In the age of genomic medicine, family health history (FHH) remains an important tool for personalized risk assessment as it can inform approaches to disease prevention and management. In primary care, including in prenatal settings, providers recognize that FHH enables them to assess the risk for birth defects and complex conditions that not only affect the fetus’ health, but also the mother’s. However, many providers lack the time to gather FHH or the knowledge to confidently interpret the data. Electronic tools providing clinical decision support using FHH data can aid the busy provider with data collection and interpretation. We describe the scope of conditions included in a patient-entered FHH tool that provides clinical decision support and point-of-care education to assist with patient management. This report details how we selected the conditions for which it is appropriate to use FHH as a means to promote personalized medicine in primary prenatal care.

KEYWORDS: clinical decision support  family health history  genetic screening  personalized risk assessment  prenatal care

Among the personalized medicine tools currently available at the point of care, family health history (FHH) continues to be recognized as a baseline genetic screen for care, a ‘free’ risk assessment tool that has been underused by primary care providers [1–4]. Barriers to incorporating FHH in clinical care have been well characterized and include:

- Limited time to carefully collect and discuss FHH during the clinical encounter;
- Inadequate provider knowledge about, and confidence in, performing genetic risk assessment;
- Insufficient data to inform genetically based management for many conditions;
- The rapid pace of genomic discoveries that translate into patient care, exacerbating these issues [5–8].

Solutions using health information technology to address these barriers remain elusive. Although data standards and models exist [10], there is no uniformly accepted approach to capturing FHH data in personal or electronic health records (EHRs), enhancing the value of the data using clinical decision support (CDS) or exchanging information between systems (interoperability) [9,10]. Additional challenges include the feasibility of collecting and using FHH in the context of the US healthcare system, which places constraints on time, does not focus on prevention and, currently, does not reimburse appropriately for this activity [7].

FHH that includes ethnic background has long been recognized as valuable in assessing risk for Mendelian diseases in the preconception and prenatal periods, leading to support among public health agencies and professional societies for genetic screening for autosomal recessive conditions such as cystic fibrosis, Tay–Sachs disease, sickle-cell disease and others [11–16]. Research has also established FHH as a means to identify risk for chronic conditions with significant public health impact, including cardiovascular disease, diabetes and cancer [17,18]. Encouraged by the prospect of reducing the burden of these diseases in the USA, federal agencies, including the CDC, have recommended the use of FHH as a public health screening tool for various common chronic conditions [102,103], leading to public health initiatives, targeting providers and consumers, to encourage the use of FHH in disease prevention [3,19,20].

To establish the utility of FHH in public health, further research must address the ability of FHH information to identify disease accurately and reliably in an individual’s relatives (analytic validity), stratify and predict disease (clinical validity), or improve patient outcomes given effective interventions (clinical utility) [3,21,22]. A 2009 NIH State-of-the-Science Conference on Family History and Improving Care reinforced the need for direct evidence describing the accuracy and predictive value of familial risk data for common chronic conditions, as well as perinatal and pediatric outcomes [23].
collaborative efforts of investigators and stakeholders in the NIH, government agencies, academia and clinical research continue to gather evidence for the clinical utility of FHH for common chronic conditions such as heart disease, cancer and diabetes [24]. For some conditions, findings have been promising: Qureshi et al. recently demonstrated in a primary care setting that systematically collecting and integrating detailed FHH into a standard risk assessment protocol for cardiovascular disease increased the proportion of patients considered high risk by 4.5%, compared with standard practice [25].

**Genetic screening during the first prenatal visit**

While the scientific and medical communities continue to investigate the use of FHH as a public health tool, in practice, FHH remains an influential component of risk assessment for many perinatal conditions [16]. During the initial prenatal or preconception visit, the provider synthesizes health information from the patient, the father of the baby and the family to assess risks to the pregnancy – for example, risk of a heritable disorder, chromosomal abnormality or complex birth outcome – and risks to the patient herself, such as for osteoporosis or breast cancer [16].

