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BRIEF COMMUNICATION

Development and implementation of an electronic medical record module to track genetic testing results

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PURPOSE: Genetic testing and results return pose many challenges, even in the era of electronic medical records. Whether results are positive or negative, genetic testing and return of results necessitate patient follow-up, referrals, and coordination between providers. Genetic evaluations typically utilize a variety of testing modalities with differing timetables and/or avenues to return. Therefore, genetic information requires a secondary, unified mechanism for storing and tracking results and communication to facilitate patient care.

METHODS: We developed an electronic medical record (EMR) episodes-based module called Pediatric Genetic Tracking to provide a centralized summary of patient tracking information in a single-institution pediatric genetics setting.

RESULTS: We created episodes for 6,133 patients evaluated in our division over a 3-year period. They highlighted clinical information for 1,901 different diagnoses and 547 genetic tests, and the involvement of 9 providers, 7 genetic counselors, 61 trainees, and 15 students using two modes of follow-up.

CONCLUSION: This Pediatric Genetic Tracking episodes system serves as a "one-stop shop" living document for updated patient genetic information and can be easily expanded to include variant content for broader population level sharing or analysis. These episodes-based modules facilitate communication to support timely and accurate return of genetic test results and follow-up.

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INTRODUCTION

Genetic testing is a powerful diagnostic tool in medical care and plays an important role in major decisions that affect health, reproductive choice, and pursuit of therapies or surgeries. Several papers have outlined the technical challenges in integrating genetic testing and electronic medical records (EMR).[1–3] The Electronic Medical Records and Genomics (eMERGE) network, which seeks to leverage the EMR for genomic research, has experienced logistical challenges in integration of genetic information into established EMR systems. These challenges include data storage, representation and exchange of patient genomic and variant data, and clinical decision support.[4]

Traditional laboratory results (such as hematology or chemistry panels) are explicitly annotated within the EMR, are often completed in an internal clinical laboratory, and are entered in the EMR as discrete data points that are interpreted as normal or abnormal. In contrast, genetic testing is frequently entered in the EMR as a generic "sendout" test, obtained from an external laboratory and scanned into the media tab as a pdf document that cannot be searched by character recognition software.[5–7] How the ordering clinician synthesizes that result is not included in the report: placement of this interpretation remains inconsistent within the EMR and may include multiple locations such as clinician notes, problem lists or, in some cases, even the allergies section.[1,8,9] Additionally, the interpretation provided by the laboratory may not completely match the assessment by the clinician, which may also change over time.[7] As such, mechanisms that help process traditional laboratory testing within the EMR do not currently translate to genetic testing.

In addition to the current challenges of integrating genetic tests into the EMR, there are unique attributes associated with genetic testing that elevate the importance of careful tracking and follow-up. These include reclassification of genetic variant data with the availability of new genome level sequence data, confusion over ownership of genetic testing (e.g., patient, provider, lab, or a combination of these), and complex health-care systems with multiple providers, all of which necessitate a reliable mechanism of secondary EMR oversight for genetic testing.[1,7,8] Genetic testing is also expensive, requiring prior authorization from payers that can take prolonged periods of time and can therefore be difficult to track.[10,11] Test processing and results can also take weeks to months, increasing the possibility of errors in the process and resultant mistakes or mishandling of results.

An EMR module and living document could help address many of these concerns and reduce the likelihood of systems failure. Our division previously relied on a combination of paper forms and a Microsoft Access database file for tracking genetic testing; however, this paradigm eventually proved insufficient for our expanding patient population. Due to storage size limitations, Microsoft Access also could not reliably handle our patient database, and its ability to run queries successfully was inconsistent. Counselors or students were often tasked with tracking down the responsible provider for any missing information, and a genetic counselor audited the database on a weekly basis, detracting from their clinical duties. To address these inefficiencies, our goal was to leverage a functionality already present in our EMR and to improve the utility of this database.

