

# THE GENE GENIE

Genetic counseling is expected to play an important role in the effective delivery of personalized medicine. Tara Funari helps families understand their genetic conditions and guides them through often difficult decisions.

By Robin Heffler

## Q. What is the purpose of genetic counseling, and why is it important in personalized medicine?

**Tara Funari:**

Genetic counselors help people understand the diagnosis of a genetic condition and the results of genetic testing, so that with the guidance of their physicians, they can make informed decisions about their health-care. We interpret family and medical histories, we assess the chance of a disease occurring or reoccurring in a family, and we educate families about inheritance patterns, genetic testing options, medical manage-

ment of conditions, and prevention. We also refer them to supportive resources—such as local organizations that deal with particular diseases or conditions—and we discuss options for being involved in research studies.

Personalized medicine involves using genetic information to identify an individual's disease risk and to determine a course of treatment or prevention that is specific to that individual's genetic makeup (i.e., based on a person's genetic profile, which drug would be the best choice for treatment?). Genetics will inevitably

play a much larger role in mainstream medical care. Genetic counselors will be instrumental in assisting physicians in interpreting and communicating results to patients.

## Q. For what kinds of medical conditions does Cedars-Sinai most often provide genetic counseling?

**A.** In addition to skeletal dysplasias, the Medical Center sees patients for everything ranging from prenatal genetic services, to pediatric genetics and metabolic disorders, to adult-onset conditions such as cardiovascular disease, neurological conditions, and cancer.

## Q. What should someone expect in a counseling session?

**A.** Typically, we begin by drawing a "pedigree," or family history tree, and assessing the risks of certain diseases. When a patient is the first one in a family to be diagnosed with a disease, we will discuss how the disease usually unfolds and the typical symptoms. Then we talk about the genetics. We explain what a gene is and its role in the body, how it changes to cause the disease, how the particular condition is

