in every patient encounter.
- [ACMG Basics: Genetics for Providers](#) is an online CME activity, which designates up to 6 hours of AMA PRA Category 1 Credit toward completion of the AMA Physician's Recognition Award.

Getting Started

Ethical, Legal, and Social Issues

Testing for genetic conditions can be complex and while there have been many advances in genetic testing, limitations remain. A positive result does not necessarily confirm a diagnosis, and a negative result does not always rule out the condition. Due to limitations in technology, genetic testing may lead to unanticipated results.

Over the past decade, significant advances in genetic testing and technologies have altered the clinical management of individuals. Advances in genetic testing are also accompanied by a string of new challenges related to the ethical, legal, and social issues (ELSI) for our society.

Genomic advancements in testing and diagnosis often outpace the development of therapeutic treatments. Although largely unsupported by professional medical associations, private companies continue to promote genetic tests to the public (direct-to-consumer tests). As PCPs grasp the new implications of genetic and genomic advances, an understanding of ELSI will allow them to anticipate these new challenges and provide better care for their patients.

Research has found that a frequent barrier to practicing genetic medicine in a primary care setting is that providers are unsure how to provide adequate information and support to their patients, particularly in the areas of ELSI. Since PCPs are frequently first to interact with patients seeking genetic information, they are encouraged to review resources to better equip them for these sensitive interactions.

Genetic Information Nondiscrimination Act (GINA)

Genetic discrimination legislation—the Genetic Information Nondiscrimination Act (GINA)—passed in May 2008. As a result of GINA, group and individual health insurers cannot use a person's genetic information to:

- Set eligibility requirements
- Establish premium or contribution amounts
- Request a person undergo a genetic test

Employers cannot use a person's genetic information to:

- Make decisions about hiring, firing, job assignments, or promotion
- Request, require, or purchase genetic information about an employee or family member

Limitations of GINA:

- Does not provide protection when a condition is already diagnosed or manifest
- Does not apply to life, disability, or long-term insurers

The following resources can assist providers and families to understand the benefits and limitations of GINA legislation and be able to assist families in navigating these issues:

- [Discussion guide from the National Coalition for Health Professional Education in Genetics for clinicians about GINA legislation](#).
- [Fact Sheet from the Department of Health and Human Services for researchers and health care professionals regarding the overview and implications of GINA legislation](#).
- The [Genetics and Public Policy Center](#) helps policymakers, the press, and the public understand and respond to the challenges and opportunities of genetic medicine and its potential to transform global public health. The Genetics and Public Policy Center has an extensive “Facts, Questions, and Answers” section about the implications of GINA legislation for patients.

Getting Started

Articles about Genetic Testing

- [Ethical and Policy Issues in Genetic Testing and Screening of Children](#)
- [Ethical Issues With Genetic Testing in Pediatrics](#)
- [Testing Children for Adult-Onset Genetic Diseases](#)
- [Disclosure of Incidental Findings From Next-Generation Sequencing in Pediatric Genomic Research](#)

Genetic Testing of Children

Many ethical concerns exist related to genetic testing of children. Providers and families should be aware of the potential risks associated with genetic testing on children. Unanticipated incidental findings (non-paternity, adult-onset disorders) may result from genetic testing in children, and providers should be equipped to handle these findings.

With the exception of state-mandated newborn screening, all genetic tests are voluntary. Genetic testing sometimes requires obtaining informed consent from the patient or guardian, and many times this form is provided by the laboratory testing company or the provider's medical institution.



Getting Started

Quality Improvement: Small Tests of Change

The quality improvement (QI) process gives you the tools and resources to conduct small tests of change in your practice. By using QI methods, you'll see how setting goals and using small steps or tests of change can help you implement system-based changes. By taking small rapid steps and learning from each cycle, you'll end up with more successful and implementable process improvements. The scope of a small test of change could be introducing a new set of questions or form with 5 patient encounters in one afternoon. Each small test is rapid, manageable and leads to evaluation and further refinement of a process.

All of the practices who went through the QI project had to use rapid cycle tests of change to get things right. That's part of the process and that's how you learn what works and what doesn't. There isn't a one size fits all approach. Every practice is unique and it's up to you to figure out what works best in your practice. This section on QI is intended to help build a framework for your practice to engage in quality improvement methods in order to improve your care for patients with genetic conditions, and in obtaining a family history.



Getting Started

Tips for an Effective QI Team

Quality improvement teams should meet regularly (every 1-2 weeks) using established ground rules to share efforts, successes, and failures throughout the practice. Remember the ground rules of a good team meeting:

- All members and opinions are equal
- Team members will speak freely and in turn
 - Listen attentively to others
 - Each must be heard
 - No one may dominate
- Problems will be discussed, analyzed or attacked, **not people**
- All agreements are kept unless re-negotiated
- Members will respect the confidentiality of the team
- Once we agree, we will speak with “one voice” even after leaving the meeting
- Honesty before cohesiveness
- Each gets his say, not his way

Develop a Quality Improvement Team

Building Your Team

An active, motivated QI team is a key ingredient to success. Your team members should have knowledge and daily involvement with the processes involved in providing care for patients in your practice. They should also be interested in and available to plan and undertake improvements in the way in which care is delivered to patients and families.

First, pick your team members and clearly define roles of each team member. A multidisciplinary team with front office staff, nurses, other health professionals as well as PCPs often yield the best results because the team aims will reflect multiple perspectives from the setting. Choose 3 members, including a primary care provider, to form a core team. Other team members are typically a nurse or other clinician and an administrative or office staff member.

It may also be helpful to have input from different disciplines/perspectives at your clinical site (or individuals involved from different parts of the care process: primary care clinician, nursing, and administrative staff, medical records, information systems/data, etc). You may choose to invite representatives of these different perspectives to join your improvement team meetings on an ad-hoc basis. Gaining support of your IT or the staff person with direct interface with your electronic health record (EHR) has shown to be a strength in QI teams. They don't need to be a regular part of your QI team, but their support can help you achieve aims that impact your EHR.

It's best to keep your core members as consistent as possible throughout the project to help ensure success in implementing new strategies in your practice. It is recommended that your team also identify one or more parent partners to participate on your improvement team. In some cases, the development of a parent advisory panel that reflects the family perspectives and needs of your practice contributes an important voice that is grounded in practicality and experience.

Your team needs a **day-to-day leader or champion** who can act as the critical driving force, someone who can assure that tests of change are implemented, data collection and entry completed, and changes in your process are resulting in improved care for patients and families. It is helpful if the leader is someone with delegated authority to “get things done,” such as a clinic manager or head nurse. The day-to-day leader/champion needs to be able to work effectively with everyone involved and functions as the “key communication contact” between your team and project staff.

Many of the strategies your team tests will depend on making changes in practice culture and infrastructure. Such changes usually require the input of a variety of individuals and groups to adapt, test, and implement these changes in your setting. Therefore, another key ingredient for success is **engaging the senior leadership** in your practice to support your goals for improved integration of genetics into primary care practice. This should be someone with enough authority in the organization to institute change, overcome barriers that arise, and allocate time and resources the team will need to be successful. Such support will help remove implementation barriers and can help assure the long-term adoption of new strategies that enhance care for your patients.

Section 1

Getting Started

“Don’t let perfect be the enemy of the good”

-Voltaire

As your practice continues to collect family health history information, remember to share patient successes with front desk and other clinic staff who are part of the process. Positive reinforcement and the motivation of providing improved patient care will ensure sustainability and renewed engagement in adding additional tasks to the clinic flow.

(Prattville Pediatrics)

Involving Others

While the QI team directs the kind of work the practice focuses on, engaging all the staff in the practice is key to success.

Start your day with a team huddle. This assists in the practice's ability to organize the day's work and staffing needs, and to get on the same page about tests of process change with patient encounters. Map out the processes and personnel to determine if there are gaps or bottlenecks. Fostering “team thinking” can help your practice organize and address issues and schedules proactively on a daily basis.

Obtain buy-in from front line members and keep them regularly updated. This is just as important as obtaining leadership support, as front line workers know the most about processes and can help you develop ideas that will work for them and for your specific clinic's context.

Display your data everywhere! Remember the Hawthorne Effect? People work harder when they know someone is observing them. Share and post your data so it can have the maximum effect on your team.

Model for Improvement

Once you've selected your team, educate all team members on QI methodology. The Model for Improvement is a framework in which teams can strategically perform QI testing and cycles of change, using Plan-Do-Study-Act (PDSA) cycles.

The model provides organizing structure to guide thinking, ensure discipline and thoughtfulness, support improvement principles, facilitate improvement, and foster a common language. To get the most from the model: listen to patients and families, tap into knowledge of the system by involving staff, understand processes and interactions within and between systems, use a disciplined method of successive cycles to test changes, test on a small scale and move rapidly to improve, and measure to learn and to understand variation.

Three Key Questions for Improvement

What are we trying to accomplish?

AIM

How will we know that a change is an improvement?

MEASURES

What changes can we make that will result in an improvement?

IDEAS

Test Ideas and Changes in Cycles for Learning and Improvement



*Langley, Nolan, Norman, and Lloyd P. Provost.
The Improvement Guide: A Practical Approach
to Enhancing Organizational Performance.
New York: Jossey-Bass Inc., 1996.*

Getting Started

SMART Aim

- Specific: answer who, what, when, why, and where
- Measurable: use a quantity, quality or period of time to determine if you met your goal
- Actionable: what steps can your practice take to achieve the aim
- Relevant: the aim should match the overall change you are trying to achieve
- Time Bound: what can you do today, next week, next month to achieve this aim

Tools & Resources

- [Quality Improvement Basics](#) provides quality improvement tools and resources from the National Center for Medical Home Implementation.
- [Quality Improvement Innovation Networks \(QuIIN\)](#) is the host of AAP member networks that have an interest in quality improvement. Visit the QuIIN online resources for examples to help your practice team improve the quality and value of care and outcomes for children and families.
- [Institute for Healthcare Improvement \(IHI\)](#) is an independent not-for-profit organization that provides tools and resources for health and health care improvement.
- [How to Improve IHI](#) uses the Model for Improvement as the framework to guide improvement work.
- [Quality Reports](#) a section in Pediatrics.
- [EQIPP Courses: \(1\) Medical Home for Pediatric Primary Care and \(2\) Newborn Screening: Evaluate and Improve Your Practice](#) are online courses from the AAP that provide information, tools and guidance needed to make systematic and continuous practice improvements.

Determine your Aims, Measures, and Ideas

QUESTION 1:

What are we trying to accomplish?

AIM: A specific, measurable, actionable, realistic, and time-bounded (SMART) statement of expected results of an improvement process.

A strong clear aim gives necessary direction to improvement efforts, and is characterized as:

- Intentional, deliberate, planned
- Unambiguous, specific, concrete
- Measurable with a numeric goal, preferably one that provides a “stretch” to motivate significant improvement
- Aligned with other organizational goals or strategic initiatives
- Agreed upon and supported by those involved in the improvement and leaders

Make your aim actionable and useful. Include:

- A general description of what you hope to accomplish
- Specific patient population who will be the focus
- Some guidance for carrying out the activities to achieve aim

Getting Started

Tools & Resources

QUESTION 2:

How will we know that a change is an improvement?

MEASURES: Measures are indicators of change. To answer this key question (“How will we know that a change is an improvement”), several measures are usually required. These measures also can be used to monitor a system’s performance over time. In Plan-Do-Study-Act (PDSA) cycles, measurement used immediately after an idea or change has been tested helps determine its effect.

In improvement, key measures and measurement should:

- Clarify and be directly linked to goals
- Seek usefulness over perfection
- Be integrated into daily work whenever possible
- Be graphically and visibly displayed
- For PDSA cycles, be simple and feasible enough to accomplish in close time proximity to tests of change

QUESTION 3:

What changes can we make that will result in an improvement?

IDEAS: Ideas for change or change concepts to be tested in a PDSA cycle can be derived from:

- Evidence or results of research/science
- Critical thinking or observation of the current system
- Creative thinking
- Theories, questions, hunches
- Extrapolations from other situations

When selecting ideas to test, consider the following:

- Direct link to the aim and goals
- Likely impact of the change (avoid low-impact changes)
- Potential for learning
- Feasibility
- Logical sequencing
- Series of tests that will build on one another
- Scale of the test (3 patients, NOT 30)
- Shortness of the cycle (1 week, NOT 1 month)

Getting Started

Tips to Make the Most of PDSA Cycles

- Think a couple of cycles ahead
- Plan multiple cycles to test and adapt change
- Scale down size of test (number of patients, location), a “cycle of 1” is often appropriate
- Do more cycles, at a smaller scale and faster pace instead of fewer, bigger, slower
- Test with volunteers first
- Seek buy-in or consensus for the test
- Be innovative and flexible to make test feasible
- Collect useful (and only just enough) data during each test
- Test over a wide range of conditions
- Learn from failures as well as successes
- Communicate what you've learned
- Engage leadership support

Tools & Resources

Plan-Do-Study-Act

Once you've decided your aims, measures, and ideas, it's time to test those changes in your clinic using one or more PDSA cycles.

PLAN: You've already done a lot of planning in determining your aims, measures, and ideas. Now, plan the test or observation, making sure you have a plan for collecting data.

- State the objective of the test.
- Make predictions about what will happen and why.
- Develop a plan to test the change. (Who? What? When? Where? What data needs to be collected?)

DO: Try out the test on a small scale. A test of 1 patient for 1 day is appropriate for your first test. Make changes and then test with more patients.

- Carry out the test.
- Document problems and unexpected observations.
- Begin analysis of the data.

STUDY: Set aside time to analyze the data and study the results. What worked, what didn't work?

- Complete the analysis of the data.
- Compare the data to your predictions.
- Summarize and reflect on what was learned.

ACT: Make changes based on what you learned during the test.

- Determine what modifications should be made.
- Prepare a plan for the next test.

Adapted from the Institute for Healthcare Improvement.

<http://www.ihi.org/resources/Pages/HowtoImprove/ScienceofImprovementTestingChanges.aspx>

XYZ Pediatrics' QI team decided to test the completion and review of a family health history (FH) tool with all of the project's recommended components the one-month visit. The following is an example of their testing process, using a PDSA cycle:

PLAN: By the end of the next three months, our practice team will aim to complete FH tools for at least 90% of our patients seen for their one-month visit.

Copies of the Pediatric Family History tool will be made by Marie and available at the front desk. On Monday, at check-in the front desk staff will give the paper FH tool to the 3 families scheduled to come in to see Dr. Jones for their newborn's one-month visit. The staff will ask the parent(s) to complete the form while in the waiting room.

The QI team predicted that all 3 families would complete the tool while waiting to be seen. At the end of the day they will measure the number of FH tools received and completed. They will ask the front desk staff, clinicians and families for feedback about this process and the tool.

DO: On Monday, the FH tools were available for families with a scheduled one-month visit. Two of the families received the FH tool at check-in but 1 was given by the MA when rooming the family because the front desk staff forgot.

STUDY: One family (mom and dad attended the visit) completed their FH tool (they thought it was easy to understand) but needed to ask additional family members for information. One mom only knew about her side of the family, not the dad's. Dr. Jones did not get to review the tool with one family.

ACT: The team evaluated the results and decided the families need more time to complete the FH. We will try again but for the next test, Marie, our nurse, will explain the reason for collecting family histories and will plan to give the FH tool to parents at the 1-week visit, ask them to complete it at home with input from other family members and bring it back at the one-month visit.