MATERIALS AND METHODS

Development of module: The Michigan Medicine Division of Pediatric Genetics, Metabolism and Genomic Medicine in the Department of Pediatrics utilizes the Epic EMR (Verona, WI) for patient care documentation. One feature of this EMR is the Episodes of Care tab ("Episodes"). This

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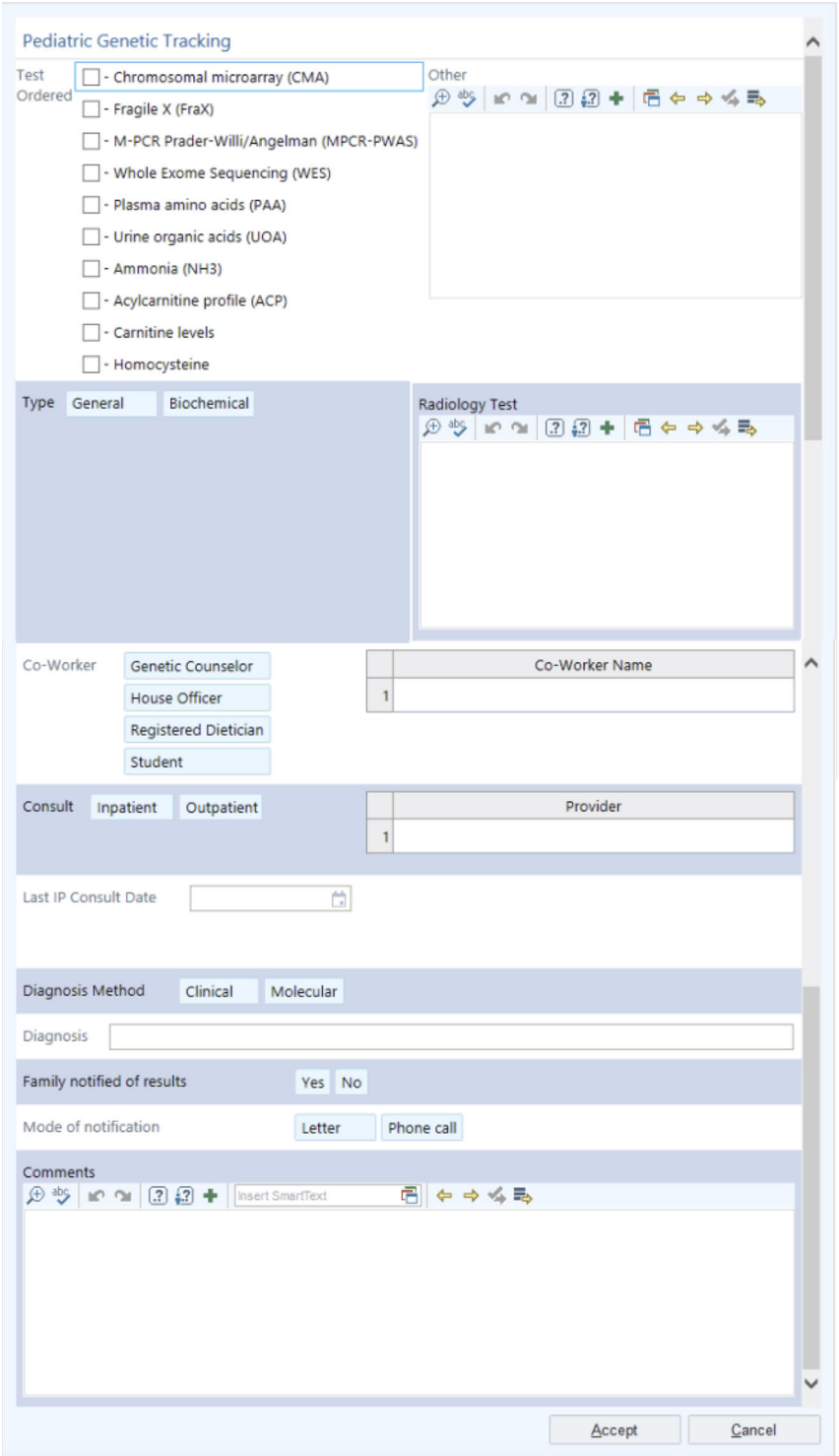

Fig. 1 Screenshot of the Pediatric Genetic Tracking Episode entry module. Information is entered using a combination of pre-selected options, free text boxes and searchable fields.