In practice, however, the timing, level of detail and scope of conditions included for FHH risk assessment for a prenatal patient can vary depending on the patient population, clinic system and resources, and provider’s knowledge about clinically relevant genomic risk factors, screening tests and professional guidelines (American College of Obstetricians and Gynecologists [ACOG], American College of Medical Genetics [ACMG] and others). Such variation results in a lack of standardization in the application of these professional guidelines in perinatal care [26-27].

FHH tools that have been developed to assist the prenatal provider – for example, the first prenatal assessment and genetic evaluation tool [28] – are largely paper-based and face challenges in dissemination, maintenance of current content and integration into emergency health information technology systems. Medical care at all levels is in need of integrated data collection and risk assessment systems that employ standardized terms and machine-readable algorithms for sustainability and interoperability. Newer electronic FHH collection tools that enable data entry by patients and computerized risk assessment [29–31] typically address adult chronic diseases. With limited tools available to support the integration of FHH into prenatal and pediatric care, there remains an important unmet opportunity for electronic FHH risk assessment and intervention [32,33].

The product and approach we describe in this report had its genesis in programmatic efforts at the Health Resources and Services Administration (HRSA) to improve the application of genetic family history in the prenatal setting and throughout the lifecycle of the female patient. In particular, HRSA wished to support the primary care disciplines of family practice, obstetrics and gynecology, and nursing, all of which have a duty to understand the content needed and processes involved with obtaining a FHH. In cooperation with HRSA, our team developed an electronic FHH tool with CDS, the Pregnancy and Health Profile (PHP): a screening and risk assessment tool that is driven by patient-entered data. The tool, for use in primary care offices during the first prenatal care visit, aims to support FHH collection and assessment at the point of care by identifying patients at increased risk for genetic conditions that would require appropriate follow-up.

PHP, which is free to download, features an electronic patient questionnaire, a clinician management system, CDS in the form of qualitative risk assessment and management considerations for the provider, and point-of-care educational materials for the patient and provider [104-107]. **Figure 1** presents screenshots from the patient questionnaire, while **Figure 2** demonstrates a proposed clinical flow based on components of the tool.

One of the first challenges in developing any CDS tool is to determine the conditions whose inclusion could improve clinical outcomes. The remainder of this report focuses on the process by which we identified the conditions for inclusion with CDS, those for inclusion without CDS and conditions excluded from the tool (**Figure 3**).

**Identifying candidate conditions**

We identified possible conditions for inclusion in the PHP by:

- Examining professional guidelines from ACOG, ACMG, the National Society of Genetic Counselors, the American Academy of Pediatrics and other national guideline setting organizations to identify conditions for which there are recommendations to incorporate disease-specific FHH screening in primary prenatal care;

- Searching the published literature database PubMed using keywords (e.g., ‘family history’, ‘family health history’, ‘prenatal
Reviewing available prenatal FHH and genetic screening tools (Table 1) from professional organizations [108,109], foundations [110] and commercial groups [111,112] to determine the types of FHH information prenatal providers are currently collecting and using [28].

Throughout the course of the project, we continued to monitor publications from professional societies for the emergence of new or updated guidelines.

Our review of professional society guidelines identified 25 conditions or categories of conditions (e.g., a panel for Ashkenazi Jewish conditions) for which family history can be used to screen for diseases in the fetus and/or adverse pregnancy outcomes (Table 2) [11,14,15,34–51]. Although ACOG and ACMG do not have practice guidelines for all of the same conditions, there is significant overlap. In addition, in all but two cases ACOG and ACMG practice guidelines were in agreement regarding provider practice. ACMG suggests widely offering spinal muscular atrophy carrier screening, whereas ACOG’s recommendation for carrier screening is based on a positive FHH [37,38]. ACMG suggests offering a panel of nine conditions more common in the Ashkenazi Jewish population, whereas ACOG recommends offering patients a panel of four conditions but suggests making the remaining five ‘available’ to patients [35,36]. Our literature search identified additional candidate conditions for which evidence exists to support FHH screening across the lifespan.
cardiovascular disease, depression and seizure disorder \cite{52–54}.

