

# Family History in Pediatric Primary Care

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## ABSTRACT

The family history is a critical element in pediatric medicine and represents the gateway to the molecular age of medicine for both pediatric clinicians and their patients. The pediatric clinician has several opportunities to obtain a family history and multiple clinical and educational uses for that information. Available methods include paper and digital forms, classical pedigrees, online programs, and focused family history at the time of a new diagnosis or problem. Numerous barriers impede the application of family history information to primary pediatric practice. The most common barrier is the limited amount of time the typical primary care encounter allows for its collection. The family history can be used in many facets of pediatric practice: (1) as a diagnostic tool and guide to testing and evaluation; (2) to identify patterns of inheritance; and (3) as a patient-education tool. The most exciting future use of family history is as a tool for public health and preventive medicine. More accurately identifying children at risk for common chronic conditions such as diabetes, asthma, and cardiovascular disease could change the primary care clinician's approach to pediatric medicine.

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### Key Words

family history, pediatric primary care

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**A**S CLINICIANS, WE are living and practicing in the molecular age of medicine. To use the exciting products of the Human Genome Project, pediatric clinicians must be able to identify those patients or families for whom testing or intervention would be beneficial. An accurate family history represents the gateway to the molecular age of medicine for primary care pediatrics. In this review we summarize current practices, current and future uses for this information, barriers to collection of this information, and possible solutions for achieving the goal of having a pedigree in every chart.<sup>1</sup>

The family history represents the most traditional diagnostic tool in clinical genetics and remains an inexpensive approach to identifying individuals at risk for genetic disorders.<sup>2,3</sup> Also, family history information is increasingly being used for complex common conditions for which the genetic etiology is unknown. The family history is defined as the description of the genetic relationships and medical history of a family; when it is represented in diagram form using standard symbols and terms, it is referred to as a pedigree.<sup>3</sup> As the recognition of the importance of genetic factors in the causation of human disease grows,<sup>4</sup> the family history becomes not only a tool for providing appropriate care for specific diseases but also a tool for public health and preventive medicine.<sup>5</sup>

The goal of universal inclusion of family history in the medical chart is formidable. As the director of the National Human Genome Research Institute, Francis Collins, MD, PhD, noted, "This 'next revolution of medicine' will fall on the shoulders of physicians who provide primary care."<sup>6</sup> Although many primary care clinicians already incorporate genetic screening into their routine services, the demands on clinicians are increasing substantially with the growing need to provide information on new genetic tests to their patients, help interpret test results, and consider prescribing new genetic therapies as they become available.<sup>7</sup> To illustrate why the clinician must become the point person in this genetic information revolution, we need only to review the 2003 survey of the American Board of Medical Genetics, which addressed the state of the medical geneticist workforce. The survey identified 882 clinical geneticists in the United States who reported devoting an average of 30% of their professional time to direct patient care. There are additionally 1399 certified genetic counselors who devote an average of 70% of their time to direct patient care.<sup>8,9</sup> Because "all patients have genes,"<sup>10</sup> it becomes obvious that although medical geneticists and genetic counselors are the experts in the clinical use of genetic tests for the diagnosis and management of heritable disorders, it is clearly the primary care clinician who must identify and guide those patients who will benefit from their expertise.

## CURRENT PRACTICES

Family history has always been important in health care.<sup>11</sup> Families share genetic susceptibilities, environments, and behaviors, all of which interact to cause different levels of health and disease.<sup>12</sup> Family history can help identify conditions that are predominantly genetic, as well as those that might be the result of gene-environment interactions. The opportunity to use family history to focus on prevention and earlier intervention has grown with the expanding genetic knowledge gleaned from the mapping of the human genome. Pediatric clinicians (physicians, nurse practitioners, and physician's assistants) are particularly well positioned to use this knowledge, because they provide primary care to an individual from birth to adulthood, the period during which most genetic disorders will become manifest. Also, medical genetics has traditionally been incorporated into their overall practice. The challenge is to make the collection of a family history with annual updates as standard as collecting and updating information on a child's immunizations.