Section 2

Collecting Family Health History



It all starts with family

Collecting Family Health History

Step 1

Understand the Value of Family Health History in Primary Care

A family health history (FH) is an inexpensive, non-invasive, and relatively simple yet valuable diagnostic tool within the primary care provider's (PCP) arsenal. Historically used to find single-gene disorders, FH has implications for much broader clinical use, including health promotion and risk assessment.

Family members share the same genes, environments, and behaviors. Because of these factors, they often share the same health problems, too. Family health history information can provide a full picture of the health of a patient and their family.

A comprehensive FH contains information from three generations of biological family members and is constantly evolving, which is why it is important for providers to regularly update the information.

We realize that the thought of collecting FH from all of your patients may seem overwhelming. Take this one step at a time, and start testing your tools and strategies with a few patients. In the subsequent sections, many strategies are provided to help guide you. For example, some providers from the GPCI learning collaborative started with obtaining FH from patients at the 1 month visit, or contacting OB providers to share information for newborn patients. Try not to get frustrated or overwhelmed with this process. As you uncover important information from your patients, you will be surprised!

Family health history information is only as valuable as the accuracy of the information provided and the ease of which it is collected and updated in primary care practice. Benefits of collecting FH include:

- A diagnostic tool and guide to testing and evaluation
- Identification of patterns of inheritance
- Resource to provide patient-education

Section 2

Collecting Family Health History

Step 1

Understand the Value of Family Health History in Primary Care

CONTINUED

Case Examples

While taking a detailed FH can be time consuming, there are many times when this information is critical in providing care to the pediatric patient and can define medical risks for extended family members.

1. You have a four year old girl in your practice who comes in for a routine health supervision visit. You expand the FH to include aunts, uncles, and cousins and learn that this child's mother lost a brother to HIV (acquired from tainted blood products) who had been diagnosed in early childhood with hemophilia A. Mom indicates that no one else in the family had hemophilia and she assumed her risk to be a carrier was low because of the unremarkable FH. You explain that although it is possible that her brother's condition was the result of a new factor VIII gene mutation, she needs to consider genetic counseling and testing, since it is still possible that she is a carrier of this X-linked recessive condition. Genetic counseling and testing reveals that she is in fact a carrier, and this information is critical for this couple who now know their reproductive risks and the family planning options available to them.

2. A mother brings in her 10 year old son with an acute pharyngitis and tells you that her husband was diagnosed with medullary thyroid cancer 9 months ago and had widespread disease at the time of diagnosis. The doctors have given him a very poor prognosis, but they are pursuing some experimental treatments. In reviewing the extended FH, you learn that no one else in the family has had thyroid cancer, but that the boy's paternal grandmother, age 57, developed hyperparathyroidism in her 30s and required surgical resection of the parathyroid glands. Based on this FH, you are even more concerned about the possibility of MEN2 in this father, with a 50% risk for transmission to his child. Since dad is quite ill, it would be valuable to consider either doing genetic testing on the father now, or saving some DNA for future genetic testing. If this boy has inherited a RET gene mutation from his father, then he will need prophylactic thyroidectomy and close follow-up for other tumors associated with MEN2. If the boy did not inherit a mutation from dad, then he is not at risk for these other problems and requires no additional surveillance. You recommend that the family go for cancer genetic counseling through the oncology program where dad receives care.

Section 2

Collecting Family Health History

Step 2 Recognize Genetic Red Flags in Family Health History

Most disorders that have a genetic component will not follow classic Mendelian patterns (e.g., autosomal dominant, autosomal recessive, or X-linked). Therefore, providers will have to depend upon other clues, or red flags, to identify potential genetic influences on disease. Understanding these genetic red flags will facilitate the interpretation and assessment of FH information.

Genetic red flags are indications that there is a potential for genetic risk in an individual or family. The red flags indicate unusually high genetic and/or environmental risk, and the risk for close relatives may be increased dramatically. Generally, the same red flags increase risk for Mendelian and common disorders.

The Rule of Too/Two

Family health history questions that result in answers using the descriptors "too" or "two" may indicate a genetic condition.

(Source: Arthur Grix, MD)

- TOO Tall
- TOO Short
- TOO Early
- TOO Many
- TOO Young
- TOO Different
- TWO Tumors
- TWO Generations
- TWO in the Family
- TWO Birth defects

Examples of some genetic red flags include:

Family history of multiple affected family members with the same or related disorders. These disorders may or may not follow an identifiable pattern in the family.

- Multiple family members affected by the same or related disorders indicates increased risk, whether through genetic or environmental risk factors, or a combination of genes and environment.
- Examples: Three family members in two generations with heart disease; a father and son with diabetes; two family members with mental disability.

Earlier than expected age at onset of disease.

- Disorders that arise at a younger age than expected may occur because of a genetic predisposition that makes an individual more susceptible to environmental exposures.
- Examples: Heart disease occurring in the 30s; colon cancer in the 30s.

Condition in the less-often-affected sex.

- Due to a genetic predisposition, a disorder may occur in the less common sex. Genetic factors can override other hormonal, developmental and environmental factors and contribute to the condition's occurrence.
- Example: Breast cancer in a male.

Disease in the absence of known risk factors.

- Occurrence of a disorder in the absence of obvious environmental factors may be attributed to a genetic predisposition.
- Example: High cholesterol in an individual with an ideal diet and exercise regimen.

Ethnic predisposition to certain genetic disorders.

- Some genetic disorders are more common in certain ethnic groups. Awareness of a patient's ethnicity or ancestral background can aid in recommending genetic testing and evaluation of genetic conditions.
- Examples: Thalassemia in an individual of African ancestry; BRCA 1/2 in an individual of Ashkenazi Jewish ancestry.

Close biological relationship between parents (ie, consanguinity).

- Consanguinity is a relationship by blood or through common ancestor. Relatives are more likely to share the same genes; therefore children from consanguineous couples related as first cousins or closer have an increased risk of having an autosomal recessive condition.
- Examples: Autosomal recessive disorders, including those that are part of the newborn screen (including MCADD and cystic fibrosis), are more common in consanguineous couples.

Collecting Family Health History

Step 3 Review the Components of a Meaningful Family Health History

Tips for Collecting Family Health History

Recorded FH information should include the following information:

- Date the information was collected
- Name of person providing information
- Ancestral background/ethnicity
- Consanguinity
- Adoption status

A comprehensive FH contains information from three generations of biological family members and is constantly evolving, which is why it is important for providers to regularly update the information. Family health history should contain the following components for each relative:

- Relationship (eg, full or half sibling, adopted)
- Sex
- Age or year of birth
- Medical conditions and age at diagnosis
- Pregnancies and any complications
- Age at death and cause of death (if known)

A new generation is defined by the birth of an individual's offspring (eg, great-grandparent, grandparent, and parent). Relationships to family members can be defined in terms of degrees:

- First-degree relatives: parents, brothers, sisters, and children
- Second-degree relatives: aunts, uncles, nieces, nephews, grandparents, and grandchildren
- Third-degree relatives: first cousins

Section 2

Collecting Family Health History

Step 3

Review the Components of a Meaningful Family Health History

CONTINUED

Components of a Family Health History

A comprehensive FH tool should include general family health history elements for three generations of family members (defined below), including: parents, brothers, sisters, aunts, uncles, first cousins, and grandparents.

Defining three-generations:

- First generation relatives: child, siblings, cousins
- Second generation relatives: parents, aunts, uncles
- Third generation relatives: grandparents

Ask the family about medical conditions that run in the family (2 or more family members with the condition). Be sure to document who and what for each of the following:

- Birth Defects
 - Structural birth defects (ie, congenital heart disease, spina bifida, extra or missing fingers, clubfeet)
 - Sensory birth defects (ie, congenital deafness, congenital blindness)
- Cancer <50 years and specify type
- Carrier of genetic condition
- Clotting, bleeding, or blood disorder (ie, sickle cell, von Willebrands, hemophilia)
- Developmental delay, intellectual disability, autism spectrum disorder, or learning disability
- Early Death: Sudden, unexpected, or unexplained death <50 and details of event
- Heart attack: <55 years for males and <65 years for females
- Known genetic condition
- Multiple miscarriages or stillbirths
- Received special education services
- Seizures

Unknown Family History

In some cases, the family history information is missing from one side of the family, due to estranged family members, adoption, or other reason. In this situation, it is most important to remember that the absence of family history information does not mean that the child does not have genetic risk from the missing information, or that they are not a carrier of a genetic condition. Strategies to obtain information include asking if they know any other family members who could provide information, asking the adoption agency for access to health records if available, or contacting the birth family for information. However, if the information is not retrievable, the PCP should focus on the child's clinical symptoms.

Collecting Family Health History

Step 4 Choose a Family Health History Tool for your Practice

Now it's time to start collecting FH. First, make an aim statement for collecting FH and then pick a FH tool. You may want to start with a tool you already use and incorporate the recommended components from the Components of a Family Health History section, or you may want to use a new tool. If you choose to use a new tool, there are two examples included in the toolkit that you can use as is or modify to meet your practice's needs. There are additional tools, including Spanish versions, on the [Genetics in Primary Care Institute Web site](#).

Make sure to consider the demographics of your patient population when choosing a FH tool. Some practices participating in the QI project found that having a Spanish version or determining the culturally appropriate ways to ask sensitive questions was important to ensuring the FH is correct.

Plan Your QI

Throughout this toolkit you will see sections called "Plan your QI." These sections are intended to help you develop aims, changes, and ideas relating to QI PDSA cycles. You can also refer to the case study on page x for an example of aims, changes, and ideas.

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next quarter, our practice team will assess FH, using a tool with all of the recommended components, for all of our patients at their 1-month visit.

How will we know that a change is an improvement? Our measures are:

- At least 90% of our 1-mo. old patients each month will have a documented FH in their record. We will collect feedback from staff, physicians and families about this process and the tool.

What changes can we make that will result in an improvement? Our ideas for change are:

- We will give the FH tool to all families at their newborn's 1-week visit and ask them to complete the questions and bring it with them to the 1-month visit.

Collecting Family Health History

Step 4

Choose a Family Health History Tool for your Practice

CONTINUED

Changing the Family Health History Documentation Tools in Your Electronic Health Record

If you decide to document FH in your electronic health record (EHR), you may need to work with your organization's information technology department or your vendor representative to adjust the EHR family history documentation tools to include the recommended family health history components.

Depending on your EHR and what changes you decide to make to your FH, the practices that participated in the QI project recommend:

- Document that you reviewed FH with the patient/family. In order to assist with documentation of the FH within the EHR, create “dot” phrases that will give you a statement that will say “family history collected and reviewed” and the action that you took, such as placing the concern on a problem list or requesting additional information from the family.
- Add a prompt to ask for 3 generations of family history. Most practices don't ask for 3 generations of family history. Adding a prompt can help staff remember to collect information about all three generations.
- Model the FH section in your EHR to match the paper FH forms. Once your practice has decided upon a FH tool, it will be helpful have the EHR mirror the paper form. For example, the conditions are listed on the left and the corresponding family members are shown on the top. This modification will decrease data entry time and is useful because scanning the form into the medical record doesn't help with tracking and making the data/information quantifiable.
- The FH should be reflective of pediatrics. Many EHR systems are designed to serve adult patient populations. The FH concerns of adults are different than for pediatrics. As you begin collecting FH information, consider the following types of structured data and how it can be modeled to be reflective of your pediatric patient population:
 - Addition of elements such as adoption status, conception through IVF, and ethnicity
 - Pediatric conditions/diagnoses that should be included (intellectual disability, congenital hearing loss, etc)
 - Family member structure (half-siblings, step-parents, etc)
 - Family member functionality (link siblings' information)

Section 2

Collecting Family Health History

American Academy of Pediatrics-Committee on Genetics Pediatric Genetic Screening Questionnaire

Child's Name:

Date of Birth:

Today's Date:

Mom's Name:

Dad's Name:

Check One: The child is born to the above parents is Adopted has a Legal Guardian

What is (are) the ethnic background(s) of the child's family members?

(Examples: Jewish, French, African-American, Irish, Hispanic, Asian, Native American, etc.)

Please answer all questions if possible. Remember that the questions are about the biological parents/relatives of the child. We hope this information will help us to better care for your child.

Child's Health Survey

NO	YES	Don't know
----	-----	------------

Do you have concerns about your child's growth and/or size?

Do you have concerns about your child's ability to learn?

Has your child attended an Early Intervention Program (EIP) or special education classes?

Are you concerned that your child may have an inherited (passed on in the family) problem?

Pregnancy Background

NO	YES	Don't know
----	-----	------------

Did the mother have any illnesses in the first three months of pregnancy or take any medications?

Did the mother take drugs or drink alcohol during her pregnancy?

Has the mother had 2 or more miscarriages?

Has the mother had a stillbirth or a baby that died in the first weeks of life?

Family History

Have you been told or do you think that your child or anyone in the child's mother's or father's family (including brothers/sisters, grandparents, aunts, uncles, cousins) had any of the following:

NO	YES	Don't know
----	-----	------------

Learning disabilities, mental retardation, or autism?

If yes, has this person had any testing done?

Down syndrome or other change in the chromosomes?

Spinal or cranial defect (open spine, spina bifida, anencephaly)?

Opening in the lip or roof of the mouth (cleft lip, cleft palate)?

An abnormal brain or skull (water on the brain, missing skull)?

Partial or total deafness or blindness?

Heart defect present at birth?

Abnormalities of an arm or a leg (missing or extra parts)?

Any chronic (long-lasting) skin condition or many birth marks?

Any other birth defects or physical problems in the child's family members?

Any bleeding disorder or hemophilia?

Muscle disease or muscular dystrophy?

Any severe anemia, sickle cell trait or sickle cell disease?

Any serious lung disease or cystic fibrosis?

Seizures or convulsions?

A heart problem or heart attack before age 55 in males or 65 in females?

Cancer or tumors before age 50?

Sudden, unexpected, or unexplained death before age 50?

Has anyone in the families been told he or she is a carrier for any genetic disease?

Do you suspect any other genetic or hereditary conditions in the child's family members?

Are the parents of the child related to each other?

Section 2

Collecting Family Health History

Family History for your Child

Child's Name:

Today's Date:

Put an X for all of **your child's** biological relatives who have the condition.

Comments:

	Child's Mother	Child's Father	Child's Sister	Child's Brother	Child's Grandmother Mother's side	Child's Grandfather Mother's side	Child's Grandmother Father's side	Child's Grandfather Father's side	Child's Aunt(s)	Child's Uncle(s)	Child's Cousin(s)
ADHD/ADD											
Asthma											
Autism spectrum disorder, PDD-NOS, Asperger's											
Birth Defect											
Bleeding or clotting disorder											
Cancer before age 50											
Born with an eye/vision problem											
Born with hearing loss											
Born with a heart problem											
Diabetes											
Early heart disease (<55 in men, <65 in women)											
Genetic syndrome or condition											
High blood pressure											
High cholesterol or triglycerides											
Kidney Disease											
Mental or mood disorder											
Obesity											
Seizures											
Sudden cardiac death											
Other condition that affects 2 or more family members											
Does not have any of the conditions listed above											
No information about this relative											

Are the child's mother and father related to each other except by marriage? No Yes

Do you have any other concerns about your child's family health history?

Any suggestions?

Collecting Family Health History

One practice waited two months before they transitioned all providers to the family health history tool. At first, a small group of providers started testing forms and processes. As other providers began collecting family health history as well, they used medical assistants to help with the data input. The practice conducted a structured training for how to input the family health history information and this worked well for improving the quality of data being collected and recorded.