Our assessment of paper-based tools identified 79 additional conditions or symptoms to be considered for inclusion. Areas of overlap included conditions for which race/ethnicity is a risk factor (e.g., Tay–Sachs and sickle-cell anemia) and factors that affect FHH interpretation (e.g., consanguinity and adoption status). Areas of discordance included conditions that ranged from patient symptoms (e.g., headaches and patches of differently colored skin) to general classes of birth defects (e.g., limb defects).

We identified a total of 107 candidate conditions from professional society recommendations or prenatal screening tools (Box 1). For each candidate condition, we considered the following questions, adapted from Yoon and colleagues \cite{3}:

- Does the condition have a known genetic cause or contribution?
- Is FHH a good screening tool for the condition?
- Is an intervention available based on identification of increased risk?
- What is the public health impact of the condition?

We then grouped each condition into one of five categories based on the application of the above criteria (Box 1):

- Conditions that have significant genetic or FHH contributions and for which there are professional society guidelines to support screening in the prenatal period, using FHH;
- Conditions that have significant genetic or FHH contributions and for which screening is not specifically recommended for the prenatal period, but for which evidence and guidelines support screening across the female lifespan;
- Conditions that are not genetic, but which are routinely screened for during the prenatal period, according to professional society guidelines;
- Conditions that have significant genetic or FHH contributions and for which screening is not specifically recommended for the prenatal period, but for which evidence and guidelines support screening across the female lifespan;
- Conditions that are not genetic, but which are routinely screened for during the prenatal period, according to professional society guidelines;

\begin{figure}
\centering
\includegraphics[width=\textwidth]{figure2}
\caption{Clinical flow of the Pregnancy and Health Profile. (A) The patient completes the electronic questionnaire on a tablet computer in the waiting room before the first prenatal appointment. (B) Patient-entered data is wirelessly transmitted to the Pregnancy and Health Profile (PHP) tool database and automated risk assessment is performed. (C) The clinician prints and reviews the PHP report that includes genetic clinical decision support based on patient-entered data and a pedigree. The clinician reviews point-of-care educational materials, also generated with the report. (D) The patient and clinician discuss the risk assessment and determine a management plan during the clinical encounter. (E) Patient receives targeted and personalized educational materials. (F) The clinician documents the encounter and uploads the PHP report into the paper or electronic medical record. Patient history can be updated and risk assessment recalculated as needed. Images attributed as follows: doctor by Andrew McKinley, printer by James Fenton from The Noun Project \cite{114}.}
\end{figure}

\begin{figure}
\centering
\includegraphics[width=\textwidth]{figure3}
\caption{Deriving a set of conditions for the systematic collection and interpretation of family history during the first prenatal visit. CDS: Clinical decision support; PHP: Pregnancy and Health Profile.}
\end{figure}
Conditions that have genetic or FHH contributions and are not screened for, based on FHH, in the prenatal period because they lack professional society support or other evidence;

Nonspecific congenital conditions that lack sensitivity if used as a patient-reported screen.

After categorizing the conditions, we presented this final list of conditions and rationale for inclusion to the project’s advisory group for review.

Finalizing conditions with CDS: rationale for inclusion & exclusion
With input from the advisory group, we developed a patient-friendly FHH questionnaire to collect the necessary FHH data required for risk assessment for each condition of interest. We created machine-readable risk assessment algorithms and CDS messages for each condition, derived from professional society guidelines and recommendations. In the two instances where society guidelines differed, we deferred to the actions recommended by ACOG in our CDS algorithms and subsequent provider messages. External experts in clinical obstetrics and genetics reviewed each set of algorithms for accuracy and relevance to the primary care setting. The CDS messages that are supported by professional society guidelines include clinical actions, such as ordering a screening test (e.g., Fragile X carrier screening), referring to a specialist (e.g., maternal fetal medicine and genetics) and providing patient counseling or education.

