Appendix A

Instructions for Chart and Genetic Condition Registry Review

For questions contact:
Ruth S. Gubernick, MPH
Quality Improvement Advisor
Phone: 856/751-0115
Fax: 856/489-9035
Email: gubernrs@hlh.com

Overview
Each month for the next 6 months, from April 15, 2013 through September 30, 2013 you will be asked to submit (1) a total of 10 patient chart reviews and (2) answer a set of questions about patients with known genetic conditions in your registry, using the AAP’s Quality Improvement Data Aggregator (QIDA) System. You will also submit a total of 20 patient chart reviews and answer a set of questions about patients with known genetic conditions in your registry for baseline data on January 30, 2013.

Data cycles will be open on the 15th of each month. This means that you should not submit your monthly data until the 15th of every month. Data will be due by the 30th of each month. See the Data Cycle Table provided below for details.

<table>
<thead>
<tr>
<th>Data Cycle Label</th>
<th>Month of Data Cycle</th>
<th>Data Cycle Opens</th>
<th>Data Cycle Closes</th>
<th>Data to be submitted</th>
</tr>
</thead>
<tbody>
<tr>
<td>Baseline</td>
<td>January</td>
<td>January 15, 2013</td>
<td>January 30, 2013</td>
<td>1st 20 chart of patients 0-21 yrs, seen in January for a Health Supervision Visit. Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 1</td>
<td>April</td>
<td>April 15, 2013</td>
<td>April 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in April for a Health Supervision Visit Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 2</td>
<td>May</td>
<td>May 15, 2013</td>
<td>May 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in May for a Health Supervision Visit Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 3</td>
<td>June</td>
<td>June 15, 2013</td>
<td>June 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in June for a Health Supervision Visit Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 4</td>
<td>July</td>
<td>July 15, 2013</td>
<td>July 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in July for a Health Supervision Visit Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 5</td>
<td>August</td>
<td>August 15, 2013</td>
<td>August 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in August for a Health Supervision Visit Answer a set of questions about patients with known genetic conditions in your registry</td>
</tr>
<tr>
<td>Follow-up 6</td>
<td>September</td>
<td>September 15, 2013</td>
<td>September 30, 2013</td>
<td>1st 10 charts of patients 0-21 yrs, seen in</td>
</tr>
</tbody>
</table>
Teams should not submit data until the Data Cycle Opens (15th of month)

Each month, you will receive an email from project staff with instructions for data submission. You will need to complete the on-line chart review survey for each patient chart you review and the answers to a set of questions regarding your registry of patients with identified genetic conditions. You have the option of first completing a “hardcopy” survey of each chart reviewed and the registry questions, using the PDF Data Collection Tool (provided in the Learning Session packet and posted on the Monthly Data Collection page of the Project Web site each month), but you must then submit that data as described using the QIDA. Note: Only the practice/group administrator identified in your project application will have access to enter data into the QIDA.

Doing this data collection each month will help to maximize your team’s learning during the 6-month Action Period following the Learning Session. The chart and registry reviewer for the six month Action Period should be someone designated from your core improvement team. Below is a brief set of instructions to guide you in reviewing your charts and registry of patients with genetic conditions:

- Select charts of the first 10 patients 0-21 yrs. seen by any provider in the practice for a health supervision visit. You will submit a total of 10 total chart reviews per practice each month (with the exception of baseline, which will be a total of 20 chart reviews per practice).

- Access your registry of patients with defined genetic conditions so you can answer the set of questions regarding the status and care management of this population of patients.

- Project staff will send an email each month with instructions for accessing QIDA to enter your data. You can also find information about your data collection requirements each month in the QIDA system. Your group administrator will login to the QIDA, by entering the ID and password provided to them. All core improvement team members will have access to the QIDA, but the group administrator will have another level of access to enter data each month. Each team member will log into QIDA with a unique user ID and password. AAP members will use their existing login information (for instance, the login used to enter the AAP Member Center). Other team members will be emailed their user ID and password. The email will come from Jason Newsome.

- As an option, you may choose to first complete the appropriate “hardcopy” Data Collection Tool for each record of interest and your registry and then use these “hardcopy” forms to complete the on-line survey for each record and your registry of patients with defined genetic conditions.

