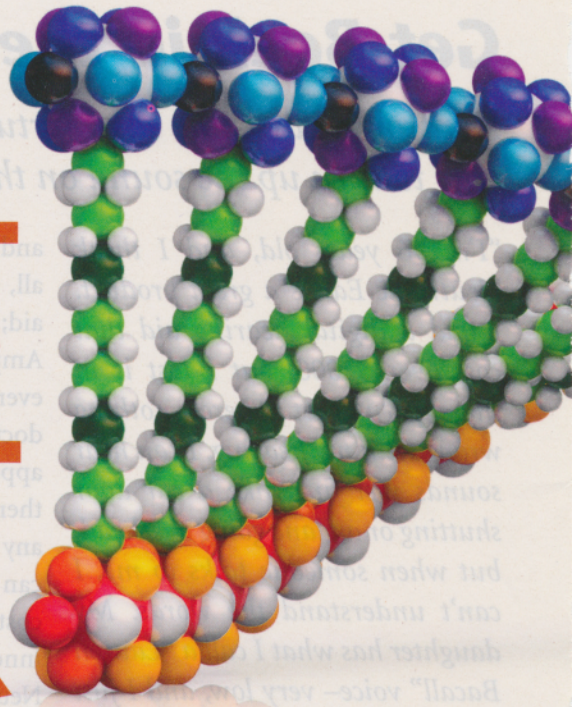


MORE POWER TO YOU

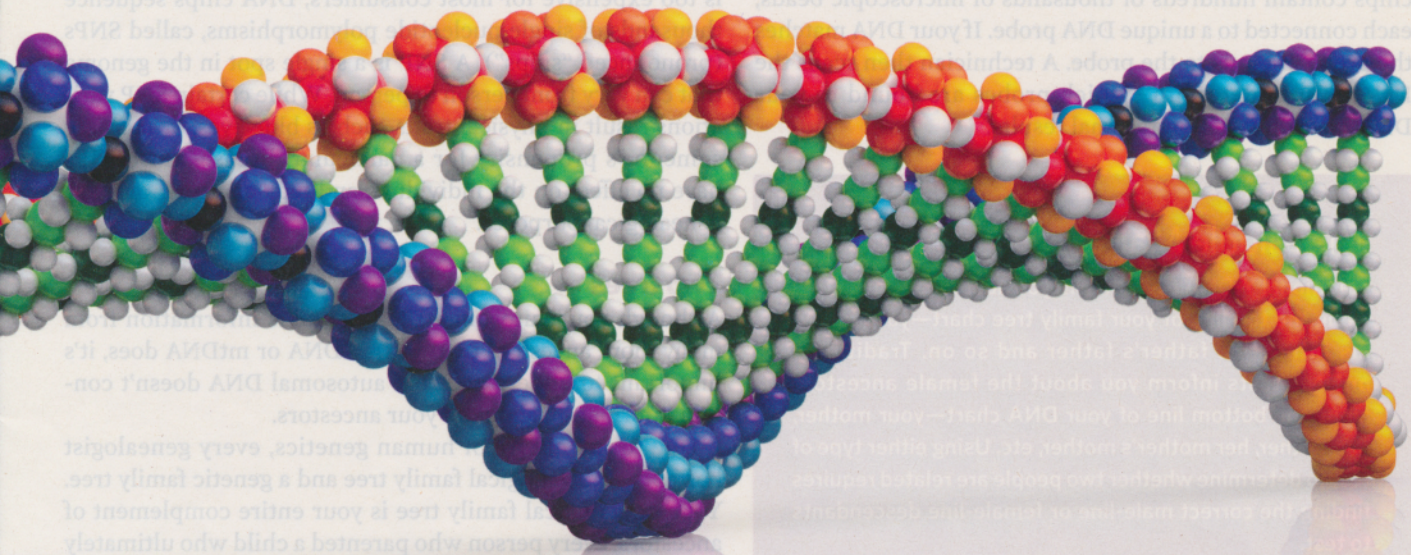
BY BLAINE BETTINGER

Traditional genetic genealogy tests limit you to male-only or female-only family lines—but autosomal DNA tests offer the potential to research the rest of your tree.