Categories 1 & 2
We included in the tool 27 conditions with CDS for which we identified guidelines and/or evidence that supported use of FHH as a screen for disease in the prenatal period and across the female lifespan (see Box 1 & Table 3, categories 1 and 2). In addition to the conditions originally identified through professional society and national guidelines, we also included cardiovascular disease, hemophilia and seizure disorder, conditions for which FHH is known to confer risk and carries important management implications during pregnancy or infancy [44,52,53]. For instance, a FHH of hemophilia conveys similar bleeding risks as von Willebrand disease, for which professional guidelines do recommend screening, and is a risk factor for having a male child with a severe bleeding disorder [44].

Table 1. Paper-based prenatal genetic screening tools in 2009.

<table>
<thead>
<tr>
<th>Publishing organization</th>
<th>Screening tools</th>
</tr>
</thead>
<tbody>
<tr>
<td>ACOG</td>
<td>Antepartum Record, version 6</td>
</tr>
<tr>
<td>ACOG</td>
<td>Obstetric Medical History, version 2</td>
</tr>
<tr>
<td>ACOG</td>
<td>Obstetric Women’s Health Record, 2005</td>
</tr>
<tr>
<td>American Medical Association</td>
<td>Prenatal Screening Questionnaire</td>
</tr>
<tr>
<td>Genzyme Genetics</td>
<td>Prenatal Genetic Screening Questionnaire</td>
</tr>
<tr>
<td>March of Dimes</td>
<td>Preconception/Prenatal Family Health History Questionnaire</td>
</tr>
<tr>
<td>Foundation for Blood Research</td>
<td>First Prenatal Assessment and Genetic Evaluation</td>
</tr>
</tbody>
</table>

ACOG: American College of Obstetricians and Gynecologists.

Table 2. Twenty five conditions addressed in professional guidelines for prenatal providers for which family health history can be used to screen.

<table>
<thead>
<tr>
<th>Mendelian or complex</th>
<th>Conditions impacting pregnancy and women’s health</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mendelian congenital (n = 8)</td>
<td>Ashkenazi Jewish-associated diseases, cystic fibrosis, Fragile X syndrome, maternal phenylketonuria, sickle-cell disease, spinal muscular atrophy, Tay–Sachs disease and thalassemia</td>
</tr>
<tr>
<td>Mendelian pregnancy and lifespan (n = 4)</td>
<td>Thrombophilia, von Willebrand disease, hereditary breast and ovarian cancer, and Lynch syndrome</td>
</tr>
<tr>
<td>Complex congenital (n = 7)</td>
<td>Autism, consanguinity, congenital and early-onset hearing loss, congenital and early-onset vision loss, congenital heart defect, intellectual disability and neural tube defects</td>
</tr>
<tr>
<td>Complex pregnancy and lifespan (n = 6)</td>
<td>Hypertension, osteoporosis, preterm birth, recurrent pregnancy loss (≥2), sudden death and Type II diabetes</td>
</tr>
</tbody>
</table>
Box 1. Conditions and syndromes (n = 107) evaluated for inclusion in the Pregnancy and Health Profile.

### Category 1 conditions or groups of conditions (n = 20), included with CDS
- Ashkenazi Jewish disease risk
- Autism
- Congenital or early-onset blindness
- Congenital or early-onset deafness
- Congenital heart defect
- Consanguinity
- Cystic fibrosis
- Fragile X syndrome
- Hemophilia/von Willebrand disease
- Intellectual disability (formerly known as development delay and mental retardation)
- Neural tube defect
- Recurrent pregnancy loss (≥2)
- Preterm birth
- Phenylketonuria
- Sickle-cell disease
- Spinal muscular atrophy
- Sudden unexpected death
- Tay–Sachs disease
- Thalassemia
- Thrombophilia

### Category 2 conditions (n = 7), included with CDS
- Cardiovascular disease
- Depression (bipolar and unipolar)
- Diabetes
- Hereditary breast and ovarian cancer syndrome
- Hypertension
- Lynch syndrome
- Osteoporosis