feature exists to capture financial implications and multidisciplinary provider effort incurred by chronic medical conditions. It allows clinical information to be consolidated in a single location within the patient's chart to coordinate care and remuneration.[12–14]

We developed a reporting mechanism whereby a provider populates an electronic module to capture information from patient encounters (shown in Fig. 1). After seeing a patient, providers enter information into this Episodes of Care functionality using a combination of free text, preselected options to click on, and searchable fields as outlined in Table 1. This information includes the testing that was ordered (such as radiological studies if applicable), the type of patient (general genetics versus biochemical patient), providers participating in the case, location (inpatient versus outpatient), date of the consult, whether the diagnosis was based on clinical criteria or molecular testing, patient diagnosis, and information about results return (whether the family was notified and if so, how [phone call versus letter]). The Episodes module is visible and editable to all

Table 1. Data fields and mode of entry for Pediatric Genetic Tracking Episodes.

Field	Method of entry
Medical record number	Autopopulated
Patient name	Autopopulated
Date of birth	Autopopulated
Age	Autopopulated
Sex	Autopopulated
Admitted to hospital? (yes/no)	Autopopulated
Consult (inpatient vs. outpatient)	Clickable selection
Last inpatient consult date	Dropdown box
Last outpatient visit date	Autopopulated
Provider	Dropdown box
Test(s) ordered	Clickable selection
Radiology test(s) ordered	Free text
Other test(s)	Free text
Genetic visit type (general vs. biochemical)	Clickable selection
Diagnosis method (clinical vs. molecular)	Clickable selection
Diagnosis	Free text
Coworker type (counselor, house officer, student)	Clickable selection
Coworker name	Dropdown box
Family notified? (yes/no)	Clickable selection
Notification mode (letter, phone call)	Clickable selection
Comments (e.g., follow-up recommendations)	Free text

providers who have access to the EMR, and can be queried using the Epic EMR's reporting mechanism.

Table 1 details the fields that can be queried for filtering to run a report in Epic, which includes the above categories plus some data automatically populated from a patient's EMR. After the query has been run, individual patients can be selected to navigate to their clinical information, or providers can optionally export their query to an Excel file for further analysis, modification, and/or distribution.

Implementation of module

We implemented the module on 1 May 2017, and built a reporting mechanism within the Epic EMR that could be used to access the data entered. Using this reporting mechanism, we obtained the total number of patients entered into the Pediatric Genetic Tracking module over a three-month period from 1 October to 31 December 2019. To assess utilization of the Episodes, we compared the list of patients seen in our division as reflected in the Episode report to a list generated manually by review of inpatient consult orders and outpatient clinic schedules in the EMR over the same time period. Patients without an Episode of Care were identified and entered into an Excel spreadsheet.

RESULTS

Over three years (1 May 2017–30 April 2020), information from 6,133 patients evaluated by our division was entered into the Pediatric Genetic Tracking Episode. This patient population exhibited 1,901 different diagnoses and had undergone 547 different genetic tests. These 6,133 patients were evaluated by 9 clinical physician providers, 7 genetic counselors, 61 resident or fellow trainees, 15 medical students, and 2 modes of testing follow-up (letter versus phone call).

Table 1 lists all data fields that can be queried in the Episodes. Data in these fields can be collected by running a report and downloading a filterable, searchable Excel file, which can be further modified, analyzed, or distributed. Wherever possible, structured elements (such as autopopulation, dropdown boxes, and clickable selections) were utilized to facilitate standardization and consistency in data entry.

Several additional functionalities of the Episode reports have proven to be useful in the clinic. They can assist in follow-up of inpatient evaluations for weekly handoffs on the consult service. Episode data can also be used for tracking patient return visits and identifying patients that were recommended for follow-up. Episode reports can identify pairings between trainees or counselors and attending physicians as well as patients with specific diagnoses. These are some of the many ways we have used these data, and we expect there are many more.

Assessing module implementation by providers: To determine provider utilization of these Episodes, we queried a three-month period from 1 October to 31 December 2019. Among the 688 patients seen by our division during this time period, only 24 did not have a Pediatric Genetic Tracking Episode, indicating 96.5% provider utilization of Episodes and data entry over this timeframe.