Step 5 Test Your Family Health History Tool and Process

Once you've selected your FH tool, you want to develop and test a process for collecting the information. Process mapping can help you figure out the best place in your clinic to collect FH.

Process Mapping

Process mapping is an exercise where everyone on your team who is involved in a process, creates a workflow diagram of the entire process. Process mapping can help your team identify waste, identify communication or hand-off issues, identify all the people involved in a process, show complexity, explain the whole process to the group, and identify who should be involved in a project to improve the process.

According to the American Society for Quality, the basic process for process mapping is to:

- Gather the necessary materials: sticky notes or cards, a large piece of flipchart paper, and pens (different colors can be helpful)
- Define the process to be diagrammed
- Write its title at the top of the work surface
- Discuss and decide on the boundaries of your process: Where or when does the process start? Where or when does it end?
- Discuss and decide on the level of detail to be included in the diagram
- Brainstorm the activities that take place
 - Write each activity on a card or sticky note
 - Sequence is not important at this point, although thinking in sequence may help people remember all the steps
- Arrange the activities in proper sequence
- When all activities are included and everyone agrees that the sequence is correct, draw arrows to show the flow of the process
- Review the flowchart with others involved in the process (workers, supervisors, suppliers, customers) to see if they agree that the process is drawn accurately

Once you've developed a complete process map, look for things that you can improve or places where you can add the QI strategy to your practice. You want to look for places where you can:

- Eliminate things that aren't used
- Eliminate multiple entry
- Minimize handoffs
- Move steps in the process closer together
- Do tasks in parallel
- Reduce choice of features
- Give people access to information
- Take care of basics
- Coach staff/clinicians to use the product, service, or tool
- Reduce setup time

Collecting Family Health History

As you begin collecting family health history information from your patients, ensure that your patient population is clear about which family members the family health history information is about. Particularly among low-literacy populations, if a parent is completing a paper family health history form on behalf of the child, descriptions such as “grandparent” and “aunt” may be confusing. Clarify “child’s grandmother/mom’s mother” instead of “maternal grandmother,” or ask the front desk/nursing team to explain the structure of the form to the family.

Example Practice Policy for Family Health History Collection: Advice from a primary care practice:

As part of each patient's health supervision visit, our practice will collect a multi-generational family health history, using a standardized collection form. The family health history will be documented, discussed with the patient/family, and updated annually.

Step 5

Test Your Family Health History Tool and Process

CONTINUED

Test Your Process

Once you have an idea of how this could work, start a small PDSA cycle with your new tool. Try it on 1 patient on 1 day and get feedback from all involved. If it works great, expand it to 3 patients on 1 day, and then get more feedback. Keep going like this until you are using the tool in your whole practice. These iterative cycles of change allow you to improve your FH tool, your process for getting the FH, and get feedback and buy-in from the front line staff as you are listening to their ideas on how to improve the process!

Practices participating in the QI project used different processes to collect family history. One of these options might work for your practice.

“The one month well-baby visit has shown to be a good time to review family health history with the parent because there are no vaccinations to give and the parent is more out of the new parent ‘fog.’ Sometimes it works best to give the form to the parent and have them bring it back at the next health supervision visit, too.”

“It can take time to take a full family health history. We began to collect the family health history at the newborn visit and then at subsequent visits we added another layer of questions, so that by age 2 there is a complete family health history for that patient.”

“We discovered that a somewhat comprehensive family health history is taken at the OB visit when the expecting mother is pregnant, so we tried to connect our EHR record with the previous appointments. Beginning to take the family health history when and if the expecting parents visit the pediatrician before the birth of their child may also be a good strategy.”

After your team has planned, tested, and implemented practice-based processes for the use of a standardized FH tool for all patients, it will be important to write them down! A written process or protocol ensures everyone in the practice knows what they are supposed to do.

Collecting Family Health History

Motivation to Adopt Change:

"After using a family health history tool, our practice found three kids with genetic conditions, which we do not think we would have found without taking the family health history. It has been positive to be able to find information that may help prevent future health issues. It has also been positive to talk with patients and get to know them."

Step 6 Review and Discuss Family Health History with Patients

Discuss FH information with the patient and family, regardless of the results. As noted above, first make an aim statement about reviewing FH with patients. Then identify some measures and change strategies, and implement in small PDSA cycles.

Explain the Importance of Family Health History

Family members share the same genes, environments, and behaviors. Because of these factors, they often share the same health problems, too. Family health history can provide a full picture of the health of a patient and their family. Explain why it is important to gather FH information and how it will allow you to provide better care for the patient.

Family History Resources for Patients

- [Family Health History](#) is a patient-friendly resource for family history and genetic testing information from the Centers for Disease Control and Prevention.
- [My Family Health Portrait](#) is a tool from the Surgeon General for patients and family members to enter their family health history information and turn the submitted information into a pedigree that individuals can bring to their PCP. Patients can also share their information with other family members.
- [Family History is Important for Your Health](#) patient handout from the Centers for Disease Control and Prevention (English and Spanish).
- [Family Health History Record Keeper](#) is a printable tool from the Illinois Department of Public Health that providers can use to talk to their patients about the importance of gathering family health history. It includes a place for patients to write their family health history information down.
- [Does It Run In the Family?](#) is a downloadable tool, created by Genetic Alliance, that allows individuals to customize, download, and print a set of two booklets that promote collection of family health history information to increase knowledge of and communication about family health.
- [National Coordinating Center for the Regional Genetic and Newborn Screening Collaboratives](#) provides the resources to assist consumers and their families in accessing information about genetic services and resources at a national level.
- [The American Society of Human Genetics video series](#) about collecting family history information.

Collecting Family Health History

Step 6

Review and Discuss Family Health History with Patients

CONTINUED

Be Aware of Sensitive Topics

Exert caution and compassion for families when discussing sensitive topics. Some examples include:

- Adoption
- Assisted reproduction
- Consanguinity
- Estranged family member
- Death and illness in the family
- Health behaviors, including substance abuse

Consanguinity

Talking about consanguinity with families can be difficult. Families may not want to reveal their relatedness if they feel it may be perceived negatively. It's important that families feel comfortable sharing this information so it can be documented for future reference and they can receive the appropriate medical care.

First cousins share 1/8 of their genes in common and their children are genetically identical for 1/16 of their genetic material. For consanguineous unions there is an increased risk for health problems related to possible autosomal recessive conditions, but should not prompt an automatic genetic referral. Second cousins and more distant unions appear to have no greater risk for adverse outcomes than the general population.

Tips for talking to families about consanguinity from practices participating in the QI project:

- Explain that as part of a way to provide better care for all of their patients, this question is being asked of all families. This strategy is helpful so families didn't feel singled out.
- Explain to families that in order to provide better quality of care, you are updating family histories and these questions will help providers with understanding pieces of the child's health.
- Ensure patient privacy and confidentiality by asking about things like adoption status and consanguinity in private, when the child is out of the room.

Section 2

Collecting Family Health History

Step 6

Review and Discuss Family Health History with Patients

CONTINUED

Notice Clues to Missing Information

Notice clues to potentially inaccurate or missing health information, for example, there are no reported cases of cancer in the family and everyone in the family is described as “healthy.” If the parent seems doubtful of recalling the exact condition, recommend that they communicate with their family to obtain accurate information. The following strategies may help you communicate FH information to families:

General Tips

- Exercise compassion when discussing the family health history
- Avoid the terms, “positive”, “negative”, and “uneventful”
- Use the term “condition” instead of “disease”
- Soften your questions with phrases such as, “Is there a possibility of...?”
- If family members are unsure of specifics, such as the type of cancer, make a note or flag in the EMR to prompt recording the information at a future health supervision visit

Suggested Questions for Families

- “I would like to ask you some questions about your health and about the health of your family members. Having this information will help to provide the best care for your child.”
- Address clues to inaccurate or missing information by probing, “Does anyone in the family take any medications?”
- “Do your (brothers, sisters, mother, father) have any health problems?”
- “Are all of your children with the same partner?”
- “Do you share the same mother and father with your siblings?”
- “Is there a possibility that you and your partner/spouse are blood related?”
- “Are there any family members with intellectual impairment or severe learning disabilities?”
- “Are there any family members who had health problems from birth (for example, of the hands or feet, spine, heart)?”
- “How much alcohol did you use during pregnancy?”
- “What medications did you take during pregnancy?”
- “Do you know your ethnic background?”
- “Are there any other conditions that run in your family?”
- “Do you have any concerns about a condition that may be genetic/that runs in your family?”

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 3 months, our clinical team will review and discuss FH with 95% of our patients/families, using the recommended strategies to help communicate FH information.

How will we know that a change is an improvement? Our measures are:

- 95% of our patients with a completed FH will have documentation in their record that the information was reviewed and discussed. Feedback will be collected from clinical staff and families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Education of clinical staff about suggested strategies/scripts for correcting inaccurate or incomplete FH information. Add prompt to EHR to review/discuss FH and documentation that FH information was reviewed/discussed with family.

Section 2

Collecting Family Health History

Step 7 Evaluate your Family Health History Tool

Part of QI is evaluating your changes and making the necessary adjustments. Staff and families can provide valuable input about what is working well and what needs to be adjusted.

Check in with your team to obtain feedback about how the FH collection is being received. Ask other providers and clinicians:

- What is working well with the FH form or the process for collecting the information?
- Do you have any recommendations to change formatting or language of the FH form?
- Are there any ways we can improve efficiency of using the form?
- Have you noticed any patient concerns regarding the questions on the FH form?
- Are there areas where patients/families could not provide complete or accurate information?
- Does anyone have suggestions for how to improve the clinic workflow for collecting FH?

Collect feedback from your patients either formally or informally. Here are some suggestions:

- On the bottom of the family health history form, add a spot to ask families how long it took them to complete the questionnaire, and if they have any questions or suggestions regarding the form.
- As you review the FH with the patient/family, ask them if they have any suggestions for improving the form or what their reaction to providing this information was.

Use the feedback to modify your form and process as needed.



Collecting Family Health History

Tip:

Collecting family health history information from your patients should be an iterative process. If another family member attends an appointment—such as the father or a grandparent—use it as an opportunity to gather additional information.

Step 8 Maintain and Update your Patient's Family Health History

It's important to update FH at each visit. You'll want to follow up on information families didn't know during the previous visit and ask about new diagnoses/conditions. Think about the best way to collect this new information. Will you put a note in the chart to follow up on missing information? Will families fill out a form to update their FH at each visit?

Questions for Updating Family Health History

The following questions can be useful to ask families when updating a family health history.

Since your last visit, has anyone in the family died? If so, how is this family member related to the child? How old were they when they died? What was the cause of death?

Since your last visit, are there any individuals in the extended family including brothers, sisters, parents, grandparents, aunts, uncles, or cousins of the child who have been born and have or are newly diagnosed with any of the following? If so, how is that individual related to the child?

- Blindness
- Birth defects (examples: congenital heart disease, spina bifida, extra or missing fingers, clubfeet)
- Congenital deafness (born deaf)
- Developmental delay, intellectual disability, autism spectrum disorder
- Facial features that look different from other family members
- Known genetic disorder
- Multiple handicaps
- Multiple miscarriages or stillbirths
- Organ failure (heart failure, kidney failure, liver failure) prior to age 40
- Seizures

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 6 months, our practice team will routinely create/maintain/update a patient's FJH as part of a health supervision visit for at least 90% of our patients 1-21 years old.

How will we know that a change is an improvement? Our measures are:

- 90% of our patients 1-21 yrs. old will have a FH created/maintained/updated as part of each health supervision visit. Feedback will be collected from clinical staff and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Conduct education/training of clinical staff about creating/maintaining/updating a FH, using the suggested questions. Test having MA/Nursing staff ask questions when patient/family is roomed and have physician review/discuss changes noted.

Section 3

Identifying Patients with Suspected Genetic Conditions



Recognize, Ask, Discuss

Section 3

Identifying Patients with Suspected Genetic Conditions

Step 1 Recognize Clinical Red Flags

Become comfortable with recognizing red flags in the family and clinical history of the patient. Page X of this toolkit provides information about red flags in the FH. The following are medical conditions/concerns that may suggest an underlying genetic etiology:

Significant Neurological Problems

- Autism spectrum disorder
- Brain malformation/abnormal brain MRI findings
- Congenital or acquired hearing loss
- Developmental delay/intellectual disability
- Seizure disorder (especially hard to control)
- Significant hypotonia, hypertonia, and/or spasticity
- Progressive muscle weakness/peripheral neuropathy/ataxia

Congenital Anomalies

- Congenital heart disease
- Congenital diaphragmatic hernia
- Dysmorphic features
- Limb or bone malformation such as clubfeet, missing, or extra digits
- Renal agenesis
- Tracheal esophageal fistula

Growth Problems

- Disproportionate growth/hemihyperplasia/marfanoid habitus
- Failure to thrive
- Excessively tall structure
- Intrauterine growth restriction/small for gestational age
- Microcephaly or macrocephaly
- Short stature

Other

- Abnormal skin findings: café-au-lait spots, multiple lipomas, ash-leaf spots
- Bilateral or multifocal malignancies: Wilms tumor or retinoblastoma
- Cardiomyopathy not due to viral infection
- Clotting abnormalities: thrombophilia or excessive bleeding



Identifying Patients with Suspected Genetic Conditions

Motivation to Adopt Change:

"The parents have expressed gratefulness that we had brought up questions about their family health history. Taking time to connect the dots and connections between the health status of their relatives and tendencies for their children's health has meant a lot to them."

Step 2 Identify and Follow-up with Patients Identified at Risk

An at-risk patient is a patient with a positive FH and/or identified clinical concerns (physical signs, cognitive concerns) of a genetic condition. Once you know a patient has a positive FH or signs of a genetic condition, you can provide the appropriate follow-up.

Follow-Up

When collecting FH information, ask for more information if the family indicates "yes" to any conditions on the FH questionnaire. Recommended follow-up questions are broken up by condition to help you gather more information about any condition the family indicates "yes" to in the FH.

If there are multiple miscarriages, stillbirths or unexplained pregnancy losses (3 or more for one couple or person)

- What is the relationship to the patient?
 - Were any birth defects or problems noted in the fetus/baby prior to the pregnancy losses?
 - Were genetic tests done on POC* or baby?
- *POC = products of conception/tissue from miscarriage or stillborn

If there are developmental delays/intellectual disabilities/autism spectrum disorder/significant learning disabilities

- What is the relationship to the patient?
- Are there any other health problems? If yes, what problem(s)?
- Was any genetic testing done? If yes, what testing?

If there are structural birth defects (such as heart defects, spina bifida, extra or missing fingers, cleft lip/palate, etc)

- What is the relationship to the patient?
- What type of birth defect?
- Isolated or with additional defect?
- Isolated or with developmental problems?
- Are there other health problems?
- Was any genetic testing done?

If there is congenital or acquired blindness

- What is the relationship to the patient?
- What is the cause of blindness?

If there is congenital or acquired hearing loss

- What is the relationship to the patient?
- Does the person wear hearing aids?
- Is the person able to speak?
- Does the person have abnormal external ears?
- Are there any birth defects?
- Was any genetic testing done?

If there is a chronic long lasting skin conditions or many birth marks

- What is the relationship to the patient?
- What is the name of the condition or description of birthmarks?
- Are there any other health problems besides the skin condition?

Identifying Patients with Suspected Genetic Conditions

Step 2

Identify and Follow-up with Patients Identified at Risk

CONTINUED

If there is a bleeding disorder or hemophilia

- What is the relationship to the patient?
- What is the specific name of the condition?
- Has this person had any genetic testing for their condition? If yes, does your relative have a copy of their test result that they could/would share with you?