WHEN SHE DIED at age 93, Marley Snell had no knowledge of her biological parents. Born Helen Marley Johnson in the spring of 1889 in rural Upstate New York, Marley was adopted by a family that often took in orphans. Following her adoptive parents' deaths a few years later, Marley was passed along to at least two other families in different counties. Although my great-grandmother later married and went on to live a long, full life, her search for her biological parents never ended. Assisted by technology Marley couldn't have dreamed of, I'm hoping to finish her search.

Just a few years ago, my quest would've been limited to paper records and family lore—both of which are susceptible to human error and deception. DNA wouldn't have helped, because Marley didn't have Y-DNA, and mitochondrial DNA (mtDNA) from one of her descendants failed to provide any useful clues. But now, a new type of DNA testing—using autosomal chromosomes—will let me use biology to explore her past. With hard work and a bit of luck, I can identify pieces of Marley's DNA in her living descendants and discover genetic cousins who'll eventually lead me to her biological parents.

Want to take a similar route around brick walls in your research? Here's the 411 on autosomal DNA and how you can use it to explore your ancestry.



Chromosomal normalities

What makes autosomal DNA so special? A quick high school biology review is in order. Each of us has 23 pairs of chromosomes—long chains of DNA—in our cells. These 46 chromosomes make up your genome. One pair of these chromosomes are the sex chromosomes. A male has a Y-chromosome from his father and an X-chromosome from his mother; a female has two X-chromosomes, one from each parent. The remaining 22 pairs of chromosomes are your autosomal DNA.

One parent provides each member of a pair of autosomal chromosomes. In other words, your father furnished an entire complement of 22 autosomal chromosomes and your mother gave you a corresponding set of 22 autosomal chromosomes. As a result, your autosomal chromosomes represent DNA from both the maternal and paternal sides of your ancestry.

This pattern of inheritance sets up one of the major differences between autosomal DNA testing and other types of DNA testing. At 10 generations in the past, a Y-DNA test (which examines only male-line ancestors) or an mtDNA test (which examines only female line ancestors) will tell you about only one individual out of roughly 1,024 ancestors. Autosomal DNA, however, contains information from as many as 100 or more of those 1,024 ancestors.

A SNP here, a SNP there

Commercial autosomal DNA testing began around 2002 with the launch of DNAPrint Genomics' AncestryByDNA test, which estimated percentages of DNA inherited from various world regions. It initially examined approximately 71 chromosomal locations throughout the genome. Today it's offered by DNA Diagnostics Center **<www.ancestrybydna.com>** and gathers ancestral information from many more genome locations. Additional companies have since introduced higher-resolution autosomal tests that examine many more markers.

The majority of autosomal DNA ancestry tests are purchased through 23andMe **<23andme.com>** and Family Tree DNA **<www.familytreedna.com>**, which typically test hundreds of thousands to a million locations in the genome. Each compares test-takers' DNA to a proprietary database to identify genetic cousins, and offers other services and tools for genealogy research. (DeCODE Genetics **<decodme.com>** also provides an autosomal test, though it focuses on genetic risks for medical conditions.)

As with most DNA tests, taking an autosomal test is quick and painless. After receiving a kit in the mail from your chosen testing company, you take a DNA sample by swabbing your cheek or spitting into a cup. Back at the laboratory, technicians extract DNA from the sample and chop it up into small pieces. They wash the chopped-up DNA over a DNA microarray, a small testing device also known as a gene chip or a DNA chip (23andMe and Family Tree DNA use microarray technology