In general, clinicians do ask about family history in the context of routine physical examinations,<sup>13</sup> but they need to do a better job collecting an initial family history<sup>14-17</sup> and updating the history annually.<sup>18,19</sup> Clinicians use family history for a variety of purposes: to establish patterns of inheritance, assist with medical diagnoses, identify risk, provide appropriate screening, target education and preventive efforts, and inform counseling for preconception health care.<sup>3</sup> The family history also helps to establish rapport with patients and families, understand familial relationships, and identify shared environments that may predispose one to risk.

Family history information is considered one of the key elements in a new or return patient visit for a complete physical examination and must be collected to warrant billing for that encounter.<sup>20</sup> Pediatric and adolescent medicine textbooks provide varying recommendations for the collection of family history information from the very general<sup>21,22</sup> to the specific.<sup>23</sup> The Genetics in Primary Care Family History Working Group, a program that is funded by the Health Resources and Services Administration and has been ongoing since 1998, has created an easy-to-use mnemonic for family history collection for the clinical setting entitled SCREEN (Table 1).<sup>24</sup> If there were significant findings from this initial screen, more in-depth information or a family pedigree could then be obtained. As a mnemonic, its main benefit is to remind the clinician to collect some limited, key family history information; it is not intended to be used as a tool for collection of a complete family history.

The pedigree, the gold standard for the collection of family history, is a visual way to enhance recognition of patterns of inheritance.<sup>2,25</sup> Pedigrees have the advantage of standardized nomenclature and structure, but their use might not be feasible for many primary care prac-

**TABLE 1** The SCREEN Mnemonic for Family History Collection

SC	Some concerns	"Do you have any (some) concerns about diseases or conditions that run in the family?"
R	Reproduction	"Have there been any problems with pregnancy, infertility, or birth defects in your family?"
E	Early disease, death, or disability	"Have any members of your family died or become sick at an early age?"
E	Ethnicity	"How would you describe your ethnicity?" or "Where were your parents born?"
N	Nongenetic	"Are there any other risk factors or nonmedical conditions that run in your family?"

tices,<sup>26</sup> and some clinicians might not feel capable of creating or accurately interpreting a pedigree.<sup>27,28</sup> As one approach, genetic counselors often start the history collection over the telephone, before the initial appointment, so that they and the families can begin the research needed for a complete pedigree.<sup>2</sup> Although this approach might be less practical for primary care because of the time involved, the content of a 3-generation family history remains the important factor regardless of whether an actual pedigree is drawn.

No matter which tool or form a practice uses, clinicians have limited time to collect family history in the context of a preventive care visit. The average duration of time spent with a patient during a visit is 15.2 minutes for pediatricians and 17.9 minutes for family practitioners.<sup>29</sup> According to a study by Acheson et al,<sup>18</sup> the amount of time spent by family physicians on family history discussions averaged 2.6 minutes for a new patient and 1.8 minutes for a return patient. The challenge for clinicians is to collect a useful family history in a brief period of time.

Pediatric clinicians have an advantage over adult clinicians in that children are seen more frequently for preventive care visits, especially in the first 2 years of life. If the periodicity recommended by the American Academy of Pediatrics is followed,<sup>30</sup> a child will be seen for 10 visits in that time period, which provides multiple opportunities to obtain and expand a family history. An extensive family history is recommended at the first visit, and an assessment of interval change is recommended at each subsequent visit.

The conditions that are the focus of interest can shift depending on the developmental stage of the child. For example, a family history of developmental delay might be the focus during the infant stage, but interest might shift to a family history of depression when the child is an adolescent. Creation of a 3-generation pedigree for a child and his or her family could provide years of prevention, risk reduction, and early identification if the pedigree were updated regularly and shared with the family. A recent survey of people with a family history of cancer suggests that detailed family histories of younger patients (starting at 18 years of age) might detect those at higher risk for a hereditary cancer syndrome.<sup>31</sup> When a particular condition or syndrome is identified or suspected, disease-specific family history tools are available to direct and extend collection of family history information.<sup>3</sup>

Parents can facilitate the collection of family history information through the use of Internet sites such as the US Surgeon General's My Family Health Portrait.<sup>32</sup> This site allows a user to create a simple pedigree that is kept on the user's computer and is not part of any central database. Web-based tools are becoming more popular and reflect the trend toward electronic medical charts as well as consumer-driven preferences. Computer-savvy children could work with their parents to create their own pedigree to bring to their preventive visit. Families can be encouraged to collect family history information from relatives on Thanksgiving Day, which was designated as Family History Day by former Surgeon General Richard Carmona because it is a day when families repeatedly come together in a multigenerational manner.