If there is muscle disease or muscular dystrophy

- What is the relationship to the patient?
- What is the specific name of the condition?
- Has this person had any genetic testing for their condition? If yes, does your relative have a copy of their test result that they could/would share with you?

If there is severe anemia, Cooley's anemia/beta-thalassemia or sickle cell disease, beta-thalassemia trait or sickle cell trait

- What is the relationship to the patient?
- What is the specific name of the condition?
- Does your relative have a copy of their test result that they could/would share with you?

If there is serious lung disease or cystic fibrosis

- What is the relationship to the patient?
- What is the name of the condition?
- At what age was the family member diagnosed?
- Has this person had any genetic testing for their condition? If yes, does your relative have a copy of their test result that they could/would share with you?

If there are seizures

- What is the relationship to the patient?
- What age did the seizures start?
- Are there any associated problems?

If there are heart attacks under the age of 55 in males or 65 in females

- What is the relationship to the patient?
- At what age did the family member have the first heart attack?
- Are there any risk factors, such as high cholesterol/hyperlipidemia, smoking, obesity/overweight, or hypertension/high blood pressure?

If there are cancers under the age of 50 years

- What is the relationship to the patient?
- What is the location and type of cancer?
- What was the family member's age at diagnosis?
- Anyone else in the family with a related cancer?

If there is sudden, unexpected, or unexplained death <50

- What is the relationship to the patient?
- What was the family member's age and cause of death?
- If not accidental or traumatic, was an autopsy or postmortem exam done?

Section 3

Identifying Patients with Suspected Genetic Conditions

Step 2

Identify and Follow-up with Patients Identified at Risk

CONTINUED

If there is a known carrier of a genetic condition

- What is the relationship to the patient?
- What is the name of genetic condition/trait?
- Does your relative have a copy of their test result that they could/would share with you?

If there is a known genetic condition

- What is the relationship to the patient?
- What is the name of genetic condition?
- How was the diagnosis made?

If the parents of the child are related to each other

- How are the parents related?
- Has anyone in the family been told they have an autosomal recessive condition or carry an autosomal recessive condition?

Tools & Resources

- [Genetic red flags: clues to thinking genetically in primary care practice \(article\)](#)
- [Role of ethnicity in genetics, resources from the Genetics in Primary Care Institute \(GPCI\)](#)



Section 3

Identifying Patients with Suspected Genetic Conditions

Motivation to Adopt Change:

"There are amazing stories almost every day in taking the three generation family health history. Sometimes a tad more probing yields great results. Asking the right questions can sometimes result in building a heightened awareness of health conditions in the family in parents, and point families in the direction of a genetic referral."

Step 3 Talk to Parents about an At-risk Family Health History or Genetic Referral

Many PCPs find it difficult to broach the subject of a genetic referral. They are often concerned that these discussions will cause great parental anxiety and that families may resist going for the needed evaluations. Below are some of the reasons that genetic referrals may be helpful for a child in your practice and some language that may be helpful in beginning the dialogue about the referral. Also, at the end there is an outline of what a typical genetic evaluation entails.

Possible Benefits of a Genetic Diagnosis

- **Plan for future care.** Having a specific diagnosis of a genetic condition may help you as the healthcare provider plan for future care. Perhaps this is a bit of a twist on anticipatory guidance, but if a child was known to have a condition associated with kidney problems or metabolic derangements, you would know which tests to order or what additional surveillance you need to employ to make sure the child remains healthy.
- **Information for long-term outcome.** Having a specific genetic diagnosis can provide valuable information to the family about the long-term outcome which may include how other children and young adults have done with this particular condition, specific therapies or interventions that are especially helpful, and/ or focused approaches to educational interventions.
- **Prevent unnecessary care.** Knowing a specific genetic diagnosis may also prevent unnecessary testing and subspecialty referrals, since clarification of the diagnosis may preclude the medical odyssey that many children with no diagnosis are engaged in.
- **Inform health of family members.** Having a specific diagnosis may provide valuable information for the parents, siblings, and other family members regarding risks for recurrence. For many genetic conditions there is no increased risk for future children or reproductive risks for siblings, but either way, having this information gives individuals the chance to make informed decisions.

Case Examples: Genetic Referrals

Referral for speech delays, streaky skin and poor handwriting reveals significant learning and physical issues

A 5-year-old was referred for speech delays and streaky hyperpigmentation of the skin. Her teacher reports poor handwriting and that she didn't seem to be trying very hard. Testing revealed a mosaic marker chromosome associated with more significant learning issues and physical issues such as hypotonia, which caused weakness in her hands and accounted for her poor handwriting. School placement and support services such as occupational/physical therapy were re-evaluated.

Referral for albinism avoids potential disaster

A 12-year-old boy with albinism was referred for genetic evaluation because his mother was pregnant. When asked about bruising, they confirmed that he bruised easily. Based on that, it was suspected and confirmed that he had Hermansky-Pudlak syndrome instead of simple albinism, which put him at risk for excessive bleeding with surgical procedures, pulmonary fibrosis and granulomatous colitis. Knowing this, he could be better monitored and prepared for future surgeries.

Referral of mom with disabled children demonstrates risk for sister's pregnancy

A woman was referred for genetic counseling because she had two children with severe intellectual disabilities and minor birth defects and her sister was pregnant and wanted to know if she could be a carrier of the condition. Family health history was otherwise unremarkable and prior genetic testing was negative. A microarray was ordered on her children that revealed an unbalanced translocation. Specialized cytogenetic studies were performed to determine if prenatal testing was indicated.

Identifying Patients with Suspected Genetic Conditions

Step 3

Talk to Parents about an At-risk Family Health History or Genetic Referral

CONTINUED

Strategies and Scripts for Communicating with Families

Possible introductory statements before suggesting a genetic referral

Child with multiple café-au-lait spots and you are concerned about the possibility of neurofibromatosis (you can substitute any physical finding or potential diagnosis of concern in here)

"On the exam today I see that _____ has a number of these flat birth marks. Although many people have **these same birth marks**, when I see **more than six** on exam, I want to be sure that there isn't a genetic connection. The **birth marks themselves** are perfectly harmless, but because I want to be very thorough, I am recommending that _____ be seen by a genetic specialist who can help determine if we need to look any further." If the parent asks about the condition you might add "The name of the condition that is sometimes associated with **these birth marks** is **neurofibromatosis or NF**, but we cannot possibly make this diagnosis unless there are other things we find on exam. While I do not recommend that you go online and read up about this condition before your genetic evaluation, if you feel you need more information, I can provide you with a Web site that has correct information because there is a lot of misinformation on the Internet."

Child with developmental disabilities and/or autism spectrum disorder

"As you know _____ has been diagnosed to have learning disabilities/developmental delays/autism/autism spectrum disorder. Although many times we do not identify a specific cause for these developmental or behavioral issues, in some cases there may be an underlying genetic cause for these findings. Although _____ is receiving the necessary therapies to help him/her to maximize his/her developmental potential, if there is a specific genetic cause for these problems, then knowing a more precise diagnosis can help me provide better medical care and help the school/therapists design the best educational plan for _____."

"About 5 to 10% of children with developmental difficulties are found to have a genetic cause for their problems. If we know the precise cause, then it can help us plan better for _____'s future and also to provide information to you about whether or not this is a change that only _____ has within his/her genetic make-up or something that could possibly run in the family or be passed along to future generations."

Dealing with reticent or anxious parents

"On my exam today I see _____. I want to be sure there isn't a genetic connection and, in order to be thorough, I am recommending that we seek the advice of a geneticist."

"_____ has been diagnosed with _____ and I would like to see if we can identify a possible cause for these difficulties. Therefore I am suggesting that _____ be seen by a geneticist."

"In order to address your question, I would like to refer to medical genetics."

"In order to best care for your child, I would like medical genetics to help me with the following question: _____?"

"I understand you are not concerned, but in order to provide the best care, I need _____ from medical genetics."

Identifying Patients with Suspected Genetic Conditions

Patient-friendly Genetics Resources

- [What is a Pediatric Geneticist?](#) provides information for families from healthychildren.org
- [Making Sense of Your Genes: A Guide to Genetic Counseling](#) provides a patient-friendly introduction to genetic counseling, guidance on how to prepare for an appointment, and additional details about genetic counseling specialties.
- [Informed Consent for Genetic Testing: Patient Education Information for use with the Michigan Model Consent Form for Genetic Testing](#)
- Direct-to-Consumer Testing information from the Federal Trade Commission

Step 3

Talk to Parents about an At-risk Family Health History or Genetic Referral

CONTINUED

Steps in a Typical Genetic Evaluation and What to Expect

Prior to the Visit

Many times the geneticist will want to review medical records in advance of the appointment, so it is helpful for the medical office to fax or send these in advance, or make copies for the patient to bring with them for the visit. It is especially important to have detailed birth history, results of imaging studies (especially brain CTs/MRIs), operative reports, details about growth including growth curves and developmental evaluations.

The genetic evaluation will often take an hour or more, so patients should be prepared for a lengthy initial visit. During the initial visit the entire history is taken, including a three generation FH. If there are specific features in multiple family members, then bringing family photos to the visit is very helpful.

During the Visit

A careful exam will also be performed looking for minor or subtle signs of a genetic condition.

Following the exam, a number of tests will likely be recommended. These may include blood work, urine tests, X-rays, and sometimes other evaluations such as an eye exam. Sometimes the patients will need to return to their primary care office to arrange for these referrals or testing.

After the Visit

Genetic tests may take some time to come back, so the family should anticipate a bit of a wait for results. If the test uncovers a change of significance, then the parents will need to revisit the geneticists' office to discuss the results and what this means for their child.

Plan your QI

What are we trying to accomplish? Our aim is:

• By the end of the next 3 months, our practice team will have a system in place to communicate with at least 95% of our patients/families with an at-risk FH and in need of a genetic referral.

How will we know that a change is an improvement? Our measures are:

• 95% of patients/families with an at-risk FH will have documentation that a genetic referral was communicated/made, when appropriate. We will also collect feedback from staff and patients/families about this process.

What changes can we make that will result in an improvement? Our ideas for change are:

• Conduct an education/training session with clinic staff/physicians about strategies/scripts for communicating with families about an at-risk FH and recommended genetic referral. We will test the use of suggested patient/family-friendly resources on this topic with the next patients found to be at-risk and in need of a genetics referral.

Identifying Patients with Suspected Genetic Conditions

Step 4 Develop Relationships with Genetic Professionals

It is important to establish relationships with genetic professionals in your referral network as you would with any other specialty. In many cases, PCPs can order first-line genetic testing. However, it is crucial to have some guidance from experts to ensure that the appropriate tests are being ordered and that patients who would benefit from a genetic evaluation are seen in a timely manner. As you become more comfortable with genetic services, you may find you need less help with navigating genetic tests, but certainly having a trusted source of information will help improve your skills and comfort.

If you're part of a large health system, there may be genetic professionals within your organization that you can work with. If not, there are a few ways to find and develop a relationship with a genetics professional.

Finding a Genetic Professional

- [Find a Genetic Counselor](#) from the National Society of Genetic Counselors, is a directory to assist PCPs in locating genetic counseling services. Genetic Counselors can be searched by state, city, counselor's name, institution, work setting, type of specialty, or zip code.
- [Clinical Services Search Engine](#) from the American College of Medical Genetics, enables users to search for a genetics center. Centers can be located by name, type of service provided, all clinics, or by state, city, or zip code.

A **genetic counselor** can help people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. They can assist the patient in understanding the way heredity contributes to genetic disease, and provide support and guidance for a diagnosis. This process integrates: (1) interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; (2) education about inheritance, testing management, prevention, resources, and research; and (3) counseling to promote informed choices and adaptation to the risk or condition.

A **clinical geneticist** is a physician who can provide both a medical evaluation and genetic counseling. Depending on their needs, families may see both a genetic counselor and a clinical geneticist.

Many PCPs find that working with a genetic counselor can be extremely beneficial. Most genetics centers employ genetic counselors, and they may be more available or accessible to a PCP who is seeking guidance around genetic testing and patient consults.

Most genetic counselors are happy to connect with PCPs in their referral network. Here are some strategies for building a successful relationship with a genetic counselor:

Questions to ask:

- What is the best way to contact you? (eg, telephone, email, or text message)
- When I contact you for advice, what patient information will you need?
- Ask genetic professionals in your referral network if they would like you to order first-line genetic tests, and for which indications

Other communication/relationship strategies to consider:

- Genetic counselors who are affiliated with an academic center, and not a lab, are often the best source of unbiased information about which genetic tests to order
- When requesting assistance, make it clear that you are either
 - 1) requesting an official patient consultation for a differential diagnosis
 - 2) are considering a certain genetic test and would like the counselor's opinion
- When requesting assistance, provide the case presentation and symptoms
- If the genetics center is comfortable with PCPs ordering genetic tests, establish processes or a plan for the genetic counselor or other professional to be available immediately to meet with the family after a positive test result

Identifying Patients with Suspected Genetic Conditions

Working with Genetic Counselors Can Improve Patient Care

"My role as a GC typically consists of counseling patients, taking family histories and drawing pedigrees, and doing the many unspoken tasks of a genetic counselor (calling out abnormal newborn screens, writing letters, talking to insurance companies).

During the quality improvement project, I was a mentor to a privately-owned primary care practice who was already in my referral network. One of the practice's goals was to connect with their local genetic specialists and improve referral patterns. The team had lots of questions along the way but were able to streamline the process. They first worked on collecting family history to help identify red flags, and then they contacted me with questions regarding referrals they were considering. While the practice was already a good referral source for our center, the fact that they were better informed regarding appropriate use of genetic services helped. It showed me how building relationships as they had could help with our other practices in the area and our referral pattern with them."

-Cecilia Rajakaruna, MS, CGC, Mentor from the Region 4 Newborn Screening Collaborative to a practice from the GPCI QIP

Step 4

Develop Relationships with Genetic Professionals

CONTINUED

Tools & Resources

- [Pediatrics & Clinical Genetics](#) from the National Society of Genetic Counselors, provides a provider-focused overview of the role of a genetic counselor specializing in pediatric and clinical genetics and the benefits they can provide to patients. Also included is a list of conditions for which a referral would be appropriate and tips on communicating with your patients regarding what will happen during the evaluation.
- [Genetic Counseling in Primary Care](#), a webinar from the GPCI, provides an overview of the key competencies and skills in practice of genetic counseling, genetic counseling skills used for the risk assessment, evaluation, and management of patients in the primary care setting, and opportunities for collaboration with genetic counselors for improved patient care.
- [Newborn Screening and Genetics Collaboratives](#) provide newborn screening and genetics services, tools, and resources to both providers and families.

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the month, our practice team will meet with the genetic counselor/mentor in our Regional Genetics Collaborative, as part of a scheduled "lunch and learn" session in our office.

How will we know that a change is an improvement? Our measures are:

- Documentation of the meeting held with our genetic counselor/mentor in our Regional Genetics Collaborative. (including meeting agenda, meeting notes and list of attendees). Pre- and Post-tests will be conducted. Feedback will be collected from staff who attend the session.

What changes can we make that will result in an improvement? Our ideas for change are:

- Our physician champion will outreach a genetic counselor/mentor in our Regional Genetics Collaborative and schedule a staff education "lunch and learn" session in our office to discuss genetics in primary care and how to build a relationship with genetic professionals.