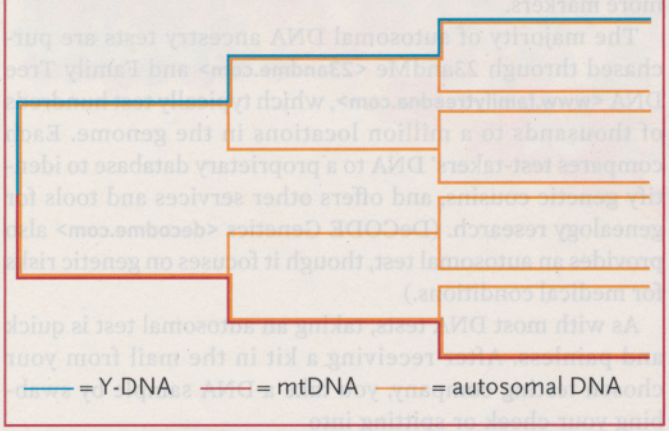
TIP: To identify which ancestors likely contributed to your autosomal DNA, find a genetic cousin and compare your family tree chart to his. Ancestors who are named on both charts likely provided the DNA sequences you share with your cousin.

from a company called Illumina <www.illumina.com>). DNA chips contain hundreds of thousands of microscopic beads, each connected to a unique DNA probe. If your DNA matches the probe, it binds to the probe. A technician then scans the DNA chip to determine which probes are bound to your DNA, revealing your autosomal results.

The Middle Matters

Traditional Y-DNA tests inform you about male ancestors along the top line of your family tree chart—your father, his father, his father's father and so on. Traditional mtDNA tests inform you about the female ancestors along the bottom line of your DNA chart—your mother, her mother, her mother's mother, etc. Using either type of test to determine whether two people are related requires finding the correct male-line or female-line descendants to test.

Autosomal DNA, however, contains genetic material from people in the top, bottom *and* middle branches of your family tree. But there's a catch: Beyond your great-great-grandparents, not all your ancestors are reflected in your autosomal DNA. And we don't have a test—yet—that tells you which part of your autosomal DNA came from which ancestor.



Rather than sequence a test-taker's entire genome, which is too expensive for most consumers, DNA chips sequence thousands of single nucleotide polymorphisms, called SNPs (pronounced "snips"). A SNP is a single spot in the genome that can vary from person to person. While certain SNP variations result in physical traits such as blue eyes, or increase someone's propensity for a particular disease, most SNPs have no effect on the individual, and may be useful only for genealogical purposes.

Two trees

Although your autosomal DNA contains information from many more ancestors than your Y-DNA or mtDNA does, it's important to understand that autosomal DNA doesn't contain information about *all* your ancestors.

Due to the nature of human genetics, every genealogist has both a genealogical family tree and a genetic family tree. Your genealogical family tree is your entire complement of ancestors, every person who parented a child who ultimately led to you. But because parents pass along only pieces of their genomes to their children, not every person in your genealogical tree contributed to your autosomal DNA. With every new person, pieces of DNA are lost and gained, resulting in a brand-new genome that holds DNA from a random assortment of ancestors.

Accordingly, everyone has a genetic family tree, which is a small subset of the genealogical family tree. Your genetic tree contains only those people who contributed a portion of your autosomal DNA. While you can be sure that your parents, grandparents and great-grandparents are members of both your genealogical and genetic trees, at about the third-great-grandparent generation, ancestors quickly begin to fall off your genetic family tree.

One challenge of autosomal DNA testing is to identify the ancestors who likely contributed to your DNA (those on your genetic tree). This is typically done by comparing your genealogical tree to the genealogical tree of a genetic cousin in order to identify overlap. Genetic cousins can successfully identify their shared ancestors by using autosomal DNA testing to trace individual segments of their DNA across time and space.

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Oh, the possibilities

Mountain View, Calif.-based 23andMe, launched in 2007, was one of the first companies to offer large-scale autosomal DNA testing. The price (at press time) is $99 plus $9 a month with a 12-month commitment to the Personal Genome Service, or $399 without the service. Keep your eyes peeled for specials around the holidays and DNA day (April 25).

In addition to information about traits from eye color to bitter taste perception, risk factors for almost 100 different diseases, and predicted sensitivities to a variety of common pharmaceuticals, the 23andMe test offers a wealth of genealogical information.