- **IMPORTANT: Do not submit charts for the next month before the 15th of the next month.**

- See specific question instructions on the next page for completing Data Collection Tool and/or the QIDA on-line survey for each chart and the registry you are reviewing at baseline and for the following 6 month period.
**Genetics in Primary Care Institute Quality Improvement Project**
**Chart and Registry Review Instructions**

**Chart Review:** The purpose of the chart review is to help identify current performance and areas for improvement. Some information may not be clearly documented in the patient’s chart and may be inferred based on other information listed in the chart. While every attempt should be made to record data as accurately as possible, the goal in quality improvement data collection is usefulness, not perfection.

---

**Chart review for the first 20 patients seen by any provider in the practice during January 2013.**

***Please select and complete a chart review for a total of 20 patients meeting the criteria above for the Prework period. For all other Chart Reviews during the Action Period, please select and complete a chart review for a total of 10 patients meeting the criteria above. ***

**Please review the patient’s chart to collect the information needed** on the *Data Collection Tool* and/or in the QIDA system.

**Questions to qualify patient record:**

1) Month of review:

2) Is a multi-generational family history obtainable for this patient?

Answer ‘NO’ if there is documentation in the chart that there are reasons that a multi-generational family history is not obtainable for this patient (i.e., unknown birth parent(s)) and EXCLUDE this patient from this review.

3) Is there documentation in the chart that a multi-generational family history was created or updated/maintained for this patient using the components defined by the project?

Answer ‘Yes, All project-defined components included’ if there is documentation that a multi-generational family history was created or updated/maintained at the health supervision visit using all components defined for the project (Note: see pg. 28 at the end of this Appendix for a list of defined family history components). Answer, ‘Yes, one or more project-defined components included’ if there is documentation that a multi-generational family history was created or updated/maintained at the health supervision visit using one or more components defined by the project (pg. 28) Otherwise, answer ‘No.’

4) Is there documentation in the chart that the current multi-generational family history was discussed with the patient/family?

Answer ‘Yes’ if there is documentation that the family history was discussed with the patient/family at this health supervision visit. Otherwise, answer ‘No.’

5) Did the multi-generational family history indicate risk? (Was the family history positive)?

Answer ‘Yes’ if there is documentation that the family history indicated risk. Otherwise, answer ‘No.’
6) Did the patient present with any clinical concerns (physical signs, cognitive concerns) of a genetic condition?

Answer ‘Yes’ if there is documentation that the patient presented with any clinical concerns (physical signs, cognitive concerns) of a genetic condition. Otherwise, answer ‘No.’

If you answered ‘Yes’ to 5 and/OR 6:

7) Is there documentation in the chart that a follow-up plan of care was discussed with the patient/family?

Answer ‘Yes’ if there is documentation that a follow-up plan of care was discussed with the patient/family. Otherwise, answer ‘No.’

If you answered ‘Yes’ to 7:

8) Did you make the referral(s) (to a lab, geneticist, or other program/service) that were documented in the follow-up plan?

Answer ‘Yes’ if there is documentation that referral(s) (to a lab, geneticist, or other program/service) that were documented in the follow-up plan. Otherwise, answer ‘No.’ Note: Answer ‘N/A,’ if there were no referrals necessary for this follow up plan.

If yes to 8:

9) Did you track this/these referral(s) (enter it into your registry and/or referral tracking mechanism)?

Answer ‘Yes’ if there is documentation that referral(s) (to a lab, geneticist, or other program/service) were documented in your registry and/or referral tracking mechanism. Otherwise, answer ‘No.’

**Aggregate Registry Review:** The purpose of the registry review is to help identify current performance and areas for improvement regarding the care and management of your patients age 0-21 yrs. with diagnosed medical conditions, as a population.

<table>
<thead>
<tr>
<th>Registry review for all of the patients with diagnosed genetic conditions in your practice registry during January 2013.</th>
</tr>
</thead>
</table>

**Please note:** The instructions for creating a registry for your patients with the identified ICD-9 Codes and an example of an Excel file of data fields for tracking care management were included as a separate attachment with the materials you received upon acceptance. Please reference that document for information about building your registry.