Identifying Patients with Suspected Genetic Conditions

Step 5 Order Appropriate Genetic Tests

Genetic testing is used for screening and diagnostic purposes. It has improved tremendously over the last few years and genetic tests are now available for over 2,200 diseases (both rare and common).

The newborn screening is the most common genetic test. It is performed on all babies in the US to screen for disorders. Prenatal screening is another kind of genetic testing. It is performed during pregnancy to determine the risk of possible health issues such as a chromosomal abnormality or birth defect in the developing fetus.

Newborn Screening

- Newborn Screening is testing performed in the first few days of a newborn's life to assess for disorders that might otherwise be missed due to the baby's normal appearance. By law, all states require newborn screening for at least 26 conditions, and some require screening for up to 50 conditions.
- While newborn screening was beyond the scope of the GPCI Quality Improvement Initiative, it is important for pediatric primary care providers to be aware of what to do with a positive newborn screen as well as tracking newborn screening results and having a plan for repeat screens in the event of a false negative. The following resources may be helpful for you to review:
 - [Baby's First Test](#)
 - [Newborn Screening ACT Sheets and Confirmatory Algorithms](#)
 - Your Regional Genetic and Newborn Screening Collaborative, as well as your state's newborn screening laboratory
 - AAP EQIPP Course "[Newborn Screening: Evaluate and Improve Your Practice](#)"



Identifying Patients with Suspected Genetic Conditions

Genetic testing can also be helpful for diagnostic or predictive testing. If you have a patient who might benefit from genetic testing, consider the following testing options. Many pediatricians should feel comfortable ordering a chromosome microarray after working with a local genetics professional to identify a reliable laboratory to which samples can be sent. Single gene sequencing, gene panels and whole genome/exome sequencing is best ordered by, or under the direction of, a geneticist. As discussed in the previous section, maintaining open communication and a working relationship with genetic professionals in your referral base will be invaluable to your practice.

Disclosures: Consider consulting a genetic professional before ordering genetic tests with which you are less familiar. Costs of testing may vary, and on occasion, a letter of medical necessity must be written to obtain insurance approval for ordering the test. Genetic professionals are familiar with writing such letters and may be able to assist you.

- **Chromosome microarray** is used to study number of copies of all genes all at once at much higher resolution than a karyotype and is therefore a **first line test before a karyotype or FISH in some instances**. It is useful for detecting microdeletions/microduplications and specific breakpoints in an unbalanced translocation. It's commonly used to identify etiologies of patients with multiple congenital anomalies, dysmorphic features, autism, developmental delay, and failure to thrive. It only detects segments of deleted/duplicated DNA and provides resolution down to ~ 50,000 base pairs, not to the individual's nucleotides.
- **Karyotype** is used to identify and evaluate the size, shape, and number of chromosomes. It can be used to identify trisomies like Down syndrome, sex chromosome anomalies, visible deletions/duplications, and translocations. It's not effective at clearly defining breakpoints in a chromosome with a deletion or translocation. However, it is the best method to identify a balanced translocation or inversion.
- **Fluorescence in situ hybridization (FISH) Analysis** is a laboratory technique for detecting and locating a specific DNA sequence on a chromosome. It is commonly used to detect specific syndromic deletions such as 22q11 deletion (DiGeorge syndrome) and Williams syndrome.
- **Single gene sequencing** is used to read out the DNA sequence for a specific gene such as the gene for neurofibromatosis 1 when that diagnosis is suspected. This is a good test when there is only one gene for the condition and when that condition is suspected in the patient or the family.
- **Panels of genes for a single clinical indication.** Some diseases are caused by more than one gene so to effectively evaluate the condition, you need to sequence several genes (e.g. cardiomyopathy or epilepsy). Panels of well-defined genes associated with these diseases have been developed to analyze several known genes for the disease.
- **Whole Genome/Exome Sequencing** sequences only the protein-coding regions (exons) of nearly all genes simultaneously. The current limitation of this process is the expense, but this is expected to decrease over time. This test is most useful when the clinical diagnosis is not obvious, when there are many genes for the condition (e.g., intellectual disability), or when genetic testing is not otherwise available for the condition by any other means.

Identifying Patients with Suspected Genetic Conditions

Step 5

Order Appropriate Genetic Tests

Uses of Genetic Tests

	Microarray	Karyotype	FISH Image	DNA Sequencing
Detects large deletions or duplication	X	X	X	
Detects deletions or duplications in part of a chromosome	X		X	
Detects small deletions or duplications	X			
Detects translocations		X		
Detects sequence changes and single gene mutations				X

Due to advances in genetic testing, you may want to consider retesting patients with suspected genetic conditions who previously tested negative or had inconclusive genetic test results.

Cost and Insurance Coverage

While some genetic tests can be expensive, there are other less expensive options. For example, a karyotype or routine chromosome analysis can cost between \$650 to \$800, and a plasma amino acid test can cost about \$270.

Private insurance and Medicaid coverage of more customary genetic tests has improved. A letter of medical necessity may be necessary for some tests. For patients without insurance coverage, some labs will offer a lower price. Genetic testing laboratories are familiar with this and can also assist with obtaining pre-authorization.



Identifying Patients with Suspected Genetic Conditions

Step 5

Order Appropriate Genetic Tests

CONTINUED

Case Examples: Genetic Testing

Testing for specific or common mutations

A baby was diagnosed with meconium ileus, a sign of Cystic Fibrosis (CF), at birth. Sweat chloride testing was positive for CF. The couple has had carrier screening for CF during pregnancy and the mother had been found to have a common CF mutation; however, the father's screening test was negative. The newborn screening test was also positive for CF. However, reflex testing only revealed the one mutation found in the mother. Because standard newborn screening programs use a panel of the most common mutations as a reflex test, the baby had sequencing done of the whole CF gene and was found to have a second rare mutation that was not included in the prior testing panels. The father was subsequently found to carry the rare mutation.

Chromosome testing

A 12 month old in your practice was born with tetralogy of Fallot and is now not gaining weight or reaching his developmental milestones despite a successful surgical repair of his congenital heart disease and good cardiac function. You order a chromosome microarray, and he is found to have a 300 kb duplication of 1q21 containing 2 genes which the lab classifies as a variant of uncertain clinical significance. The lab recommends testing the parents, and you find that the healthy father carries the same 1q21 duplication. Because the healthy father carries the same duplication and because the duplication is relatively small and contains few genes, it is likely that this duplication is not pathogenic and that this is a normal genetic variant.

Sudden death

The father of three children in your practice dies suddenly and unexpectedly in his sleep. His autopsy determines that he had hypertrophic cardiomyopathy. Because this is an autosomal dominantly inherited condition, each of the children are at 50% risk to inherit this genetic condition. You arrange for the medical examiner to perform genetic testing on the father because genetic testing is always best initially performed on a symptomatic family member when you want to offer predictive testing to other asymptomatic family members. The father's test shows a MYBPC3 mutation. Genetic testing in only one of the three children is positive for the father's MYBPC3. The two mutation negative children do not require longitudinal echocardiograms. The one mutation positive child requires serial echocardiograms to monitor for hypertrophic cardiomyopathy.

Section 3

Identifying Patients with Suspected Genetic Conditions

Step 5

Order Appropriate Genetic Tests CONTINUED

Tools & Resources

- [Genomic Testing](#) provides information and resources from the Centers for Disease Control and Prevention.
- [Frequently Asked Questions about Genetic Testing](#) provides information from the National Human Genome Research Institute (NHGRI).
- [Genetic Disorders, Genomics, and Healthcare](#) provides information about genetic disorders, genetic testing, genetic counseling and evaluation, and how to locate a genetics professional from the NHGRI.
- [The Genomics in Medicine Lecture Series](#) is a recorded lectures from the NHGRI to enhance health-care professionals' understanding of the intersection between genomics and medicine.
- [GeneReviews](#) are expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.
- [Genetic Testing Registry](#) provides a central location for voluntary submission of genetic test information by providers. Information is provided on the test's purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts and credentials.
- [Improving Newborn Preventive Services at the Birth Hospitalization: A Collaborative, Hospital-Based Quality-Improvement Project](#) (article)
- [Overview of Genetic Testing and Screening](#) is a webinar from the GPCI covering the difference between diagnostic and screening tests and their appropriate use in pediatrics, several types of diagnostic genetic testing and their limits and applications, and how to access current information and resources for genetic testing.
- [Ordering the Right Tests – Genetics in Primary Care](#) is a webinar from the GPCI covering the different types of genetic testing and advances in testing technology, the role of genetic testing in primary care, and the ethical principles involved in genetic testing in pediatrics.
- [Genetic Testing in Primary Care](#) is a webinar from the GPCI covering the two basic categories of genetic variation/mutation, the types of genetic testing used to identify these variants, the limitations of each methodology.
- [Insurance Coverage Toolkit for Chromosomal Microarray Testing](#), developed by the International Standards for Cytogenetic Arrays Consortium to assist providers in advocating for insurance coverage and reimbursement on CMA tests.

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 3 months, our clinical team will be more knowledgeable about genetic testing and report being more comfortable and confident about ordering the right genetic tests.

How will we know that a change is an improvement? Our measures are:

- 90% of our patients identified at-risk and in need of testing/screening will have documentation in their record that the appropriate genetic tests/screens were ordered. Feedback will be collected from clinical staff and families.

What changes can we make that will result in an improvement? Our ideas for change are:

- All appropriate clinical staff will watch the recommended GPCI webinars on the topic of Genetic Testing/Screening. Documentation of consultation(s) with local genetic professional(s) about appropriate testing/screening, reliable labs and costs will be noted in the patient's record.

Section 4

Providing Appropriate Care for Patients with Genetic Conditions



Care and tracking

Providing Appropriate Care for Patients with Genetic Conditions

Once a patient is diagnosed with a genetic condition, you'll want to provide the appropriate follow-up and additional services, so the patient receives the best possible care.

Much of the information included in this section is consistent with the patient-centered medical home model of care. Medical home is an approach to providing comprehensive primary care that facilitates partnership between patients, physicians, and families. The American Academy of Pediatrics (AAP) believes that every child deserves a medical home, where care is accessible, continuous, comprehensive, patient- and family-centered, coordinated, compassionate, and culturally effective.

While the information in this toolkit is focused on patients with genetic conditions, implementing strategies based on the medical home model will improve care for all your patients. If you're trying to achieve patient-centered medical home recognition, many of the suggestions included here align with the requirements and can help you achieve patient-centered medical home recognition. While the following sections provide steps you can take to care for patients with genetic conditions, the following Web sites provide additional information, tools, and resources for child health and medical home.

- [Bright Futures](#) is a national health promotion and disease prevention initiative through the AAP. Bright Futures offers many different resources, including education, family resources, and clinical practice materials for your use.
- [Medical Home Portal](#) provides information about genetic conditions, family-centered care, health supervision guidelines, transitions, and more for both PCPs and families.
- [The National Center for Medical Home Implementation](#) (NCMHI), a cooperative agreement between the Maternal and Child Health Bureau and the AAP, works to ensure that all children have access to a medical home. Through the [NCMHI Web site](#), you can create a free account and access tools to help you implement many of the medical home components discussed in this section of the toolkit. You'll find family surveys, focus group information, and more.



Providing Appropriate Care for Patients with Genetic Conditions

Step 1 Create a Patient Registry

Registries vary in format, from simple Excel spreadsheets to EHRs. The use of a patient registry can help primary care providers effectively manage a patient population as part of a successful medical home, especially in the management of chronic conditions.

To develop and maintain a registry to identify and manage children with genetic conditions in your practice:

- Obtain a list of patients with genetic conditions to include in your patient registry. You can use a list of ICD-9 codes to select the patients.
- Decide if you will store the data in your EHR, in a database, or on a spreadsheet. Structured data entry can prompt for completeness and provide better search and retrieval capabilities and reporting features.
- Identify someone in your practice to help build and maintain the registry. This person could be a care coordinator or other appropriate staff member in your practice.
- Develop a registry that meets your practice's needs with simple or more extensive fields. If using a spreadsheet or database to track patient data, consider adding some conditional formatting that flags when patients need certain laboratory tests and other preventive services.
- Build a genetic patient registry with your EHR or use the electronic Excel template (see the *Using a Simple Patient Registry to Improve Your Chronic Disease Care* article for a template).
- Identify the structured data that should be included within the patient registry. According to 2011 National Committee on Quality Assurance (NCQA) Patient-centered Medical Home (PCMH) Standards, your registry should be able to record the following as structured data:
 - Date of birth
 - Gender
 - Race
 - Ethnicity
 - Preferred language
 - Contact information (telephone numbers, email address)
 - Dates of previous clinical visits
 - Legal guardian/health care proxy
 - Primary caregiver
 - Health insurance information
 - An up-to-date problem list with current and active diagnoses
 - Allergies, including medication allergies and adverse reactions
 - Blood pressure, with the date of update
 - Height
 - Weight
 - Length/height, weight and head circumference (less than 2 years of age) and BMI percentile (2–20 years) of pediatric patients, with the capability to plot changes over time
 - Status of tobacco use for patients 13 years and older
 - List of prescription medications with the date of updates
- Continually add to your registry as patients are diagnosed with genetic conditions and remove those who are no longer your patients.
- Develop a process to identify high-risk or complex patients, such as a complexity of care score system, to assist you in understanding the level of need of patients. The benefit of assigning a complexity score is that it allows a practice to not only quantify but qualify their population and to plan for care coordination needs and staffing. Below is a sample complexity of care scoring system, developed by Jennifer Lail, MD, FAAP for Chapel Hill Pediatrics and Adolescents, PA.

Section 4

Providing Appropriate Care for Patients with Genetic Conditions

Step 1

Create a Patient Registry

CONTINUED

Name:	Race:	
Sex: Birthdate:	Patient ID:	
Insurer:	Primary care provider:	
Diagnoses: 1)	2)	
3)	4)	
CSYCN Complexity Rating	Description	Examples
1	1 chronic condition, well-controlled OR Significant PMH, quiescent or resolved	Asthma, mild per Repaired VSD
2	1 evolving chronic condition, unstable OR 2 chronic conditions, both well-controlled	Asthma PCOS + Type 2 DM Asthma +ADHD
3	2 or more chronic conditions, with either unstable	GERD Asthma w/ER visit
4	Any tech. dependent patient Mod/sever cognitive delays	Wheelchair, walker, GT, Trach MR, Autism, Group Home res.
+1	Language barrier	Non-English speaker
+1	Behavioral disorder	OCD, Anxiety in addition to above
+1	Family/Social complications	Divorce, Horizons
Total complexity score		

Tools & Resources

- [Using a Simple Patient Registry to Improve Your Chronic Disease Care \(article\)](#)
- [Newborn Screening ACT Sheets and Confirmatory Algorithms](#) from the American College of Medical Genetics provides step-by-step instructions for treating patients with positive newborn screening markers.
- [Clinical Genetic Evaluation of the Child with Mental Retardation or Developmental Delays \(AAP policy/clinical report\)](#)
- [Genetic and Rare Conditions Site](#) for lay advocacy and support groups, information on genetic conditions/birth defects, from the University of Kansas Medical Center.

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next quarter, our practice will utilize an Excel-templated patient registry which will include our current patients with genetic conditions, using ICD-9 codes as our selection criteria.

How will we know that a change is an improvement? Our measures are:

- 100% of our patients with diagnosed genetic conditions will be included in our registry, scored for complexity and followed up by our care coordinator for preventive services, other needed referrals and tests. Feedback will be collected from our care coordinator, clinicians and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- A list of appropriate ICD-9 codes and structured data to be tracked will be agreed upon by our physicians/clinicians (task). Our care coordinator will create and maintain our registry (as part of her role/job description) and will follow up with specialists, labs and families, as appropriate.