### Category 3 conditions (n = 22), included without CDS
- Alcohol use
- Allergies
- Blood transfusion
- Cigarette use or smoking exposure
- Consumption of raw meat
- Epilepsy or seizures
- Exercise frequency
- Hospitalizations
- Illicit drug use
- Infectious diseases (HIV, CMV and others)
- Infertility
- Medication use
- Past pregnancies
- Religious objections to medical treatment
- Seat belt use
- Sexually transmitted diseases
- Thyroid disease
- Urinary tract infections
- Vaccination status
- Violence, physical or sexual abuse
- Workplace exposures (lead, mercury and others)
- X-ray exposure

### Category 4 conditions (n = 20), excluded
- Achondroplasia
- α-1-antitrypsin deficiency
- Alzheimer’s disease or other form of dementia
- Cataracts
- Chromosomal abnormality (other)
- Cleft lip and/or cleft palate
- Congenital hip dislocation
- Crouzon syndrome
- Down’s syndrome
- Dwarfism
- Genital or urinary tract defects
- Huntington’s disease
- Hydrocephalus
- Marfan syndrome
- Muscular dystrophy
- Neurofibromatosis
- Neuromuscular disease
- Polycystic kidneys
- Scoliosis
- Turner syndrome

Conditions in category 1 and 2 are included with CDS, those in category 3 are included as personal risk factors only, without the tool providing decision support recommendations, and categories 4 and 5 were excluded from the tool.  
†Includes screening for Tay–Sachs disease, cystic fibrosis, Canavan disease and familial dysautonomia, and may include screening for mucolipidosis IV, Niemann–Pick disease Type A, Fanconi anemia group C, Bloom syndrome and Gaucher disease [36].  
‡Defined as occurring at 40 years of age or younger.  
§Defined as occurring at 40 years of age or younger.  
Includes screening for some conditions in category 3 in the current release of the tool.  
CDS: Clinical decision support; CMV: Cytomegalovirus.
### Category 3

The complete list of data collected in the PHP expands beyond genetics and FHH to include risk factors commonly reviewed during the initial prenatal visit and that may be flagged in the patient report. The realities and infrastructure of everyday clinical practice necessitated this expansion of the scope of the electronic questionnaire. For a FHH tool to be incorporated into the prenatal setting and be accepted by primary care providers, questions pertaining to FHH and genetic risk must be integrated with those that are part of routine prenatal care, including personal health history, past pregnancy history and exposure to potential teratogens. Such environmental risk factors, in addition to social and cultural ones have a significant impact on pregnancy outcomes. We identified these risk factors based on a discussion with clinicians in our advisory group and from collaborating prenatal care clinics, followed by a review of existing paper-based screening questionnaires with an emphasis on the ACOG Antepartum Record. Although we elected to include screening questions for these category 3 conditions in the tool to streamline the prenatal intake process, we did not, however, create CDS because management of these risk factors is part of routine obstetric care and beyond the educational objectives of our FHH screening tool (see Box 1 & Table 3, category 3).

### Category 4

Screening questionnaires also included different lists of rare genetic syndromes and specific birth defects with known genetic contributions. However, many of these rare conditions lacked demonstrated clinical utility for prenatal screening for the general population. We did not include such conditions in the tool (see Box 1 & Table 3, category 4); instead, the questionnaire allows the patient to type in a free-text response to

---

**Box 1. Conditions and syndromes (n = 107) evaluated for inclusion in the Pregnancy and Health Profile (cont.).**

<table>
<thead>
<tr>
<th>Category 5 conditions (n = 38), excluded</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Anemia</td>
</tr>
<tr>
<td>- Arthritis</td>
</tr>
<tr>
<td>- Asthma</td>
</tr>
<tr>
<td>- Attention-deficit/hyperactivity disorder</td>
</tr>
<tr>
<td>- Autoimmune disorder/lupus</td>
</tr>
<tr>
<td>- Birth defect (other)</td>
</tr>
<tr>
<td>- Birth defect of an arm or a leg (including extra or missing figures or toes)</td>
</tr>
<tr>
<td>- Birth defects of the bowels or intestines</td>
</tr>
<tr>
<td>- Bowel disease</td>
</tr>
<tr>
<td>- Cataracts</td>
</tr>
<tr>
<td>- Chronic or significant skin problems</td>
</tr>
<tr>
<td>- Deformities</td>
</tr>
<tr>
<td>- Dental problems (missing, extra or abnormally formed teeth)</td>
</tr>
<tr>
<td>- Eating disorder</td>
</tr>
<tr>
<td>- Excess of broken bones</td>
</tr>
<tr>
<td>- Fibroids</td>
</tr>
<tr>
<td>- Glaucoma</td>
</tr>
<tr>
<td>- Headaches</td>
</tr>
<tr>
<td>- Kidney disease</td>
</tr>
<tr>
<td>- Limb defects (extra or missing digits, malformed arms, legs, hands or feet)</td>
</tr>
</tbody>
</table>