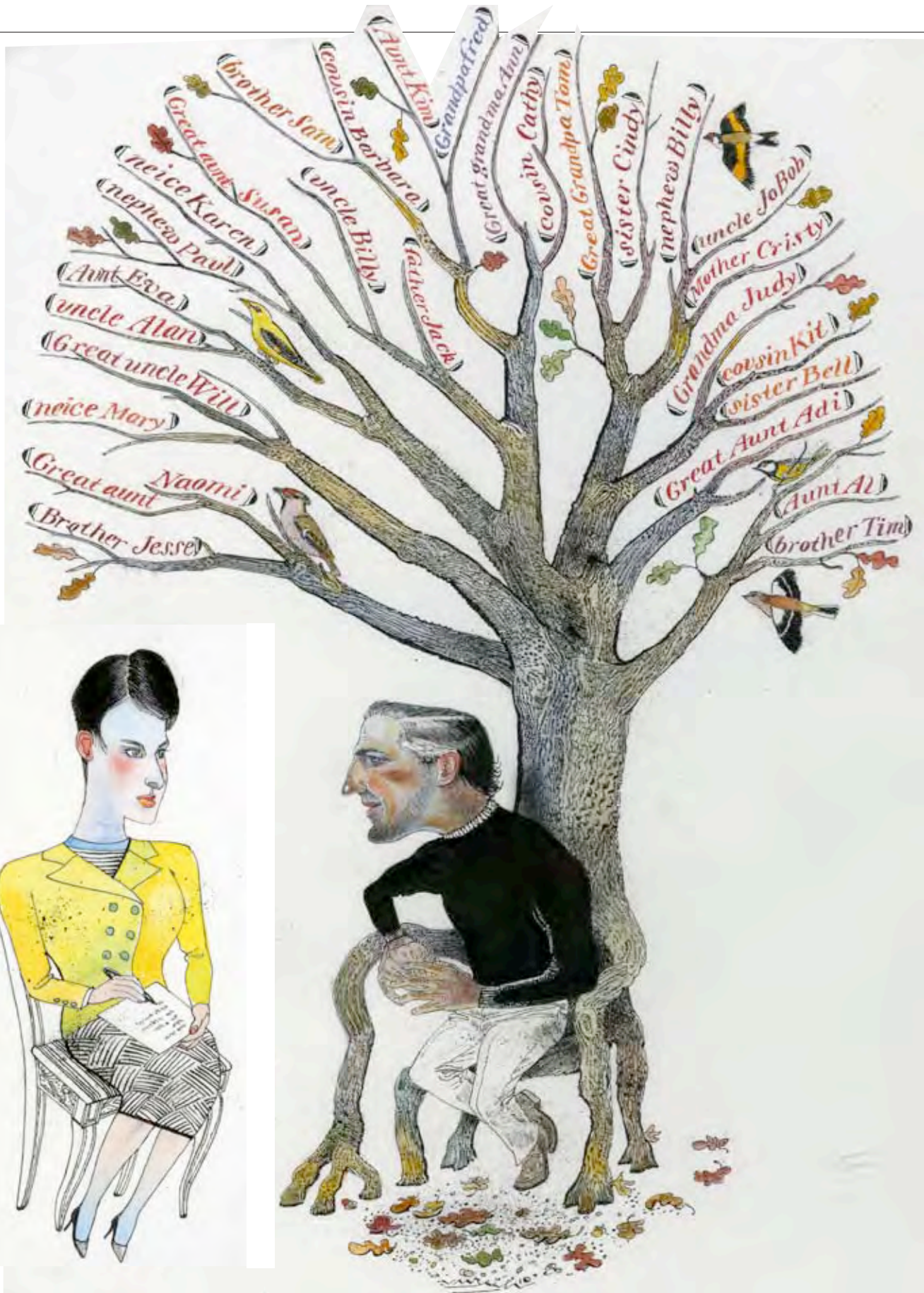
### WHAT TO ASK WHEN CONSIDERING

#### Genetic Testing

- What is the test looking for?
- How much of the risk is it going to identify?
- What are the limitations?
- Is it possible to get an ambiguous result or a non-interpretive result?
- How will the results translate into a difference in care?

#### Targeted Therapies

- Before you give me this drug, is there a genetic test that might suggest whether or not I would be a good responder?
- What does the data say about effectiveness in general?
- Are other drugs likely to interfere with results?



## GINA—Protection for Your Precious Genetic Material

**T**he Genetic Information Nondiscrimination Act (GINA) was signed in May 2008 but took over a decade to get through Congress. Until recently, people turned down lifesaving genetic testing for fear that genetic information would be used against them. The law, which went into full effect November 21, 2009, helps to alleviate that fear and sets clear restrictions on the use of genetic information.

### GINA

- Prohibits employers from requesting, requiring or purchasing genetic information about an employee or an employee's family member
- Prohibits health insurers from requesting or requiring a person to take a genetic test
- Forbids the use of a person's genetic information by health insurance companies for determining eligibility or insurance premiums
- Does not interfere with a health care workers' ability to request that a person or family member take a genetic test, or to provide patients with information on genetic tests that are available

Source: The Genetics and Public Policy Center at Johns Hopkins University

inherited, and what the chances are of a reoccurrence.

We also talk about the benefits and limitations of genetic testing, which sometimes can give ambiguous or uncertain results. We explain detection rates, the chances of false positives and negatives, and implications of the results for future medical management. We also counsel the patient about psychological implications of genetic testing for the individual and the family. After genetic testing is completed, patients and their families will usually return for a second counseling session to discuss results.

### Q. What fears do people commonly bring into a counseling session, and how do counselors provide emotional support?

**A.** People often think that the risk of having an abnormality is much greater than it actually is. Many people are afraid to learn about their genetic makeup, sometimes out of fear that a child has inherited a disease. Our goal is to help them understand the true risk and put their minds at ease, if possible. When we have to give bad news, we try to empower patients by giving them as much information as we can about the disease, because the scariest part is the unknown.

After giving test results, we typically sit down and help patients work through their feelings and design a game plan. In the process, we assess the person's coping mechanism, and occasionally we will call in a social worker or make a referral to a therapist.

### Q. Can you give an example of a situation in which you provided counseling and how it helped the patient?

**A.** One case involved parents who did not believe the pediatrician's diagnosis of dwarfism in their two-month-old baby. When they came to see us, we told them that tests revealed it was the correct diagnosis, and explained that sometimes it happens to average-size parents. They were well-educated people, but were in denial and wanted no part of the supportive resources we offered them. We had some follow-up phone calls, and at the next appointment there was a complete turnaround. They accepted the baby's diagnosis, took the information we gave to them, and became very involved in the national support organization Little People of America.

The opportunity to interact with patients and help people like this is one of the reasons I enjoy my work. It is very rewarding when you talk to patients and see their cases come full circle, helping them through difficult times. ☺

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Since 2006, Tara Funari has been a certified genetic counselor in the Cedars-Sinai Skeletal Dysplasia Clinic,



which sees patients with abnormally developed bones and connective tissues. She also serves as research coordinator for the International Skeletal Dysplasia Registry, and trains graduate students in genetic counseling.