Providing Appropriate Care for Patients with Genetic Conditions

Step 2 Implement Systems to Improve Genetic Services

Creating systems of care that enable you to track, follow-up, and document all results and activities for patients with genetic conditions, will help you better care for these patients. It's also important to communicate results and their implications to patients and families. Once you've discussed the results or diagnosis with the family, make sure to document the follow-up plan in patient's chart and that the follow-up/plan of care was discussed with the family.

Establish a tracking mechanism for future orders, referrals, and follow-ups.

- Use future orders in an EHR as a tracking mechanism.
- Track lab orders for genetic tests.
- Put a note at the next well visit in the EHR to trigger follow-up discussion.

Implement a process to identify and contact patients who are not current for age-appropriate preventive services (reminder-recall system).

Discuss possible registry functionality within your EHR system with your information technology department or your EHR vendor representative.

Improve coding and reimbursement procedures for genetic services.

- Create a business plan for care coordination, including support for the role of care coordinator (if feasible).
- Consider how you will work with payers to finance care coordination through fee-for-service enhancements, per member per month administrative payments, or the use of care plan oversight billing codes.
- Advocate to third-party payers for adequate payment and reimbursement of genetic services.

The tracking system does not have to be complex. One practice participating in the quality improvement (QI) project developed a basic system where the practice faxes the patient information to the genetics professional. The genetics professional calls the practice back to say if they think it merits their looking at the patient. The nurse who faxes the information, keeps the fax until she gets an answer, then calls the patient with the appointment information. The appointment is placed on the calendar, and the practice calls the patient two days after the scheduled appointment to see if they made it.

Another practice uses their EHR to track referrals. They found it helps to have a staff member assigned to track referrals and follow up with families or specialists as appropriate. While developing the process was easy, a dedicated team and strong leader is needed to make it successful. The process doesn't work if the team doesn't follow though.

Providing Appropriate Care for Patients with Genetic Conditions

Step 2

Implement Systems to Improve Genetic Services

CONTINUED

Tools & Resources

- [Recalls and Reminders System](#) from Bright Futures, provides information and resources to improve your office systems.
- [Building Your Medical Home and Getting Paid Appropriately](#) from NCMHI, explains the different kinds of payment within the medical home
- [Medical Home Coding Fact Sheet](#) from the AAP contains an index of CPT codes for medical home.
- [Genetic Coding Fact Sheet](#) from the Genetics in Primary Care Institute (GPCI) contains case studies, Current Procedural Terminology, and International Classification of Diseases, 9th Revision, Clinical Modification codes will assist you in coding to the highest specificity possible.
- [Genetics and Coding: What the Primary Care Provider Needs to Know](#) is a webinar from the GPCI covering the purpose of the ICD coding system, the gaps within the current coding systems regarding genetics in primary care settings, and appropriate courses of action related to accurate coding of genetic services.
- [Insurance Coverage Toolkit for Chromosomal Microarray Testing](#), developed by the International Standards for Cytogenetic Arrays Consortium to assist providers in advocating for insurance coverage and reimbursement on CMA tests.



Providing Appropriate Care for Patients with Genetic Conditions

Step 3 Follow the Health Supervision Guidelines

Thus far, the AAP Committee on Genetics has developed guidelines for the health supervision of patients with the following genetic conditions:

- [Achondroplasia](#)
- [Down Syndrome](#)
- [Fragile X](#)
- [Marfan Syndrome](#)
- [Neurofibromatosis](#)
- [Prader-Willi Syndrome](#)
- [Sickle Cell Disease](#)
- [Turner Syndrome](#)
- [Williams Syndrome](#)

For patients with other genetic conditions, you can look for resources through the AAP, the American College of Medical Genetics, the Genetic Alliance, the National Institute of Health, and disease-specific advocacy groups.

You can also use the [Bright Futures / AAP Periodicity Schedule](#), health supervision guidelines for pediatric primary care.

Plan your QI

What are we trying to accomplish? Our aim is:

- *By the end of this month, we will use the available guidelines for the health supervision of 100% of our patients with certain identified genetic conditions.*

How will we know that a change is an improvement? Our measures are:

- *Identified HSV guidelines will be made available for 100% of patients with genetic conditions seen during the review month. HSV guidelines will be reviewed and discussed with 95% of those patients/families. Feedback will be collected from staff, physicians and patients/families.*

What changes can we make that will result in an improvement? Our ideas for change are:

- *Identify available health supervision guidelines (task). Conduct education/training session for physicians and staff on the topic. For a 1 week period, as part of our daily huddle, we will identify those patients being seen each day for whom HSV guidelines are available, and put a note/prompt in the patient's record. The nurse will provide a copy of the appropriate guidelines for the physician when rooming the patient/family. The physician will review and discuss the guidelines with patient/family. Simultaneously, our care coordinator will make reminder phone calls to patients with genetic conditions due/overdue for a HSV who do not have a scheduled appointment.*

Providing Appropriate Care for Patients with Genetic Conditions

Use a parent advisory panel to vet certain genetics-related documents and process, and tell you about the best resources to include as community-based resources/supports in your medical neighborhood.

- Create a community resources fact sheet for your practice and for parents and caregivers.
- Connect with public and private health care providers in your “medical neighborhood.” Meet to discuss services offered that may benefit your patients/families.
- Refer your patients to their state’s United Way 2-1-1 hotline. This free, 24-hour information and referral helpline is assigned by the Federal Communications Commission to streamline access to health and human services. While services vary by community, 2-1-1 can provide families access to physical and mental health resources (children’s health insurance programs), support for children (childcare, Head Start, family resource programs), and more.
- Develop a protocol in your practice that designates a representative of your practice team, like a care coordinator, to review key educational messages with patients and families at every visit.
- Provide information on parent support groups for specific medical conditions or for parents of children with special health care needs.

Step 4 | Provide Family-centered Care

The Maternal and Child Health Bureau defines family-centered care as a respectful family/professional partnership that honors the strengths, cultures, traditions, and expertise that everyone brings to the relationship. Family-centered care is the standard of practice which results in high quality services.

Community Resources

Become aware of community-based resources in your area; provide information for families about how to access these local or national services and supports. Invite community-based resources to come to your practice and learn about the programs and services that they offer.

Patient and Family Satisfaction Survey

Consider developing and implementing a patient and family satisfaction survey yourself (addressing access, communication, coordination, and whole-person care), or use an existing survey to measure and assess the “medical homeness” of a primary care practice from the family perspective.

- Conduct informal or formal discussions or interviews with families to brainstorm ideas for opening the lines of communication.
- Establish focus groups for feedback and identification of specific needs to be improved within the practice.
- Clearly post practice procedures for addressing patients’ and families’ recommendations, complaints, and questions.
- Add a suggestion box in the office waiting room.
- Establish a practice advisory group that includes family membership.
- Establish a process to elicit patient and family concerns at every visit. For example, put prompts on the intake form or hang a poster in the examination room that urges patients to express their concerns.

Develop a post-visit process that queries patients and families regarding whether their concerns were addressed at this visit or if plans were made to address them. Or, verbally check for understanding and satisfaction about the plan made to address them and document response(s).

Section 4

Providing Appropriate Care for Patients with Genetic Conditions

Step 4

Provide Family-centered Care

CONTINUED

Tools & Resources

- [Patient-Centered Medical Home Version of the Consumer Assessment of Healthcare Providers and Systems \(CAHPS\) Clinician Group Survey](#)
- [The Medical Home Family Index and Survey](#)
- [Positioning the Patient and Family at the Center: A Guide to Family and Patient Partnership in the Medical Home](#)
- [Family Voices Family-Centered Care Self-Assessment Tool for providers](#)
- [Family-Centered Care Self-Assessment Tool for families](#)

Cultural and Linguistic Changes

Take a systematic assessment of your organization's cultural and linguistic attitudes, practices, structures, and policies using the [Cultural Competence Health Practitioner Assessment](#). Use the results of the assessment to plan for and incorporate cultural and linguistic changes in your practice.

Plan your QI

What are we trying to accomplish? Our aim is:

- *By the end of the next month, we will query at least 95% of our patients at the end of their visit to assess if all of their concerns were addressed or plans made to address them.*

How will we know that a change is an improvement? Our measures are:

- *Documentation that 90% of the patients/families were queried to assess if their concerns were addressed or plans made to address concerns and 95% of those assessed will indicate "yes." Feedback will also be collected from clinicians and families about the poster and the assessment process.*

What changes can we make that will result in an improvement? Our ideas for change are:

- *A poster about patient/family-centered care will be designed by the QI team and hung in 1 exam room as a prompt for clinicians and families to ask about concerns. At the end of the visit, the patient/family will be asked by the clinician if their concerns were addressed (or plans made) and the response will be documented in the visit note.*

Section 5

Coordinating Care for Patients with Genetic Conditions



Make a plan

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 1 Improve Processes to Co-manage Care with Specialists

Increasing numbers of medically complex children require an increasing degree of complex care. Co-management can provide increased efficiency and quality of care for patients with genetic conditions.

Co-management is the process of shared, delegated care of a patient's medical conditions among providers with either similar or disparate clinical expertise and/or professional credentials.

While a co-management approach can be used in almost any care situation, including between two primary care providers (PCPs) who work in the same office and see the same patients, co-management should be considered for:

- Complex or multisystem medical issues
- Conditions requiring providers with specific technical skills
- Conditions requiring specialty teams, such as metabolic diseases
- Rare conditions
- Patients and families with many questions about diagnosis and care
- Any case where patient, family, PCP, or specialist feels co-management would benefit care

A few things to consider when coordinating care among multiple providers:

- Many patients with genetic conditions are cared for by multiple other pediatric specialists, each of whom should have defined patient care responsibilities
- All members of the co-management team must be clearly identified for family and providers
- It is best to establish a relationship with specialty providers prior to referral
- Multidisciplinary clinic care may often be preferable due to better coordination of care, proximity, communication, like-mindedness, and more

Is Co-management the Right Fit?

Co-management offers many benefits, but also can add complexity to patient management if it's not the right fit. A few determinants include:

- Geography
- Clinic systems and insurance limitations
- Specialist availability/desired involvement
- Frequency of need for specialty visits
- Patient/family buy-in to team approach
- Family barriers (ie, finances, time, etc)

Work with Genetics

While geneticists' time can be quite limited, their expertise in patient care, ability to discuss and address rare or complex issues, and access to resources make them a valuable member to a co-management team. Benefits include:

- Expertise in areas such as diagnosis, inheritance pattern and recurrence risk, mechanism of disease, evolution of clinical manifestations with age, and syndrome management
- Access to patient information, support resources, and additional genetics expertise
- Ability to share current information with families about issues such as testing and biobanking
- Experience and comfort with rare conditions, end-of-life issues, and post-mortem investigations

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 1

Improve Processes to Co-manage Care with Specialists

CONTINUED

Develop a Care Plan

A care plan can be an effective tool for sharing information among providers and defining each provider's responsibility in the patient's care. A care plan should contain a comprehensive record of the patient's medical, social, and school history as well as action related to how that patient's care will be managed and by whom.

- Develop a protocol/written process for co-managing the care of patients with a genetic condition in your practice among the patient/family, pediatric medical home, and specialists. Consider developing and implementing the following tools with specialists in your area:
 - Co-management letter and agreement with specialists that specify the responsibilities of the medical home and those of the specialist.
 - Fax-back form for expedited communication with specialists when requesting information.
 - Referral forms to accompany the patient/family on the visit with the subspecialist that clarify the purpose of the referral, the questions to be answered, and the degree to which you want the specialist's help (eg, assume care of the problem, provide initial diagnosis and recommendations, share care according to a specific plan).
 - Comprehensive care plans to share information that can travel back and forth between primary care and specialty visits with updates added following each visit.
 - Standard format for referral letters that provide a brief history, state the question or concern, and specify your and the patient's preference for ongoing care/co-management.
- Obtain information for local or regional genetic professionals (see the *Develop Relationships with Genetic Professionals* section).
- Establish a mechanism to track specialist referrals (see the Implement Systems to Improve Genetic Services section).
- Document a problem list and identify who manages each aspect of care (specialist or primary care).

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 6 months, our practice will implement a co-management letter and agreement to use with specialists who share delegated care of our patients with diagnosed genetic conditions.

How will we know that a change is an improvement? Our measures are:

- We will generate a letter and have a signed agreement for 90% of our patients with genetic conditions seen by their specialist during each month of this 6 month period. We will also collect feedback from primary care and specialty staff and physicians and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Use our registry to identify patients referred or due to be seen for specialty care during each month (task). Create a letter to test using with 1 or 2 of the specialists who will see our patients with genetic conditions. The agreement will indicate information we will make available and expectations of updated information we need to receive (and by when).

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 2 Obtain Current Emergency Plans (if applicable)

An emergency plan is a comprehensive set of instructions for families that outlines when, where, who, and how to contact emergency personnel should their child need emergent care. This plan also provides critical information for emergency providers. The plan should list the child's diagnoses with base-line findings, current medications, allergies, immunization status, common presenting problems/findings with specific suggested management strategies, and contact information for the child's subspecialty providers.

- Establish processes to obtain emergency plans from specialists.
- Modify and use example letters to specialists to obtain emergency management protocols for patients in your practice that require an emergency plan for management of genetic conditions.
- Obtain and update emergency plans for your patients with a genetic condition that require a plan, as indicated in the Genetic Patient Registry ICD-9 list.

Sample/Template Emergency Plans

- [New England Consortium](#)
- [British Inherited Metabolic Disease Group](#)

Genetic conditions that should have an emergency plan (All conditions are Endocrine/Metabolic conditions)

ICD-9 Code	Condition
270.2	Tyrosinemia
270.3	Disturbances of branched-chain amino-acid metabolism (MSUD, PA, MMA, IVA, 3-MCC)
270.3	MSUD
270.6	Disorders of urea cycle metabolism (Urea cycle defects: NAGS, OTC, CPS, Citrullinemia, ASA Lyase deficiency, Arginase deficiency)
270.7	GA-1
271	Glycogen storage diseases (Types 0, I-VII, IX, XI-XIII)
277.1	Disorders of porphyrin metabolism (Porphyria)
277.81	Primary carnitine deficiency
277.85	Disorders of acid oxidation (VLCADD, LCHADD, MCADD, SCADD, MTFP, GA-II A/B/C, CPT1/2)
277.86	Peroxisomal disorders
277.87	Mitochondrial disorders
	Organic Acid Disorders (Isovaleric academia, glutaric aciduria type 1, 3-hydroxy 3-methylglutaric aciduria, multiple carboxylase deficiency, methylmalonic academia, 3-methyl-crotonyl-CoA carboxylase deficiency, methylmalonic academia, propionic academia, -Ketothiolase deficiency, 2-Methylbutyryl-CoA dehydrogenase deficiency, 2-Methyl-3-hydroxybutyric aciduria, 3-Methyl-glutaconic aciduria, methylmalonic academia, isobutyryl-CoA dehydrogenase deficiency, malonic aciduria)

Emergency plans should be considered for other, non-genetic complex conditions, such as asthma, diabetes dependent on insulin, seizures, and anyone with a tube (GT, Trach, etc). Models for working with specialists to ensure appropriate information is in place are the same across specialties. The guidelines provided here for patients with genetic conditions could be replicated for other specialties.