- Large/small/ unusually shaped head
- Muscle weakness or inability to walk/poor coordination
- Other hemoglobin abnormality
- Other nerve, muscle or seizure disorder
- Patches of differently colored hair
- Patches of differently colored skin
- Primary amenorrhea
- Pyloric stenosis
- Rashes (part of descriptors)
- Reflux/hiatal hernia/ulcers
- Respiratory disease or chronic lung condition/pneumonia/lung disease
- Serious medical condition in infancy or childhood
- Slow learner
- Speech problems (e.g., significant delay)
- Unusual shape/size/position of ears
- Unusually formed bones or many broken bones
- Unusually formed hands or feet (including club foot)
- Very short or tall stature

Conditions in category 1 and 2 are included with CDS, those in category 3 are included as personal risk factors only, without the tool providing decision support recommendations, and categories 4 and 5 were excluded from the tool.

1. Includes screening for Tay–Sachs disease, cystic fibrosis, Canavan disease and familial dysautonomia, and may include screening for mucolipidosis IV, Niemann–Pick disease Type A, Fanconi anemia group C, Bloom syndrome and Gaucher disease [36].

2. Defined as occurring at 40 years of age or younger.

3. Epilepsy is included as an obstetric risk factor in category 3 in the current release of the tool.

CDS: Clinical decision support; CMV: Cytomegalovirus.
identify any of these conditions for the upcoming provider appointment. This enabled the tool to capture information about a particular family with a rare condition or symptom, while not requiring every woman to look through long lists of rare, unfamiliar conditions.

In our review of existing screening questionnaires, we also identified nonspecific conditions for which we did not identify professional society guidelines or literature recommending inclusion of the condition and that would not translate to a computerized and algorithm-based risk assessment tool. As an example, general categories of birth defects or physical anomalies (e.g., limb defects and patches of differently colored hair) may have genetic contributions and possibly confer an increased recurrence risk to family members. Without a more specific diagnosis, however, the validity of using such a condition for risk prediction in the patient or pregnancy is unknown. Therefore, no reliable recommendations for a medical intervention are possible. These nonspecific conditions or categories of conditions with unknown clinical validity and clinical utility were excluded from the tool at this time (see Table 5).

### Table 3. Categories of conditions included and excluded in the tool.

<table>
<thead>
<tr>
<th>Group</th>
<th>Category</th>
<th>Professional society support†</th>
<th>Decision</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Conditions with significant genetic or family history contributions (highly penetrant Mendelian conditions, as well as complex conditions)</td>
<td>ACOG guidelines recommend screening by the primary prenatal provider</td>
<td>Included in the tool with CDS</td>
<td>Hemoglobinopathies and Fragile X syndrome</td>
</tr>
<tr>
<td>2</td>
<td>Conditions with significant genetic or family history contributions (highly penetrant Mendelian conditions, as well as complex conditions)</td>
<td>Screening is not specifically recommended for the prenatal period, but evidence and guidelines support screening across the female lifespan</td>
<td>Included in the tool with CDS for later action (e.g., postpartum visit)</td>
<td>Hereditary breast and ovarian cancer syndrome, and osteoporosis</td>
</tr>
<tr>
<td>3</td>
<td>Conditions without significant genetic or family history contributions</td>
<td>ACOG guidelines recommend screening by the primary prenatal provider</td>
<td>Included in the tool as a risk factor without CDS†</td>
<td>Toxoplasmosis and alcohol exposure</td>
</tr>
<tr>
<td>4</td>
<td>Rare conditions with significant genetic or family history contributions (highly penetrant Mendelian conditions, as well as complex conditions), but without evidence of clinical utility of prenatal family history screening for all women</td>
<td>Lacking</td>
<td>Excluded in the tool</td>
<td>Neurofibromatosis, arthritis and lupus</td>
</tr>
<tr>
<td>5</td>
<td>Nonspecific congenital conditions that lack sensitivity if used as a patient-reported screen</td>
<td>Lacking</td>
<td>Excluded in the tool</td>
<td>Bowel disease and deformities</td>
</tr>
</tbody>
</table>