#### BARRIERS

The main barrier to collecting family history in the clinical setting is time.<sup>27,33</sup> Time constraints might also be a factor in the lack of updated<sup>18,19</sup> and detailed<sup>33</sup> family histories. In a study of maternal history of depression, a condition that can have a significant effect on children, 70% of pediatricians felt that they had inadequate time to inquire about aspects of family history.<sup>34</sup> In addition, families might not know their history or might not have access to family members. Many parents are working full-time outside the home and might lack the time, support, or motivation to obtain the information. Another potential barrier is that pedigree construction can be emotional as families remember loss, divorce, illness, or fractured family relationships. Children might have multiple family members caring for them without a single adult assuming primary responsibility. Given these barriers, an active partnership between clinicians and parents is necessary to create a useful family history, and resources must be available and shared.

#### EXAMPLES OF CURRENT USES OF FAMILY HISTORY INFORMATION

The family history is a critical factor in many facets of pediatric practice. It has traditionally been used as a diagnostic tool and as a guide to evaluation and treatment in single-gene disorders, and it is increasingly becoming important in the arenas of patient education, public health, and preventive medicine.

A recent case from Dr Trotter's pediatric practice illustrates the power of the family history to direct medical evaluations and genetic testing, aid in diagnosis, and

provide appropriate counseling for a family. The case involved a 4-year-old girl who had been diagnosed with autism spectrum disorder at 18 months of age. When she was seen by her primary care pediatrician for her 4-year well-child visit, her family history was updated, as recommended by the American Academy of Pediatrics. At that time, her mother related that in the previous 6 months her sister's 2 sons had been diagnosed with fragile X syndrome. This new history prompted molecular genetic testing specific for *FMRI*, the gene that is responsible for fragile X syndrome, which led to the subsequent diagnosis of fragile X syndrome for this patient. Additional testing of the family revealed both the mother and an older sister as premutation carriers. For this family, the family history focused the evaluation and ultimately led to the correct diagnosis and provided a basis for both prognostic and reproductive counseling.

Additional clinical scenarios commonly dealt with by pediatric clinicians further illustrate use of the family history as a diagnostic tool. For instance, in evaluating an infant with several café-au-lait spots, the knowledge that one of the child's parents has neurofibromatosis will clarify screening tests and follow-up for the infant. Likewise, awareness that a young adolescent girl with severe or prolonged vaginal bleeding at menarche has a father with von Willebrand disease will substantially guide the workup.<sup>11</sup>

A family history also helps to identify patterns of inheritance and can aid in a diagnostic workup, such as the evaluation of a boy with mental retardation in a family with a pattern of X-linked recessive mental retardation, even when no other family members carry a known diagnosis. The extension of this information is to use inheritance patterns to counsel other family members, quantify risk assessment for other relatives, and assess reproductive options.<sup>3</sup> Inheritance patterns can be explained visually by using a standard pedigree, and the variability of expression can be similarly demonstrated. This is especially useful when making a diagnosis or presumptive diagnosis at an early age. It would be important for parents to know that their toddler with presumed neurofibromatosis might have only a dozen café-au-lait spots and a few neurofibromas like her father or might develop an optic nerve glioma and severe scoliosis like her aunt.