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 2

Obtain Current Emergency Plans (if applicable)

CONTINUED

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 3 months, our practice team will obtain all emergency plans, as appropriate, from the specialists co-managing our patients with genetic conditions.

How will we know that a change is an improvement? Our measures are:

- Obtain emergency plans, when appropriate, for 95% of our patients with certain genetic conditions who are co-managed by specialists. We will also collect feedback from our care coordinator, specialists and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Our care coordinator will use our registry of patients with genetic conditions to identify and document patients who should have an emergency plan. Our team will draft a letter to send to 1 or 2 specialists, as a test, asking for a fax or electronic copy within 7 days of the most recent emergency plan created/maintained by the specialist.

Advice From a Private Primary Care Practice:

If we have a patient with a specific genetic condition that requires an emergency plan like Maple Syrup Urine Disease, that emergency plan does come from the specialist, but we rely on the family to help us get that information which in turn encourages the family to understand the plan better and anticipate the needs of the child. We do not keep the form in the patient's chart at this time, but we are looking at incorporating that into the chart in the future. This form has been successful in creating opportunities for conversation with patients to help discuss emergency situations and openly address the possible needs a child may have in times of crisis. I would advise other practices to make the emergency plans as patient-friendly and as flexible as possible. Remind the families about them because they will forget that they exist and help keep them updated when at all possible.

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 3 Obtain Current Emergency Letters (if applicable)

An emergency letter is a document stating the patients' condition and any instructions regarding the proper care for the patient under specific circumstances or in an emergency. This letter should contain contact information for the geneticist or specialist, as well as the PCP, and anyone who should be contacted if the patient becomes ill. Families should have a copy of their emergency letter on hand, and a copy should be given to the child's school, other care givers, etc.

- Review the list of suggested ICD-9 codes and conditions for these patients with a genetic condition.

This list should not be considered mutually exclusive, nor an exhaustive list of conditions and reasons for which an emergency letter is warranted. This is a decision that should be made in conjunction with the specialists involved in the patient's care.
- Review a sample emergency letter for patients with Maple Syrup Urine Disease.
- Communicate with the specialist to obtain emergency letters for your patients with a genetic condition that may benefit from such a letter (for specific manifestations or acute emergency situations, as necessary). These letters are ideally created as part of co-management between the specialist and the PCP.



Section 5

Coordinating Care for Patients with Genetic Conditions

Step 3

Obtain Current Emergency Letters (if applicable)

CONTINUED

Genetic conditions that should have an emergency letter

ICD-9 Code	Condition	Category
279.11	Velocardiofacial/DiGeorge	Syndromes
282.2	Glutathione-caused anemias (e.g., G6PD deficiency)	Endocrine/Metabolic
282.4	Thalassemias	Other/Miscellaneous
282.6	Sickle Cell Disease	Hematologic
345.9	Seizure disorder	
348.3	Epilepsy (348.3)	Neurologic
348.3	Encephalopathy, unspec	Neurologic
359.1	Duchenne/Becker	Neuromuscular
359.1	Hereditary muscular dystrophy	Neuromuscular
359.21	Myotonic dystrophy	Neuromuscular
359.22	Myotonia congenital	Neuromuscular
359.23	Myotonic chondrodystrophy	Neuromuscular
359.71	Hereditary inclusion body myopathy	Neuromuscular
359.8	Myopathy, other	Neuromuscular
359.9	Myopathy NOS	Neuromuscular
448	Osler-Weber-Rendu	Syndromes
448	Hereditary hemorrhagic telangiectasia	Syndromes
710.9	Connective tissue disorder (chest pain management)	Skeletal dysplasias/connective tissue disorders
737.3	Scoliosis/kyphoscoliosis, idiopathic	Congenital anomalies
741	Arnold-Chiari malformation	Congenital anomalies
741.9	Spina bifida	Congenital anomalies
742.3	Dandy-Walker	Congenital anomalies
742.3	Hydrocephalus, congenital	Congenital anomalies
742.3	Dandy-Walker malformation	Congenital anomalies
745.4	Ventriculoseptal defect	Congenital anomalies
745.5	Atrioseptal defect, secundum	Congenital anomalies
756.51	Osteogenesis imperfecta (Blue sclera)	Skeletal dysplasias/connective tissue disorders
756.55	Ellis van Creveld syndrome	Skeletal dysplasias/connective tissue disorders
756.56	Multiple epiphyseal dysplasia	Skeletal dysplasias/connective tissue disorders
756.59	Leri-Weill	Syndromes
756.59	McCune-Albright	Syndromes
756.6	Diaphragmatic hernia, congenital	Congenital anomalies
756.89	Schwartz-Jampel	Syndromes
756.89	Nail-Patella syndrome	Syndromes
756.89	Connective tissue disorder (not EDS)	Congenital anomalies
757.39	Bloom	Syndromes
758.33	Autosomal microdeletion syndrome	Chromosome anomalies
758.89	MELAS	Endocrine/Metabolic
758.89	MERRF	Endocrine/Metabolic
758.9	Williams syndrome	Syndromes
759.6	Hamartoses, NEC	Harmatoses
759.82	Marfan syndrome	Skeletal dysplasias/connective tissue disorders
759.89	Angelman	Syndromes
759.89	Bardet-Biedl	Syndromes
759.89	Beckwith-Wiedemann	Growth disorders
759.89	Freeman-Sheldon	Syndromes
759.89	Russell-Silver	Syndromes
759.89	Smith-Lemli-Opitz	Syndromes
759.89	Other congenital syndrome	Syndromes
775.9	Newborn metabolic disturbance, NOS	Endocrine/Metabolic
	Mechanical ventilation	
	Tracheostomy	
	VP Shunt	
	Endocrine (congenital hypothyroidism, congenital adrenal hyperplasia)	
	Gastrostomy	

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 3

Obtain Current Emergency Letters (if applicable)

CONTINUED

Plan your QI

What are we trying to accomplish? Our aim is:

- By the end of the next 3 months, our practice team will obtain all emergency letters, as appropriate, from the specialists co-managing our patients with genetic conditions.

How will we know that a change is an improvement? Our measures are:

- Obtain emergency letters, when appropriate, for 95% of our patients with certain genetic conditions who are co-managed by specialists. We will also collect feedback from our care coordinator, specialists and patients/families

What changes can we make that will result in an improvement? Our ideas for change are:

- Our care coordinator will use our registry of patients with genetic conditions to identify and document patients who should have an emergency letter. Our team will draft a letter to send to 1 or 2 specialists, as a test, asking for a fax or electronic copy within 7 days of the most recent emergency letter created/maintained by the specialist.



Section 5

Coordinating Care for Patients with Genetic Conditions

Step 4 Plan for Transitions to Adult Care

Patients experience a number of transitions throughout their life, whether typical transitions like prenatal to birth, or from “normal” to “something’s wrong.” For pediatric patients, a critical transition is the transition from pediatric patient to adult patient. The goal for a successful transition is for the young adult, parent/guardian, and professionals to work together to help the patient realize his/her potential, satisfy his/her own needs, develop capacities needed to interact successfully with biological, physical, and social environments, and become a responsible medical consumer.

Develop a process/written protocol for your practice to discuss and document transition to adult care for patients with a genetic condition aged 12-21 at least annually.

Transition Policy Excerpt from a Practice in the Quality Improvement Project

The Transition Plan starts at age 12 years. You will receive a copy of our policy before his/her checkup. Read our policy with your child before his/her visit. The age 12 years is chosen because your child starts thinking about his/her own future.

Before your child's 14-15 year checkup your family receives an e message directing you to the Patient Portal. Have your child fill out the questionnaire prior to his/her checkup. The answers are used for us to work together on developing his/her transition plan. This plan is updated each year at the checkup CYSHCN have transitions and CCM plans before age 12 years.

Your 18 year old is an adult. Their medical home cares for him/her as an adult. Working together we decide to transfer care to adult practice between the ages of 18 and 21 yrs.

For the patient's eventual transition from adolescence to adulthood, the AAP recommends that a written health care transition plan is developed together with the youth and family and includes what services need to be provided, by whom, when, and how they will be financed.

The following are keys to managing transitions:

- Decide on your age (or other) cut-offs
- Ask genetics specialists about their scope of practice
- Ask genetics what management or follow-up is needed in adulthood
- Identify options for appropriate adult providers—genetics and others
- Look into adult care coordination
- Let your patients and families know well in advance about transitions
- Have a written plan for transitions
- Discuss quality-of-life and end-of-life issues openly, as indicated
- Document and track transition process
- Ask for feedback from patients and families

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 4

Plan for Transitions to Adult Care

CONTINUED

Tools & Resources

- [Supporting the Health Care Transition from Adolescence to Adulthood in the Medical Home \(clinical report\)](#)
- Got Transition works with pediatric and adult primary care practices in a learning collaborative model to develop a practical package of resources that align with the report's guidance.
- [Six Core Elements of Health Care Transition toolkit](#)
- [Family-friendly resources](#)
- [Models of Transitional Care for Young People with Complex Health Needs: A Scoping Review \(article\)](#)
- [Transitions to Adult Care ACT Sheets and Confirmatory Algorithms](#) are clinical decision support tools from the American College of Medical Genetics and Genomics

Plan your QI

What are we trying to accomplish? Our aim is:

- To begin at age 12 yrs. to document annual discussions about transition to adult care for all of our patients with a diagnosed genetic condition.

How will we know that a change is an improvement? Our measures are:

- 95% of our patients 12 yrs. old or above seen for a HSV in the prior month will have documentation in their record about age- and ability -appropriate transition care planning. Feedback will also be collected from our care coordinator, physicians and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Identify adult physicians in our community who are willing/able to become the medical homes for our patients diagnosed with genetic conditions transitioning to adult care (task). Our care coordinator will use our registry to identify patients 12 yrs. old and older with no documentation of an annual discussion about transition to adult care. She will add a note/prompt about transition in the patient's record for the next health supervision visit.

Section 5

Coordinating Care for Patients with Genetic Conditions

Step 5 Discuss Palliative Care (if applicable)

Frequently thought of as end of life care, this perspective is limited. Instead, palliative care can be thought of as the art and science of family-centered care aimed at enhancing quality of life and minimizing suffering, often provided to patients with a life-threatening or life-limiting illness. Care can be provided from the point of diagnosis onward by an interdisciplinary team. Care is focused on improving quality of life, through relief of symptoms and provision of psychological, social, and spiritual care for children and families living with life-threatening or terminal conditions.

Develop a process/written protocol to offer a palliative care discussion for patients with a genetic condition, as appropriate, on an annual basis.

One practice that participated in the quality improvement project said, “We used the project as an ‘excuse’ to broach the subject and then let the families talk to us about what they might want.” While this practice focused more on end-of-life care, they had a few kids with conditions such as muscular dystrophy, quadriplegic cerebral palsy, and metachromatic leukodystrophy where the focus of palliative care was on quality of life. Open ended discussions let the family share ways the care team could help them and things that could be better. Quality of life wasn’t limited to the patient; it included the whole family and things like respite and support services.



Section 5

Coordinating Care for Patients with Genetic Conditions

Step 2 Identify and Follow-up with Patients Identified at Risk

Tools & Resources

Articles

- [Palliative Care for Children](#) (policy statement)
- [Communicating With Children and Families: From Everyday Interactions to Skill in Conveying Distressing Information](#) (technical report)
- [Analysis of enacted difficult conversations in neonatal intensive care](#)
- [Difficult Conversations: Improving communication skills and relational abilities in health care](#)
- [Difficult Conversations in Health Care: Cultivating Relational Learning to Address the Hidden Curriculum](#)

Textbooks

- Carter BS, Levetown M, Friebert SE. Palliative Care for Infants, Children & Adolescents. 2nd ed. Baltimore, MD: The John Hopkins University Press; 2011:441-64
- Wolfe J, Hinds P, Sourkes B. Textbook of Interdisciplinary Pediatric Palliative Care. Philadelphia, PA: Saunders; 2011:402-7

Resources

- [Children's Hospice and Palliative Care Coalition: Partnership for Parents support network](#) provides information about sibling support, financial assistance, and emotional/grief support for families and friends of children with life-threatening conditions.
- [NHPCO Facts and Figures: Pediatric Palliative and Hospice Care in America](#) provides an overview of the landscape of pediatric palliative and hospice care for providers, policy makers, funders, and the media.
- [Partnering for Children: Pediatric Outreach Guide](#) offers information for implementing the “Partnering for Children” campaign in the community. Resources on community assessment and outreach, parent survey reports, engaging parents as messengers, working with the media, and funding strategies are included.
- [Get Palliative Care](#) is a consumer and provider Web site geared at providing consumer-friendly information regarding palliative care, a provider directory, and resources for the media, clinicians, policymakers, and family caregivers.

Plan your QI

What are we trying to accomplish? Our aim is:

- To implement in our practice a protocol/system of care to offer a palliative care discussion with patients with a genetic condition, as appropriate, and their families on an annual basis.

How will we know that a change is an improvement? Our measures are:

- Documentation of a discussion or an offer to discuss palliative care with all (100%) patients/families, as appropriate, at least annually. Feedback will be collected from our care coordinator, staff, physicians and patients/families.

What changes can we make that will result in an improvement? Our ideas for change are:

- Care coordinator will use our registry of patients with diagnosed genetic conditions who could benefit from a discussion about palliative care and resources and will add a prompt/note in each patient record, as appropriate. Physicians will document outcomes of discussion/offer to discuss in the patient record, as part of the visit notes.

A comprehensive listing and citations for all the tools and resources listed in the toolkit. Tools and resources are listed in the order they appear in the toolkit.

Section 1 Getting Started

Genetics 101

- Genetics in Primary Care Institute Web site. www.geneticsinprimarycare.org
- Genetics Science Learning Center. University of Utah Health Sciences Web site. <http://learn.genetics.utah.edu/>
- National Coordinating Center and Regional Genetic Services Collaboratives Web site. <http://www.nccrcg.org/>
- Saul, RA. Medical Genetics in Pediatric Practice. Elk Grove Village, IL: American Academy of Pediatrics; 2013
- Time Out for Genetics Webinar Series. Genetics in Primary Care Institute Web site. <http://www.geneticsinprimarycare.org/Provider%20Education/Pages/gpci-webinars.aspx>
- Integrating Genetics into Your Practice Webinar Series. Genetics in Primary Care Institute Web site. <http://www.geneticsinprimarycare.org/Provider%20Education/Pages/gpci-webinars.aspx>
- ACMG Basics: Genetics for Providers. American College of Medical Genetics Web site. https://www.acmg.net/ACMG/Sign_In.aspx?