DISCUSSION

This paper describes the creation and implementation of Episodes as living documents to support adequate genetics follow-up for patients. They do not rely on any external program or database beyond the Epic EMR. Additionally, they facilitate communication between providers, trainees, and administrators while maintaining personal patient health information privacy. Information entered into Episode modules is visible to all other providers who have access to the same EMR (including those in other departments), allowing them to familiarize themselves with the patient's genetic diagnoses and workup without needing to search through clinic notes and test results.

We observed several advantages of the Episodes module compared with our previous paper-based manual entry tracking system. First, providers enter data themselves after or during an encounter, eliminating the need for an administrator, genetic counselor, or student to enter and audit this information manually. Second, cumulative data within the Episodes can be seamlessly obtained in real time through generation of reports. This enables analysis of information such as (1) which patients were seen by a specific provider, (2) which diagnoses were established for various patients, and (3) which specific genetic tests have been ordered, among others. Third, active and consistent use of Episodes allows for a comprehensive system that fosters coordination of care and provides a centralized accessible location for patient information. Instead of searching through a variety of visit notes, information such as tests ordered and working diagnoses are now stored in a single location. Finally, Episodes allows the provider to be the arbiter of data entry, compared with our previous system, which relied on other individuals who may have been unfamiliar with the patient's detailed information. Given the ability to query patients by multiple categories, Episodes has the potential to vastly improve the quality of patient care, medical education, and research. In addition to these user features mentioned above, the application of "Episodes of Care" is, to our knowledge, the first of its kind to be built into the EMR for a clinical genetics purpose. As the idea of Episodes of Care was initially developed to organize clinical care and remuneration for specific long-term diagnoses such as diabetes and hip fractures,[12–14] the same approach is also applicable to coordination of genetics care and testing.

This customized Episode can be easily adapted in other EMR systems to help coordinate genetic testing at other institutions. At this time, with the exception of the demographics and date of last visit, many data fields in the Episode require manual entry.

However, future versions could be developed so that additional data fields are automatically populated with information that is already entered elsewhere in the EMR to ensure adherence and accuracy. Additional improvements could include a section to specifically track follow-up (if follow-up is needed and, if so, how frequently).

Several limitations arose during our analysis of this Pediatric Genetic Tracking Episode functionality. First, providers were allowed to enter free-text diagnoses and genetic tests into this system rather than using ICD-10 designations or other structured terminology. This is a limitation that prevents standardization in our reports and searches, and lessens its utilization of algorithm-based clinical decision support. Future updates to the Episodes could include ICD-10 codes and other phenotype or genotype information. In the meantime, Episodes currently effectively captures the heterogeneity of patient diagnoses seen in our clinic, as ICD-10 codes are often imprecise and unable to describe many specific genetic diagnoses without loss of phenotypic information.[7] However, adding ICD-10 codes may make the Episodes more useful, especially for implementing management guidelines. Additionally, we attempted to quantify the improvement in efficiency (e.g., loading time of databases, data entry, etc.) between our Microsoft Access database and the Episodes module; however, the Microsoft program crashed before the data were fully loaded, preventing us from providing a concrete comparison between these two systems.

Therefore, while we cannot compare Episodes against our previous system, there are still multiple metrics that could be analyzed in the future to determine how Episodes influence patient care. Specifically, we could investigate the number of times the database is queried over a given time period and what specific queries are performed. In doing so, we could investigate how Episodes may identify deficiencies in care such as need for follow-up appointments or aid in provider handoffs. Episodes could also be assessed through surveys of providers as well as further analysis of the database. This future work would provide additional data about how Episodes of Care directly improves patient care.

In summary, our Episodes of Care module works within the framework of existing mechanisms to consolidate information relevant to a patient population receiving genetics service, thus facilitating their ongoing care. Moreover, this module can be readily applied to other Epic-based EMR systems.

DATA AVAILABILITY

Data and materials are available individually upon request.

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AUTHOR CONTRIBUTIONS

Conceptualization: A.S., D.M.M. Data curation: A.S. Formal analysis: A.S., D.M.M. Methodology: A.S., D.M.M. Writing—original draft: A.S., D.M.M. Writing—review & editing: A.S., D.M.M.

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

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