PS#87 – Family History Taking and Genetic Testing in Pediatric Practice

PS#87 was initiated by the Committee on Genetics and members of the AAP Genetics in Primary Care Institute (GPCI) to explore pediatricians’ knowledge, attitudes, practices and barriers surrounding family history taking, genetic testing and management of children with genetic conditions. Partial funding for this survey was provided by the Genetics in Primary Care Institute (GPCI), HRSA, MCHB grant #UC7MC21713.

PS#87 was an eight-page questionnaire mailed to a random sample of 1,627 nonretired U.S. members of the AAP from December 2013 to June 2014; response to the survey was 43% (705 respondents, including pediatric residents). Analysis is limited to 629 pediatricians who provide patient care.

Findings:

Experiences and Opinions on Family History Taking

Overall, 95% of pediatricians collect family health history information with at least some of their patients:

- Eight out of 10 pediatricians say they collect a family history with most or all (>76%) new patients; 29% do so with most/all established patients.

- Nearly all pediatricians (91%) say they collect information on second degree relatives, while only 28% collect information on third degree relative.

- Most pediatricians (80%) report that family history information is collected in the exam room; 16% say it is collected in the waiting room. Sixty-four percent of pediatricians report they personally take the family history verbally from the patient, 15% say a nurse or other staff collects this information, and 20% say the a parent completes a form.

- One-half of pediatricians (49%) use a standardized disease checklist to take the family history and 41% ask open-ended questions.

- Nearly all pediatricians who collect family health histories do so on disorders relevant to the family’s concerns or clinical findings (94%); 75% ask about disorders relevant to the child’s age-based health, while 54% inquire about a range of disorders, including adult-onset disorders not immediately relevant to the child’s age-based health.

- More than 80% of pediatricians say they feel confident in their ability to discuss the results of a positive family history with parents (87%) and to determine the need for further evaluation based on those results (83%).

- The main barrier to collecting or updating a family history is lack of time during a typical office visit (61% of pediatricians say this is a moderate or significant barrier); 40% report uncertainty as to the accuracy or reliability of the information collected from parents as a moderate/significant barrier. Other barriers, such as lack of an EHR system to collect and record family histories, lack of questionnaires to obtain family health histories, inadequate insurance reimbursement for collecting family histories, and language or other cultural differences between pediatrician and patient, were reported as moderate/significant by less than one-fourth of pediatricians.
Genetic Testing in Pediatric Practices

- Most pediatricians (72%) have had at least 1 patient identified as at-risk for a genetic related disorder during the 12 months prior to the survey; among those pediatricians, the average number of patients so identified is 10.8.
  - One half of pediatricians with at-risk patients say they referred a majority (>50%) of these patients to a geneticist or another specialist for tests without ordering tests themselves; 26% say they ordered genetic tests themselves and referred for tests the majority of their at-risk patients. Only 14% of pediatricians say they ordered genetic tests themselves but did not refer these at-risk patients to a geneticist or another specialist for tests.

- Nine out of 10 pediatricians say they have at some time referred a patient for genetic consultation.
  - Pediatricians named several factors as being very important in the decision to refer, including the complexity/severity of the disorder (71% so reporting), the desire for recommendations on management of the condition (67%), the ability to provide information about recurrence risks/family planning (60%), the availability of known treatments for the condition (58%), and parents’ desire for more information (58%). Few pediatricians identified the availability of insurance coverage for genetic consultation (20%) or malpractice concerns (7%) as very important factors in the decision to refer.

- Moderate to significant barriers to genetic testing/evaluation named by pediatricians include: lack of training and/or practice guidelines on genetic interpretation (59% so reporting), lack of guidelines for care management decisions once risks are identified (55%), lack of training in identifying genetic risks and choosing appropriate genetic tests (54%), and inadequate time during typical office visits to interpret genetic tests (48%). Limited access to geneticists/genetic counselors for testing and/or consultation, and inadequate insurance reimbursement for genetic tests were named as moderate/significant barriers by less than one-third of pediatricians.

- Pediatricians generally agree that genetic testing can provide helpful information about recurrence risks for other family members (91% strongly agree/agree) and that geneticists are better able than other providers to provide genetic testing and evaluation (87%), but that there are situations in which it is the role of a primary care pediatrician to provide genetic testing and evaluation (73%). Sixty-five percent also agree that genetic testing in children can help prevent certain adult-onset conditions.

- While 81% of pediatricians are confident in their ability to interpret newborn screening results, far fewer are confident in their ability to interpret genetic test results (31%) or explain genetic test results to parents (34%).

- About one-third each of pediatricians are moderately (31%) or very (34%) interested in Continuing Medical Education (CME) on genetics in primary care.