Ethical, Legal, and Social Issues

- The Genetic Information Nondiscrimination Act. National Coalition for Health Professional Education in Genetics Web site. http://www.nchpeg.org/index.php?option=com_content&view=article&id=97
- “GINA” The Genetic Information Nondiscrimination Act of 2008 Information for Researchers and Health Care Professionals. Department of Health and Human Services. <http://www.genome.gov/Pages/PolicyEthics/GeneticDiscrimination/GINAInfoDoc.pdf>. Published April 6, 2009.
- Genetics Information Nondiscrimination Act. Genetics and Public Policy Center Web site. <http://www.dnapolicy.org/gina/>
- American Academy of Pediatrics Committee on Bioethics, Committee on Genetics, the American College of Medical Genetics, Genomics Social, Ethical, and Legal Issues Committee. Ethical and Policy Issues in Genetic Testing and Screening of Children. Pediatrics. 2013; 620-622.
- American Academy of Pediatrics Committee on Bioethics. Ethical Issues with Genetic Testing in Pediatrics. Pediatrics. 2001;107(6):1451-1455
- Caga-anan EC, Smith L, Sharp RR, Lantos JD. Testing Children for Adult-Onset Genetic Diseases. Pediatrics. 2012;129(1):163-167
- Abdul-Karim R, Berkman BE, Wendler D, et al. Disclosure of Incidental Findings From Next-Generation Sequencing in Pediatric Genomic Research Pediatrics. Pediatrics. 2013;131(3):564-571

Quality Improvement

- Your Medical Home: Well Designed Using a Quality Improvement Process. National Center for Medical Home Implementation Web site. <http://www.pediatricmedhome.org/qib/>
- Quality Improvement Innovation Networks (QuIIN). American Academy of Pediatrics Web site. <http://www.aap.org/en-us/professional-resources/practice-support/quality-improvement/Quality-Improvement-Innovation-Networks/Pages/Quality-Improvement-Innovation-Networks-QuIIN.aspx>

Tools and Resources Index

- Institute for Healthcare Improvement. Institute for Healthcare Improvement Web site. <http://www.ihi.org/Pages/default.aspx>
- How to Improve. Institute for Healthcare Improvement Web site. <http://www.ihi.org/resources/Pages/HowtoImprove/default.aspx>
- Kemper A, Moyer V, First L. Introducing Quality Reports. Pediatrics. 2011;127(1):187 - 188
- EQIPP. EQIPP Web site. <http://eqipp.aap.org/>

Section 2

Collecting Family Health History

Choose a Family Health History Tool for your Practice

- Family Health History. Genetics in Primary Care Institute Web site. <http://www.geneticsinprimarycare.org/YourPractice/Family-Health-History/Pages/Family-Health-History.aspx>

Review and Discuss Family Health History with Patients

- Family Health History. Centers for Disease Control and Prevention Web site. <http://www.cdc.gov/genomics/famhistory/index.htm>
- My Family Health Portrait. Health and Human Services. <https://familyhistory.hhs.gov/fhh-web/home.action>
- Family History is Important for Your Health. Centers for Disease Control and Prevention. <http://www.cdc.gov/genomics/public/file/print/FamHistFactSheet.pdf>
- Los antecedentes familiares son importantes para su salud. Centers for Disease Control and Prevention. http://www.cdc.gov/genomics/spanish/file/print/FamHist_FS_Span.pdf
- Family Health History Record Keeper. Illinois Department of Public Health. http://www.idph.state.il.us/HealthWellness/FamilyHealthHistory_Brochure.pdf
- Does It Run In the Family? Genetic Alliance. <http://www.doesitruninthefamily.com/>
- National Coordinating Center and Regional Genetic Services Collaboratives Web site. <http://www.nccrcg.org/>
- ASHG Video Series. The American Society of Human Genetics Web site. http://www.ashg.org/education/family_history.shtml/

Section 3

Identifying Patients with Suspected Genetic Conditions

Identify and Follow-up with Patients Identified at Risk

- Whelan AJ, Ball S, Best L, et al. Genetic red flags: clues to thinking genetically in primary care practice. Prim Care. 2004;31(3):497-508
- Role of Ethnicity in Genetics. Genetics in Primary Care Institute Web site. <http://www.geneticsinprimarycare.org/YourPractice/Family-Health-History/Pages/Genetic%20Red%20Flags.aspx#jump-3>
- What is a Pediatric Geneticist? Healthychildren.org Web site. <http://www.healthychildren.org/English/family-life/health-management/pediatric-specialists/Pages/What-is-a-Pediatric-Geneticist.aspx>
- Making Sense of Your Genes: A Guide to Genetic Counseling. Genetic Alliance Web site. <http://www.geneticalliance.org/publications/guidetogeneticcounseling>

Tools and Resources Index

- Informed Consent for Genetic Testing: Patient Education Information for use with the Michigan Model Consent Form for Genetic Testing. Michigan Department of Community Health. http://www.michigan.gov/documents/InformedConsent_69182_7.pdf. Published May 2006.
- Direct-to-Consumer Genetic Tests. Federal Trade Commission Web site. <http://www.ftc.gov/bcp/edu/pubs/consumer/health/he02.pdf>

Develop Relationships with Genetic Professionals

- Find a Genetic Counselor. National Society of Genetic Counselors. <http://nsgc.org/p/cm/ld/fid=185>
- American College of Medical Genetics Clinical Services Search Engine. American College of Medical Genetics Web site. https://www.acmg.net/ACMG/Find_Genetic_Services/ACMG/ISGweb/FindaGeneticService.aspx?hkey=720856ab-a827-42fb-a788-b618b15079f9
- Pediatric & Clinical Genetics. National Society of Clinical Genetics Web site. <http://nsgc.org/p/cm/ld/fid=49#ped>
- Genetic Counseling in Primary Care. Genetics in Primary Care Institute Web site. Published January 2013. <https://www.youtube.com/watch?v=nulc3cfJkM&feature=youtu.be>
- National Coordinating Center for the Regional Genetic Services Collaboratives Web site. <http://www.nccrcg.org/>

Order Appropriate Genetic Tests

- Genomic Testing. Centers for Disease Control and Prevention Web site. <http://www.cdc.gov/genomics/gtesting/index.htm>
- Frequently Asked Questions About Genetic Testing. National Human Genome Research Institute. <http://www.genome.gov/19516567>
- Genetic Disorders, Genomics and Healthcare. National Human Genome Research Institute. <http://www.genome.gov/27527652>
- The Genomics in Medicine Lecture Series. National Human Genome Research Institute. <http://www.genome.gov/27546022>
- Pagon RA, Adam MP, Bird TD, et al., eds. GeneReviews. Seattle, WA: University of Washington; 1993-2014
- GTR: Genetic Testing Registry. National Center for Biotechnology Information. <http://www.ncbi.nlm.nih.gov/gtr/>
- Mercier CE, Barry SE, Paul K, et al. Improving Newborn Preventive Services at the Birth Hospitalization: A Collaborative, Hospital-Based Quality-Improvement Project. Pediatrics. 2007;120(3):481-488
- Overview of Genetic Testing and Screening. Genetics in Primary Care Institute Web site. <https://www.youtube.com/watch?v=AyH8n-AxDXY&feature=youtu.be>. Published October 2013.
- Ordering the Right Tests - Genetics in Primary Care. Genetics in Primary Care Institute Web site. http://www.geneticsinprimarycare.org/Provider%20Education/Documents/GPCI_OrderingTests_05.31.12.wmv. Published June 2013.
- Genetic Testing in Primary Care. Genetics in Primary Care Institute Web site. <https://www.youtube.com/watch?v=IEOounOvGGE&feature=youtu.be>. Published December 2013.

Section 4

Providing Appropriate Care for Patients with Genetic Conditions

- Bright Futures. Bright Futures Web Site. <http://brightfutures.aap.org/index.html>
- Medical Home Portal. Medical Home Portal Web site. www.medicalhomeportal.org
- National Center for Medical Home Implementation. National Center for Medical Home Implementation Web site. www.medicalhomeinfo.org

Implement Systems to Improve Genetic Services

- Recalls and Reminder Systems. American Academy of Pediatrics Bright Futures Web Site. <http://brightfutures.aap.org/clinical/volume1.html>
- Building Your Medical Home and Getting Paid Appropriately. National Center for Medical Home Implementation. http://www.pediatricmedhome.org/pdfs/6_payment.pdf
- Index of CPT Codes for Medical Home. American Academy of Pediatrics. <http://www.medicalhomeinfo.org/downloads/pdfs/MedicalHomeCodingFactSheet.pdf>. Published January 4, 2010.
- Genetic Coding Fact Sheet. Genetics in Primary Care Institute. http://www.geneticsinprimarycare.org/YourPractice/Documents/GeneticCodingFactSheet_FINAL.pdf
- Genetics and Coding: What the Primary Care Provider Needs to Know. Genetics in Primary Care Institute Web site. <http://www.geneticsinprimarycare.org/Provider%20Education/Pages/gpci-webinars.aspx>
- Insurance Coverage Toolkit for Chromosomal Microarray Testing. The International Collaboration for Clinical Genomics. <http://www.iccg.org/researcher-and-clinician-information/iccg-toolboxes/>

Create a Patient Registry

- Ortiz DD. Using a Simple Patient Registry to Improve Your Chronic Disease Care. *Fam Pract Manag.* 2006;13(4):47-52.
- Newborn Screening ACT Sheets and Confirmatory Algorithms. American College of Medical Genetics Web site. https://www.acmg.net/ACMG/Resources/ACT_Sheets_and_Confirmatory_Algorithms/ACMG/Resources/ACT_Sheets_and_Confirmatory_Algorithms/ACT_sheets_Homepage.aspx?hkey=1654e248-53f2-41a9-8d93-f3d96de05e0a
- Moeschler JB, Shevell M. American Academy of Pediatrics Committee on Genetics. Clinical Genetic Evaluation of the Child with Mental Retardation or Developmental Delays. *Pediatrics.* 2006;117(6):2304 -2316
- Genetic and Rare Conditions Site. Medical Genetics, University of Kansas Medical Center Web site. <http://www.kumc.edu/gec/support/>

Health Supervision Guidelines for Patients with Genetic Conditions

- Trotter TL, Hall JG, American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Achondroplasia. *Pediatrics.* 2005;116(3):771 -783
- Bull MJ, American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Down Syndrome. *Pediatrics.* 2011;128(2):393 -406
- Hersh JH, Saul RA, American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Fragile X. *Pediatrics.* 2011;127(5):994 -1006
- American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Marfan Syndrome. *Pediatrics.* 1996;98(5):978 -982

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- Hersh JH, American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Neurofibromatosis. *Pediatrics*. 2008;121(3):633 -642
- American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Prader-Willi Syndrome. *Pediatrics*. 2011;127(1):195 -204
- American Academy of Pediatrics Section on Hematology/Oncology, American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Sickle Cell Disease. *Pediatrics*. 2002;109(3):526 -535
- Bondy CA, Turner Syndrome Study Group. Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group. *J Clin Endocrinol Metab*. 2007;92(1):10-25
- American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Williams Syndrome. *Pediatrics*. 2001;107(5):1192 -1204
- American Academy of Pediatrics Periodicity Schedule. <http://www.aap.org/en-us/professional-resources/practice-support/Pages/PeriodicitySchedule.aspx>. American Academy of Pediatrics Web site.

Provide Family-centered Care

- Patient-Centered Medical Home Version of the Consumer Assessment of Healthcare Providers and Systems (CAHPS) Clinician Group Survey. Agency for Healthcare Quality and Research Web site. <https://cahps.ahrq.gov/surveys-guidance/cg/pcmh/index.html>
- The Medical Home Family Index and Survey. Center for Medical Home Improvement Web site. http://www.medicalhomeimprovement.org/pdf/PediatricMedicalHome-FamilyIndexandSurvey_2005.pdf. Published 2001.
- Malouin RA. Positioning the Patient and Family at the Center: A Guide to Family and Patient Partnership in the Medical Home. American Academy of Pediatrics, Elk Grove Village, IL; National Center for Medical Home Implementation. http://www.medicalhomeinfo.org/downloads/pdfs/Positioning_FINAL_May24.pdf. Published 2013.
- Family-Centered Care Self-Assessment Tool: Provider Tool. Family Voices Web site. http://www.familyvoices.org/admin/miscdocs/files/fcca_ProviderTool.pdf. Published October 2008.
- Family-Centered Care Self-Assessment Tool: Family Tool. Family Voices Web site. http://www.familyvoices.org/admin/work_family_centered/files/fcca_Family-Tool.pdf. Published October 2008.
- Cultural Competence Health Practitioner Assessment. Georgetown University Web site. <https://www4.georgetown.edu/uis/keybridge/keyform/form.cfm?formID=277>

Section 5

Coordinating Care for Patients with Genetic Conditions

Improve Processes to Co-manage Care with Specialists

- Care Co-Management Among Patient/Family, Pediatric Medical Home and Specialists. National Center for Medical Home Implementation Web site. http://www.pediatricmedhome.org/pdfs/3_Co_Management_Agreement.pdf
- Comprehensive Care Planning. Medical Home Learning Collaborative Web site. http://medicalhomeinfo.org/how/care_delivery/#coordination

Obtain Current Emergency Plans (if applicable)

- Acute Illness Protocols. New England Consortium of Metabolic Programs Web site. <http://newenglandconsortium.org/for-professionals/acute-illness-protocols/>
- Emergency Guidelines. British Inherited Metabolic Disease Group Web site. <http://www.bimdg.org.uk/protocols/disclaimer.asp>

Obtain Current Emergency Letters (if applicable)

- Maple Syrup Urine Disease (MSUD) - Treatment and Management. Medical Home Portal Web site. <http://www.medicalhomeportal.org/link/4711>

Plan for the Transition to Adult Care

- American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians, Transitions Clinical Report Authoring Group. Supporting the Health Care Transition from Adolescence to Adulthood in the Medical Home. *Pediatrics*. 2011;128(1):182 -200
- Got Transition. Got Transition Web site. <http://www.gottransition.org/>
- Six Core Elements of Health Care Transition Toolkit. Got Transitions Web site. <http://www.gottransition.org/6-core-elements>
- Family Resources. Got Transitions Web site. <http://www.gottransition.org/families-resources>
- Watson R, Parr JR, Joyce C, May C, Le Couteur AS. Models of Transitional Care for Young People with Complex Health Needs: A Scoping Review. *Child Care Health Dev*. 2011;37(6):780-91.
- Transitions to Adult Care ACT Sheets and Confirmatory Algorithms. American College of Medical Genetics and Genomics Web site.
https://www.acmg.net/ACMG/Resources/ACT_Sheets_and_Confirmatory_Algorithms/Transition_ACT_Sheets_and_Algorithm_Table/ACMG/Resources/ACT_Sheets_and_Confirmatory_Algorithms/Transition_ACT_Sheets_and_Algorithm_Table.aspx?hkey=fa2225b6-5790-4387-8018-27d8c77d1604

Discuss Palliative Care (if applicable)

- American Academy of Pediatrics Committee on Bioethics and Committee on Hospital Care. Palliative Care for Children. *Pediatrics*. 2000;106(2):351 -35.7
- Levetown M, and American Academy of Pediatrics Committee on Bioethics. Communicating With Children and Families: From Everyday Interactions to Skill in Conveying Distressing Information. *Pediatrics*. 2008;121(5):e1441 -e1460
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- Browning DM, Meyer EC, et al. Difficult Conversations in Health Care: Cultivating Relational Learning to Address the Hidden Curriculum. *Acad Med*. 2007;82(9):905-13.
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- Wolfe J, Hinds P, Sourkes B. *Textbook of Interdisciplinary Pediatric Palliative Care*. Philadelphia, PA: Saunders; 2011:402-7
- Partnership for Parents. Partnership for Parents Web site. <http://www.partnership-forparents.net/>
- NHPCO Facts and Figures: Pediatric Palliative and Hospice Care in America. National Hospice and Palliative Care Organization.
http://www.nhpco.org/sites/default/files/public/quality/Pediatric_Facts-Figures.pdf. Published April 2009
- Partnering for Children: Pediatric Outreach Guide. National Hospice and Palliative Care Organization.
http://www.caringinfo.org/files/public/outreach/Outreach_Guide_Pediatric.pdf. Published 2009
- What is Pediatric Palliative Care Get Palliative Care Web site. <http://www.getpalliativecare.org/whatis/pediatric/>