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Taking Family Histories with Genetics in Mind
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Unless your patient has a traumatic injury or an infection, the chances are better than even that his or her complaint has some genetic component. Every day researchers implicate genetic inheritance in another physical or mental disorder—from allergy to Zellweger syndrome, heart disease to osteoporosis, cancer to learning disorders. Most disorders (or predispositions to them) that "run in the family" are not determined by single genes, and it will be a long time before disease-related genes, their degrees of penetrance, and their complex interactions with other genes and the environment are completely understood. Nevertheless, knowing what runs in the family and observing its pattern of expression can provide both patient and physician with information that helps them plan the patient's health-related behaviors, including an illness-prevention life style, frequent screening and diagnostic testing, and early management if disease is detected.

In her comprehensive Practical Guide to the Genetic Family History, genetic counselor Robin Bennett strongly urges capturing patients' genetic histories by drawing their family "pedigrees" (from the French pie de grue, or crane's foot), a term that, she explains, refers to the curved claw-like diagrams that preceded today's more rectilinear family trees. Bennett calls the family tree a shorthand version of the family's genetic history, and certainly its diagram-and-symbol method of documenting relationships is speedier than writing out the likes of "maternal first cousin, once removed," then noting sex, age, and nature of the condition the individual displayed. Not only does drawing the graphic pedigree save time in the long term, it is probably more accurate. Asking how many aunts and uncles a patient has on both sides of the family and how many sons and daughters each one had calls these relatives into memory more clearly than broad questions such as, "Has anyone in the family had glaucoma?"

Taking the complete family history early in the patient-physician relationship avoids alarming a patient, perhaps unnecessarily, with inquiries about family incidence of heart disease, for example, or cancer, or Alzheimer's disease in response to a symptom complaint. Another advantage of the graphic is that it facilitates separating clinical patient records from genetic information, should a physician decide to protect patients from discrimination in insurance or employment by this means. While it seems nearly impossible to disentangle narrative family history, which may be offered piecemeal over many visits, from a
The symbols and nomenclature generally used for family genetic history taking were established by the National Society of Genetic Counselors Pedigree Standardization Task Force in 1995. They include specific representations for indicating sex, legal and biological connection, affected and carrier status, pregnancy, spontaneous abortion, termination of pregnancy, stillbirth and infertility, as well as noting connections resulting from adoption and the permutations of "blended" families. Software programs that generate pedigree charts are available as are online sites from which pedigree programs and explanations of the symbols they use can be downloaded.

**Taking the Family Genetic History**
Bennett offers some practical guidelines for the history-taking interview, reminding reader-history takers about the intimate nature of the information they are asking for and the psychological impact that such terms as "defect" or "bad gene" can have. She also warns against framing questions in ways that might direct the patient's answers, e.g., "Your brothers are both healthy?" With these provisos in place, the interviewer should ask about family health "from head to toe." Bennett asks about the (1) head, face, and neck (covering general appearance, problems with vision, speech and hearing, cleft palate, balding); (2) skeletal system (including bone formation, height, back curvature, multiple fractures); (3) skin; (4) the respiratory system; (5) cardiac system (blood pressure, heart murmurs, surgery); (6) gastrointestinal system; (7) renal system; (8) hematologic system (anemia, clotting problems, need for transfusions); (9) endocrine system (diabetes, thyroid conditions); (10) immune system; (11) reproduction (infertility, miscarriages, stillbirths); (12) neurological/neuromuscular problems (seizures and strokes, uncontrolled movements, slurred speech), and (13) mental functioning. Bennett asks about these systems at a general level of detail, progressing only to more directed questioning if there is a positive finding at the general level. At the completion of the system-by-system, head-to-toe survey, she asks separately about incidence of cancer, ethnicity, and drug and alcohol abuse.

**Using the Family Medical History**
The "therapeutic gap" that existed between diagnostic ability and effective treatment in the 19th and early 20th centuries describes the early 21st-century schism between recognizing genetic contribution to disease and effective gene therapy to correct the problem. The best "therapy" for diseases and disorders with genetic components is prevention and early diagnosis. Prevention can be most thoroughgoing in reproductive medicine through preconception decision making, prenatal testing, embryo selection, and use of alternatives to reproduction by the rearing parents, such as adoption, gamete donation, or donation and surrogate gestation. In disorders that are influenced by both genetic and environmental factors, attention to nutrition, exercise, and other life-style behaviors, such as abstention from tobacco use and moderation in alcohol consumption, can play
preventive or mitigating roles. In mutations that confer high probability of serious late-onset illness, early diagnosis and, therefore, early management can significantly ameliorate and prolong early stages of the disease. Despite the absence of effective gene therapy at present, continuing medical education emphasizes that practitioners understand genetic contribution to illness as a way to get out in front of morbidity, diagnosing early, and helping patients plan for illness management.

The AMA has developed a series of tools for family history taking, including a Prenatal Genetic Screening Questionnaire, a Pediatric Clinical Genetics Questionnaire, and an Adult Family History Form. Getting a thorough family history does take time, and, while genetic counselors are skilled in the procedure, most patients do not see genetic counselors as part of the routine intake visit, even in primary care fields. But, given the pedigree software and online tools, medical students, physician assistants, and other office and clinic professionals who currently take patient medical histories can be enrolled to take the family genetic history.

References
4. Bennett, 56.

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