Family Tree DNA has been offering DNA testing to genealogists for more than a decade. In 2010, the company entered the world of large-scale autosomal DNA testing with the Family Finder test. It offers many features similar to those of 23andMe, except that it doesn't provide information about physical traits, medical propensities or predicted drug reactions—a factor that appeals to genealogists looking for only ancestry-related results.

Besides basic SNP tests, both companies have tests that estimate your ethnic ancestry, and comparisons that search for genetic matches in their proprietary databases. Shared segments of DNA identify two people as genetic cousins. More and longer shared segments mean a closer relationship. The companies also offer an array of tools (detailed below) that help you use your test results in your genealogy research and the option to download the results of a SNP test as a zipped file. Numerous third-party applications use these results, although you should be aware of privacy issues involved with any sharing of autosomal DNA test results (see the box at right for more on these).

23ANDME OFFERS THESE AUTOSOMAL SERVICES:

- **PATERNAL LINE:** This test examines hundreds of SNPs on a man's Y chromosome and determines his Y-DNA haplogroup. Although all Y chromosomes for men alive today come from a common ancestor approximately 100,000 to 50,000 years ago, over time the descendants have gained unique SNP variations—also called mutations—that created branches of the Y-DNA haplogroup tree. Males who share a Y-DNA haplogroup are descended from a common male ancestor within the distant past.
- **MATERNAL LINE:** Similar to the paternal line test, this test examines SNPs on mtDNA to determine an mtDNA haplogroup. All people have mtDNA, although only mothers pass mtDNA to their children. This mtDNA comes from a common maternal ancestor who lived approximately 200,000 years ago (estimates vary). As with Y-DNA, we've all gained unique SNP variations that created new branches of the mtDNA haplogroup tree. People who share an mtDNA haplogroup are descended from a common female ancestor within the distant past.
- **ANCESTRY PAINTING:** This test examines autosomal chromosome segments to determine whether they're likely inherited from ancestors in Africa, Asia or Europe. Results show the segments colored by geographical region on a graph of your chromosomes. They represent geographic regions prior to the extensive movement of people around the globe in the last few hundred years. That means the ancestor from whom you inherited a segment of European DNA was likely in Europe 500 years ago.
- **RELATIVE FINDER:** Use this tool to look for relatives by comparing your DNA to the DNA of everyone in the 23andMe database (currently 100,000-plus people). If it finds a match, the tool predicts the relationship ranging from parent-child to "distant cousin." Names of matches are kept anonymous; each person can decide whether to contact the predicted relative. One drawback is that users must opt in to Relative Finder, and many aren't interested. Some test-takers have complained about finding genetic matches in the 23andMe database who have no interest in connecting with others.
- **GLOBAL SIMILARITY:** Genetically, all humans are more than 99 percent similar. This tool examines the remaining less-than-one-percent to determine how similar you are to several reference populations around the world, estimating your ethnic heritage. Results are on a bar chart and a world map highlighting the populations you most closely match.

FAMILY TREE DNA'S AUTOSOMAL TESTS AND TOOLS INCLUDE:

- **POPULATION FINDER:** This tool separates segments of your DNA into biogeographical regions by comparing each segment to numerous global populations organized into continental groups (Africa, the Americas, East Asia, Europe, Middle East, Oceania and South Asia). The results are

Autosomal Apps

Once you have a zipped file with your autosomal DNA results, you can use a multitude of third-party applications to run useful and interesting analyses. Note that some apps can be a bit challenging for those without a science background. Here are three of my favorites (all are free):

- **SNPEDIA <snpedia.com>** compares your autosomal DNA results to the SNPedia database, which contains information about thousands of SNPs. You get back information about your physical traits, conditions and propensity for certain genetic diseases.
- **DAVID PIKE'S UTILITIES <www.math.mun.ca/~dapike/FF23utils>** include one that can "phase" your autosomal DNA results (order SNPs into the chromosomes inherited from each parent) if your parents have also been tested. Normally, autosomal tests using SNP chips can't tell which of a pair of chromosomes was the source for a particular SNP result.
- **INTERPRETOME <esquilax.stanford.edu>** has several apps. One determines how many Neanderthal SNPs you might have (although this research is still early).

reported as percentages of ancestries estimated to have contributed to your DNA. As with many autosomal DNA tools, the Population Finder is in beta testing. Interpretations may change as the algorithm is refined and new comparison populations are used.

