Ancestral Regions 2025 White Paper

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1. Summary

Reporting ancestral regions (“regions”) is one of several tools that AncestryDNA offers customers on their journey to discover their heritage, ancestors, and family history. Ancestral regions is a feature that connects users directly to the populations from which their ancestors likely came. This information can be used in conjunction with ancestral journeys discovered through Genetic Communities, and with relationships discovered through DNA Matching to better understand one’s more recent and distant past.

AncestryDNA employs a team of highly trained scientists with backgrounds in population genetics, statistics, machine learning, and computational biology to develop a fast, sophisticated, and accurate method for estimating genetic ancestral regions. The AncestryDNA science team has advanced the science and technology behind the region results this year, producing an increase in both the overall accuracy of the results, as well as the number of regions available for assignment (from 107 regions to 146). By adding these new regions, we provide even greater granularity to our members.

This white paper will delve into the science behind:

1. How our reference panel samples are chosen and the makeup of our 146 reference panels for 2025
2. How our algorithm works to estimate a customer’s genetic ancestral origins
3. Some of the results from our most recent advances for inferring ancestral origins from DNA

Glossary

Admixed — Describing an individual or population that has origins from multiple populations.
Allele — A variant in the DNA sequence. For example, a SNP (defined below) could have two alleles: A or C.
Centimorgan (cM) — A unit of genetic length in the genome. Two genomic positions that are a centimorgan apart have a 1% chance during each meiosis (the cell division that creates egg cells or sperm) of experiencing a recombination event between them.
Chromosome — A large, inherited piece of DNA. Humans typically have 23 pairs of chromosomes with one copy of each pair inherited from each parent.

IBD — A term abbreviated from “Identity-by-descent.” When two individuals share DNA, we can say they have DNA that is IBD, if there is evidence that they share that DNA because they inherited it from a recent ancestor.
Genome — All of someone’s genetic information; the DNA on all chromosomes.
Genotype — A general term for observed genetic variation either for a single site or the whole genome. For example, we can refer to the results for a customer from our microarray as a “customer’s genotype.”
Haplotype — A stretch of DNA along a chromosome containing a group of nucleotide polymorphisms.
Hidden Markov model (HMM) — A statistical model for determining a series of hidden states based on a set of observations.
Locus/Loci — A location or locations in the genome. It could be a single site or a larger stretch of DNA.
Microarray — A DNA microarray is a way to analyze hundreds of thousands of DNA markers all at once.
Nucleotide — DNA is composed of strings of molecules called nucleotides (also called bases). There are four different types, and they are usually represented by their initials: A, C, G, T.
Population — A group of people.
Phasing — The assignment of DNA to contiguous segments corresponding to the DNA inherited from Mom or Dad. This is done with an algorithm.
Recombination — Before chromosomes are passed down from parent to child, each pair of chromosomes usually exchange long segments between one another and then are reattached in a process called recombination.
Reference Panel — A set of people whose DNA is typical of DNA from a certain place—people native to a place or group. The DNA of these people is used as a representation of the typical DNA from this place for the purposes of studying population genetics and history.
Single nucleotide polymorphism (SNP) — A single position (nucleotide) in the genome where different variants (alleles) are seen in different people.

2. Constructing Population Reference Panels

2.1 Reference Panels are Critical to Calculate Ancestral Regions

The basic premise behind ancestral regions inference can be summarized as follows. Two **haplotypes** from the same geographic region or the same population will share more DNA with one another than will two haplotypes from different regions or groups. So two people with a historical connection to Portugal will have more DNA in common than a person from Korea will have with a person from Portugal.

In practice, region inference involves comparing a person's DNA to the DNA of multiple **reference panels**. A reference panel is composed of individuals whose DNA is representative of a population. Our algorithm compares a person's DNA segments to these reference panels to determine the best-matching populations. If, for example, a section of a person's DNA looks most similar to DNA of people in our Norway reference panel, that section is said to be from Norway, and so on. The end result is a genome-wide report where individual sections of DNA are associated with one of the 146 regions in our reference panel. The similarity breakdown is provided as a total percentage breakdown, and also as a per-parent and per-chromosome report.

The accuracy of our results depends on the quality of our reference panel. Because of this, AncestryDNA has invested a significant amount of effort in collecting DNA data from populations across the globe and developing the best possible set of reference samples.

The rest of Section 2 describes the steps taken to develop our current reference panel.

2.2 Developing Reference Panels in Regions of the World with Significant Amounts of Data

AncestryDNA has genome-wide genotype data for over 25 million customers from around the world. Additionally, many people in our database have connected family trees to their DNA results, providing invaluable contextual information about the origins of these individuals. The wealth of DNA and genealogical information allows us to create robust reference panels for many global populations, especially in regions of the world where our customers' origins are concentrated.

However, it is problematic to rely solely on self-reported genealogical information from customer trees when deciding what individuals to include in the reference panel and which populations they should represent. Relying just on samples that have connected family trees would limit our number of reference panel candidates too dramatically. As well, family trees can be difficult to verify and can carry errors.

Therefore, we've adopted a strategy that is primarily driven by genetic relatedness (**IBD**) among people and populations, while still incorporating information in the aggregate from ancestral region results and family trees. Specifically, we leverage our Genetic Communities technology and the 3,500+ DNA-based communities we have discovered as the bases for reference panels.

Our Genetic Communities technology identifies networks of Ancestry members that are highly interconnected due to sharing DNA from a common set of ancestors. We are also able to look at family

tree information in aggregate for these networks to identify shared places of origin, patterns of movement, statistically enriched surnames, and genetic ancestral region results. These data provide insight into the specific identity and story of these genetically related groups.