†Evidence cited by guidelines ranged from ‘Level A’ to expert opinion. †Includes conditions collected as part of past obstetrical history that is part of standard prenatal care.

ACOG: American College of Obstetricians and Gynecologists; CDS: Clinical decision support.
and the environmental, social and cultural factors that may affect personal and pregnancy outcomes.

The first release of PHP was developed to assist providers during the prenatal intake process in three important ways:

- Engaging the patient prior to the clinical encounter and enhancing the subsequent discussion during the clinical encounter (e.g., clarify responses as needed and update CDS);
- Providing point-of-care considerations for immediate and personalized patient management;
- Assisting providers with the continuous challenge of integrating evolving knowledge of genomics into prenatal care.

The tool was designed to be an adjunct to the clinician’s judgment and accounts for both the clinical and work flow needs of practices with or without EHRs. For practices with an established method for collecting FHH information electronically, PHP may serve as an EHR module since components of our tool, such as the structured approach to FHH data or CDS, may be integrated into systems that comply with Health Level 7 and American Health Information Community standards [9]. For practices without an electronic medical record system, the PHP can be implemented as a standalone system, which is used along with the paper medical record. We have pilot tested the tool in four clinical sites to evaluate the drivers and barriers to its implementation, acceptability, usability and satisfaction among both providers and patients, and the clinical impact of using the tool on provider behavior and management of selected genetic risks. We are currently evaluating these data and submitting our findings [Edelman E et al. Evaluation of a novel electronic genetic screening and clinical decision support tool in prenatal clinical settings (2013), Submitted; Edelman E et al. Clinical implementation of the pregnancy and health profile (2013), Manuscript in preparation; Lin BK et al. Patient perspectives from using an electronic family health history tool in a prenatal care setting: usability, confidentiality, and preferences (2013), Manuscript in preparation].

Conclusion & future perspective

Electronic FHH tools, such as the PHP, will become essential to keep providers up to date, personalize patient care and develop evidence related to the clinical validity and utility of FHH in the prenatal care setting. Large-scale observational studies that include FHH can build upon the existing evidence for established conditions, or develop a new knowledge base for conditions or symptoms where the evidence for clinical validity or clinical utility is currently lacking (Table 3). For instance, to address the serious problem of preterm birth (PTB), which runs in families, and where recurrence is high among women with a previous spontaneous singleton PTB, a study may aim to:

- Develop a risk assessment algorithm, using a scoring system that codifies risk based on FHH and personal risk indicators for PTB;
- Enhance the PHP by stratifying the patient’s risk (e.g., average, above average and high) and suggesting personalized interventions;
- Qualitatively characterize PTB susceptibility by piloting the risk algorithm in a diverse obstetrical patient population.

Future study of the PHP in clinical practice will include calculation of the clinical validity of patient-entered FHH data in the PHP in predicting perinatal outcomes such as PTB, gestational diabetes, pre-eclampsia and postpartum depression. Additional research is needed to understand the validity of patient-entered FHH for prenatal CDS and the ability of CDS systems to improve the effectiveness of personalized FHH or genomic risk assessment tools [55–58].

As the FHH evidence base develops, new or revised guidelines that inform the standard of care in prenatal primary care practices will need to be written to accommodate CDS algorithms. Currently, the March of Dimes Foundation, as a codeveloper of PHP, already monitors emerging perinatal guidelines and literature as part of its effort to improve the health of infants and children, and will recommend to the collaboration whether CDS needs to be updated or introduced. However, in the future, as genomic CDS becomes more common, professional societies or a governing body may opt to maintain an application or web service structure to store or quickly disseminate new CDS as their guidelines change or are created.