1) Month of review:

2) How many patients with defined genetic conditions are included in your practice’s patient registry?

Enter the total number of patients age 0-21 yrs. in your registry.
3) Of those patients with defined genetic conditions included in your registry, how many are up-to-date with age appropriate Health Supervision Visits?

Enter the number of patients who are up-to-date with age appropriate Health Supervision Visits.

3a) Of those patients with defined genetic conditions included in your registry who are up-to-date with age appropriate health supervision visits, how many have genetic conditions with existing Health Supervision Guidelines?

Based only on the patients identified in Q3 (up-to-date with age appropriate health supervision visits), enter the number of those patients with conditions for which there are existing Health Supervision Guidelines.

3b) Of those patients in 3a, how many have documentation in the chart that the Health Supervision Guideline was followed at the most recent visit?

Based only on the number of patients in Q3a, enter the number of those patients with documentation in their chart that the condition-specific Health Supervision Guideline was followed at this most recent visit.

4) Of those patients with defined genetic conditions included in your registry, how many have documentation in the registry of next steps and planned follow-up?

Based on the total number of patients in your registry (Q1), enter the number of those patients with documentation in the registry of next steps and planned follow-up.

5) How many patients in your registry with defined genetic conditions require an emergency plan?

Enter the number of patients in your registry that require an emergency plan.

5a) Of those patients in 5, how many have a current (at least annual) emergency plan included in their chart?

Based only on the number of patients identified in Q5, enter the number of patients with a current (at least annual) emergency plan included in their chart.

5b) Of those patients in 5, how many have documentation that a request was made to a specialist to obtain an emergency plan within the last month if a current plan (at least annual) was not included in the chart?

Based only on the number of patients identified in Q5 and subtracting those with a current (at least annual) emergency plan, enter the number of patients that have documentation that a request was made to a specialist to obtain an emergency plan within the last month.

6) Of those patients with defined genetic conditions included in your registry, how many have documentation in the registry that genetic services were offered to the patient/family at least initially?

Enter the number of patients in your registry that have documentation in the registry that genetic services were offered to the patient/family at least initially.

7) How many patients in your registry with defined genetic conditions are age 12-21 yrs. old?
Enter the total number of patients who are 12-21 yrs. old.

7a) Of those patients in 7, how many have documentation in the registry that discussions about transition to adult care and resources have been offered at least annually to the patient/family?

Based only on the number of patients identified in Q7, enter the number of patients that have documentation in the registry that discussions about transition to adult care and resources have been offered at least annually to the patient/family.

8) How many patients in your registry require a palliative care discussion?

Enter the number of patients in your registry that require a palliative care discussion.

8a) Of those patients in 8, how many have documentation in the registry that a palliative care discussion was offered at least annually to the patient/family?

Based only on the number of patients identified in Q8, enter the number of patients with documentation of a current (at least annual) palliative care discussion included in their chart.

**QIDA User’s Manual**
A QIDA User’s Manual was included as a separate attachment with the materials you received upon acceptance. Please reference this document for information about the QIDA system.
Genetics Family History Components

GENERAL FAMILY HISTORY ELEMENTS

- Should include following family members: parents, brothers, sisters, aunts, uncles, 1st cousins, grandparents
- Medical conditions that run in the family (2 or more family members with the condition)
- Ethnicity
- Consanguinity
- Any parent and/or patient's concern about family history
- Incomplete knowledge of family history (ie adoptions, estranged family members)

ANY FAMILY MEMBERS WHO HAVE BEEN TOLD THEY HAVE:

(ask who and what for each of the below):

- Birth Defects:
  - Structural birth defects (examples: congenital heart disease, spina bifida, extra or missing fingers, clubfeet)
  - Sensory birth defects (examples: congenital deafness, congenital blindness)
- Cancer <50 years and specify type
- Carrier of genetic condition
- Clotting, bleeding or blood disorder (examples: sickle cell, von Willebrands, hemophelia)
- Developmental delay, intellectual disability, autism spectrum disorder, 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