■ **MATCHES:** This tool searches for relatives by comparing your autosomal DNA to Family Tree DNA's results database. The degree of matching between two individuals determines their predicted relationship. But unlike Relative Finder, Family Tree DNA gives you each match's name and email address, instantly enabling communication. You can assign a relationship to the genetic cousin, if you know it. You'll see a list of surnames genetic cousins have entered into their profiles, too; names that also appear in your profile are bolded.

■ **CHROMOSOME BROWSER:** Use this tool to compare your DNA to up to five matches to determine exactly which segments of DNA you share. This is a great visual tool, displaying results on a graph of 22 chromosomes. (You can opt to see the results in a table and download them in a spreadsheet.) If DNA from two of your matches overlaps, there's a chance they're also related to each other. That could help you identify a common ancestor by "triangulating" relationships. In the event all three of you are related through the same piece of DNA, the common ancestor will be someone you all share. If such an overlap occurs on your chromosomes, ask one of your matches if the third person is also in his or her results.

Another key point: Although DNA testing companies constantly refine the algorithms used to analyze results and compare DNA, genetic matches are based on probabilities, not certainties. Matches are predicted using a predetermined threshold. Two unrelated people could randomly share a DNA sequence and satisfy that threshold even if they don't share recent ancestry. Keep this in mind, especially when working with distant cousins (beyond about the fifth cousin range).

TIP: Need help using your DNA test results in your genealogy research? Download Bettinger's free e-book *I Have The Results of My Genetic Genealogy Test, Now What?* from **<www.thegeneticgenealogist.com>**.

At 10 generations, a Y-DNA test or mtDNA test tells you about one of roughly 1,024 ancestors. But autosomal DNA has information from up to 100 or more of those ancestors.

Marley and me

Let's get back to the search for Marley's parents. To obtain the most potential information about Marley and her parents, it would be ideal to send away her DNA for testing. But Marley and her two children have been dead for almost 30 years. She does have living grandchildren from each of her children, including my father, Fred, and his first cousin Simon (I've changed both names to protect their privacy). Fred and Simon have identical pieces of DNA.

As long as the two men aren't recently related in another way, their shared DNA is inherited from only their most recent common ancestors—Marley and her husband. Fred inherited the shared DNA from his father; Simon, from his mother. For finding Marley's parents, Y-DNA testing won't work because Marley didn't have Y-DNA to pass down to Fred and Simon. MtDNA comparisons aren't useful for recent ancestry, because mtDNA mutates infrequently.

Once I've identified that shared DNA with a test, I could use the Global Similarity tool (23andMe) or Population Finder (Family Tree DNA) to learn about the predicted ethnicity for those pieces of DNA and get clues to Marley's ancestry. I can also look for genetic cousins in the testing company's database, especially any cousins that match both Fred and Simon—they're also likely related to Marley or her husband. Then I can use my knowledge of Marley's husband's genealogy to narrow those genetic matches to only Marley's relatives. Together, Marley's relatives and I may be able to find Marley's parents.

Although it's a multistep process with no guarantee of success, autosomal DNA testing and analysis tools—combined with traditional genealogical research—present the most promising new avenue of research into brick walls such as Marley's unknown ancestry. ■

BLAINE BETTINGER is a lawyer and biochemist who blogs and offers a free e-book on using your DNA test results at the Genetic Genealogist **<www.thegeneticgenealogist.com>**. He's crowdsourcing the search for Marley's parents—to participate, visit **<goo.gl/WrFZ9>**.