Future applications of electronic FHH tools should incorporate the complex relationship of genetic, environmental, social and cultural factors into risk assessment, for both prenatal care as well as the lifespan of the individual and his or her family. As personalized medicine and genomic screening continues to advance, FHH will play an increasingly important role in risk
We determined a set of core conditions that could be addressed by the electronic tool based on existing criteria: conditions with
9(3), An electronic tool that allows patient entry and provides clinical decision support is one solution for practitioners who need a
Am. J. Prev. Med. 19(3), 273–280 Family health history (FHH) remains the first screening tool available to primary care providers at the point of care to personalize risk for
Numerous organizations have previously developed paper-based tools that screen for inherited conditions in the prenatal setting.
We excluded conditions or symptoms for which FHH data have no clinical utility. Entry of free text by the patient is possible to alert
'The Pregnancy and Health Profile: a screening and risk assessment tool' is freeware for primary prenatal providers.
We developed a panel of 27 conditions for which FHH data can be used for risk assessment and to establish a personal, genetic
A review of professional society guidelines, the literature and existing prenatal screening tools identified 107 conditions that could be
We determined a set of core conditions that could be addressed by the electronic tool based on existing criteria: conditions with
Family health history is already a screening tool for prenatal care
• Family health history (FHH) remains the first screening tool available to primary care providers at the point of care to personalize risk for their patients.
• Numerous organizations have previously developed paper-based tools that screen for inherited conditions in the prenatal setting.
• An electronic tool that allows patient entry and provides clinical decision support is one solution for practitioners who need a
time-sensitive method of collecting and interpreting FHH data for prenatal care.
Identifying conditions for a family history & a genetic screening tool for prenatal providers
• A review of professional society guidelines, the literature and existing prenatal screening tools identified 107 conditions that could be included for prenatal FHH assessment.
• We determined a set of core conditions that could be addressed by the electronic tool based on existing criteria: conditions with known genetic contribution; utility of FHH information for screening; whether an intervention can be offered based upon identification of an at-risk individual; and public health impact of the condition.
Finalizing conditions with clinical decision support
• We developed a panel of 27 conditions for which FHH data can be used for risk assessment and to establish a personal, genetic problem list with considerations for management.
• We excluded conditions or symptoms for which FHH data have no clinical utility. Entry of free text by the patient is possible to alert providers about rare, inherited risks.
Standard panel of conditions for genetic screening & risk assessment during prenatal intake
• The level of evidence for conditions that met our criteria for inclusion and for which professional society guidelines exist varied from good and consistent scientific evidence (level A) to expert opinion.
• 'The Pregnancy and Health Profile: a screening and risk assessment tool' is freeware for primary prenatal providers.

References
Papers of special note have been highlighted as:

9 Fick W, Bigley MB, Briner KM. New standards and enhanced utility for family health history information in the electronic health record: an update from the American Health Information Community’s Family History Multi-Stakeholder

* Describes the rationale used to develop a core data set for FHH information that will promote risk assessment using FHH and interoperability among systems.


* Takes a lifecourse perspective in describing how FHH plays a role in assessing risk for numerous conditions ranging from single-gene disorders to complex birth outcomes.


* Review of studies that examine FHH as a risk factor for common chronic conditions. Presents a framework for how to assess FHH’s impact as a public health screening tool.


* With an emphasis on common chronic conditions, this report reviews the evidence base and gaps needed to establish the effectiveness of FHH as a public health screening tool.


# Websites


105 National Coalition for Health Professional Education in Genetics (2013). www.nchpeg.org


111 Genzyme Genetics. Prenatal Genetic Screening Questionnaire. www.genzymegenetics.com

Since our review, LabCorp is the new parent company and the Genzyme Genetics Questionnaire is no longer available online. A similar family history questionnaire, which now includes obstetric risk factors, is available through the LabCorp Specialty Testing Group Integrated Genetics website.