Another major use of a 3-generation family history is as a patient-education tool. Collection of family history information can demonstrate to patients the need for medical documentation of relatives' histories and sharing of family history information with relatives. For example, did your uncle die of a myocardial infarction or of a fatal arrhythmia? Accurate documentation could be vital to a possible diagnosis of long QT syndrome in the family. Once a diagnosis of long QT syndrome is confirmed, parents can then be educated regarding signs and symptoms to be aware of in family members, such as

near-sudden infant death syndrome, syncope, and aborted cardiac arrest, and can be counseled regarding their child's participation in athletics or exercise programs that might increase the risk of an event.

The family history can also be used to clarify misconceptions that families have. Commonly, families erroneously assume that certain genetic diseases affect only one gender, because thus far, for example, only men have been affected in their family. Another common misconception is that a disease "skips" generations. This is often a result of incomplete penetrance, variability of expression, or misdiagnosis. Correcting these misunderstandings by using a simple pedigree can be invaluable to all the family members and often leads to expanded opportunities to explore levels of understanding and facilitate patient and family education.

#### **FUTURE USES**

The newest and potentially most exciting use of the family history is as a tool for public health and preventive medicine. In 2002, the Office of Genomics and Disease Prevention at the Centers for Disease Control and Prevention formed a multidisciplinary working group and developed a research initiative to explore these possible uses for family history information.<sup>5</sup> This cause was further advanced in 2004 when US Surgeon General Dr Richard Carmona announced the inaugural National Family History Day in the November 25, 2004, edition of the *New England Journal of Medicine*.<sup>11</sup>

Most diseases are the result of the interactions of genes and environmental factors. Although these interactions are complex, almost every patient today has access to a free, well-proven, personalized genomic tool that captures many of these interactions and can serve as the cornerstone for individualized disease prevention. This valuable tool is the family history.<sup>10,11,35</sup> The Valdez et al article (p S78) more completely explores this exciting and valuable use of the family history for identifying children at risk of common chronic conditions such as diabetes and cardiovascular disease.

#### **STRATEGIES: PRACTICAL CLINICIAN POINTS**

- Choose a family history tool that works for you, your patient, and your setting.
- The National Coalition for Health Professional Education in Genetics has a summary of available family history tools listed at [www.nchpeg.org/newsletter/inpracticespr05.pdf](http://www.nchpeg.org/newsletter/inpracticespr05.pdf).
- The March of Dimes has developed a preconception/prenatal family history questionnaire that is available at [www.marchofdimes.com/pnhec/4439\\_1109.asp](http://www.marchofdimes.com/pnhec/4439_1109.asp).
- Families can create their own simple pedigree with the US Surgeon General's My Family Health Portrait, which is available at <https://familyhistory.hhs.gov>.



- Introduce families to resources during their first visit.
- Add family history links to your office or clinic Web site, if available.
- Provide handouts with resource information.
- Print out fact sheets from Web sites.
- Publicize Thanksgiving Day as Family History Day.
- Take advantage of frequent well-child visits to complete a family history.
- Post reminders or create slogans for clinicians and families, such as “Five Minutes for Family History” or “Don’t Forget Family History.”
- Involve the kids by using a computer.
- Involve the parents by obtaining a maternal prenatal family history.
- Review and update family history annually.

## CONCLUSIONS

All pediatric primary care practitioners should have the basic competencies to collect and interpret a family medical history. Obtaining a family history remains an inexpensive and basic approach to identifying individuals at risk for genetic disorders. For adults, family history has also been shown to be an accurate way to approach risk identification for common complex disorders such as cardiovascular disease, diabetes, several cancers, asthma, and stroke.<sup>35</sup> Family history is a way to reach those at higher risk and to target resources to get them into screening.<sup>36</sup> A family history can establish patterns of inheritance and serve as a guide to diagnostic, therapeutic, and preventive approaches.

Pediatric clinicians are familiar with genetics in their patient population and have the advantage of frequent well-child visits for collecting and updating family history. A variety of tools exist to assist clinicians and patients in improved collection of these data. Pediatric clinicians will need to tailor the family history tool they choose to the strengths and challenges of their practice setting and their patient population. Numerous barriers impede the application of family history information to primary pediatric practice. New tools will be required to assist pediatric clinicians with the efficient collection and application of family history information in the era of genomic medicine.

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