Using the networks we discover as a basis for ancestral region reference panels has several benefits. First, the networks are entirely driven by genetics and self-organized population structure. Identifying individuals to represent a population based on patterns of genetic similarity between these individuals is preferable to hand-curating a set of individuals based on self-reported data such as origins, language, or ethnicity. Second, when we do need to rely on information about origins, language, and ethnicity for identifying and annotating the reference populations, we are using information that has been averaged from hundreds or thousands of network members. By averaging the data, we remove the disproportionate effects of outliers or the need to extensively verify several hundred individual records.

A complicating factor is that individuals' membership in these networks are non-exclusive. Specifically, an individual may belong to multiple different networks, representing various branches of their family tree. For example, if a person has one parent of Irish descent and another parent of Italian descent, they will likely belong to both Irish and Italian networks. Using this individual in any Italian or Irish reference panel would adversely impact the performance of our analysis.

In order to optimize our selection of reference panel candidates, we therefore adopted several filtering approaches based exclusively on genetic data.

1. We mapped the networks to corresponding world regions, and regions with rich data were selected for this reference panel development approach.
2. We considered reference panel candidates for each region and selected samples that were more likely to descend from a single origin population, i.e., do not have recently admixed family origins. We did this by filtering out individuals who had a weak genetic connection to their assigned networks (based on their number of matches to other members of that network) and who were assigned to multiple networks from different populations (e.g., Irish and Italian).
3. We filtered individuals based on their current genetic results, removing individuals with exceptionally high levels of additional off-target regions. Specifically, we identified the most common regions shared among individuals of each network, and through an iterative analysis, determined specific regions and percentage thresholds to ensure robust reference panels. For example, consider that in England, most individuals with deep family roots to the south and east, and who could be high quality reference panel candidates, will likely carry between 3-5% Scandinavian DNA. This is a result of the historical invasions of England by Germanic and Viking

tribes 1500 years ago. Despite these individuals having a genetic connection outside of England, we would still want to consider them as reference panel candidates.

4. We filtered out individuals who had a low number of matches with others in the network, and who had a proportionally high number of matches to individuals in other networks. Specifically, for each sample we calculated the number of matches to others in the network and to other networks. We then examined the distribution of these metrics and identified a percentile cutoff below which all individuals were excluded. The effect was to remove individuals who showed a low level of genetic connectedness to the specific region and a high level of genetic connectedness to outside regions.
5. As a final check on the individuals selected for a region's reference panel, we aggregated the birth location data for these individuals and their ancestors and plotted the locations on a map. We found strong signals of enrichment for ancestor birth locations in the geographic regions of interest, and very little signal outside the region of interest.

Using the filtering approach based on IBD-sharing outlined above, we identify the individuals who are best suited to include in the reference panel. Given the large number of samples available at AncestryDNA, we subsample to a maximum of 5000 individuals per reference panel to train our model ("training set"), along with 500 testing samples to tune parameters ("testing set"), and 500 samples for final validation ("validation set").

2.3 Developing Reference Panels in regions with limited data

In some regions of the world, AncestryDNA has not yet acquired enough customer samples to rely on the process described above (Section 2.2). In these regions, AncestryDNA relies on a collection of labeled samples from worldwide populations to construct training, testing, and validation sets.

We build up this collection using several different data sources including:

- 1,000 samples from 49 worldwide populations from a public project called the Human Genome Diversity Project (HGDP) (Cann *et al*. 2002; Cavalli-Sforza 2005)
- 2,500 samples from 19 populations from the 1000 Genomes Project (McVean *et al.*, 2012)
- 900 samples from 84 populations from the Human Origins dataset (Lazaridis *et al.*, Nature 2014).
- Proprietary AncestryDNA reference collections
- AncestryDNA samples from customers who consented to participate in the research project

Once this collection has been compiled, we again use unsupervised techniques that leverage shared IBD to identify groups of individuals with shared origins. We then use aggregated meta-data (e.g., self-reported ethnicity, spoken language) to identify the groups. We sample a subset of the total individuals to include in our reference panel.

2.3 Developing Reference Panels in Regions with significant admixture

In some parts of the world, indigenous people carry DNA originating from more than one continent. For example, people of Amerindian descent in North and South America may also have some DNA from Europe and Africa. When creating reference panels for the Americas and Oceania, we use only the parts of the genome inherited from the indigenous populations. We do this by looking at our previous assignments to select only the segments of DNA where both chromosomes have an assignment to an indigenous population. So, whereas most of our regions use DNA from the entire genome of each reference panel candidate, when creating reference panels for populations in areas that are now admixed we only use a fraction of each person's genomes. The regions where we employ this approach are:

Bolivia & Peru
Colombia & Venezuela
Mexico
Canada & USA
Yucatan Peninsula
Central
Chile
Ecuador
Panama & Costa Rica
Eastern South America
Puerto Rico
New Zealand Māori
Aboriginal and/or Torres Strait Islander
Hawaii
Samoa
Tonga

For two other regions, Cuba and Haiti & Dominican Republic, we use windows where only one chromosome has assignment to the indigenous population. We then combine single chromosomes from two different people in the same window. This creates a window homozygous for the indigenous DNA.

2.4 Reference Panel Quality Control

For each sample, we analyze a set of approximately 300,000 SNPs that are shared between the Illumina OmniExpress platform and the Illumina HumanHap 650Y platform (which was used to genotype HGDP samples). Samples with large amounts of missing data are removed. We also remove samples which are likely to degrade the performance of the reference panel. Samples can be removed because 1) they are closely related to another reference sample, or 2) the underlying genetic information about a sample's origins disagrees with the sample labels, as determined through principal component analysis (PCA) (Jackson 2003, Patterson 2006) and our previous genetic analyses (Figure 2.1).

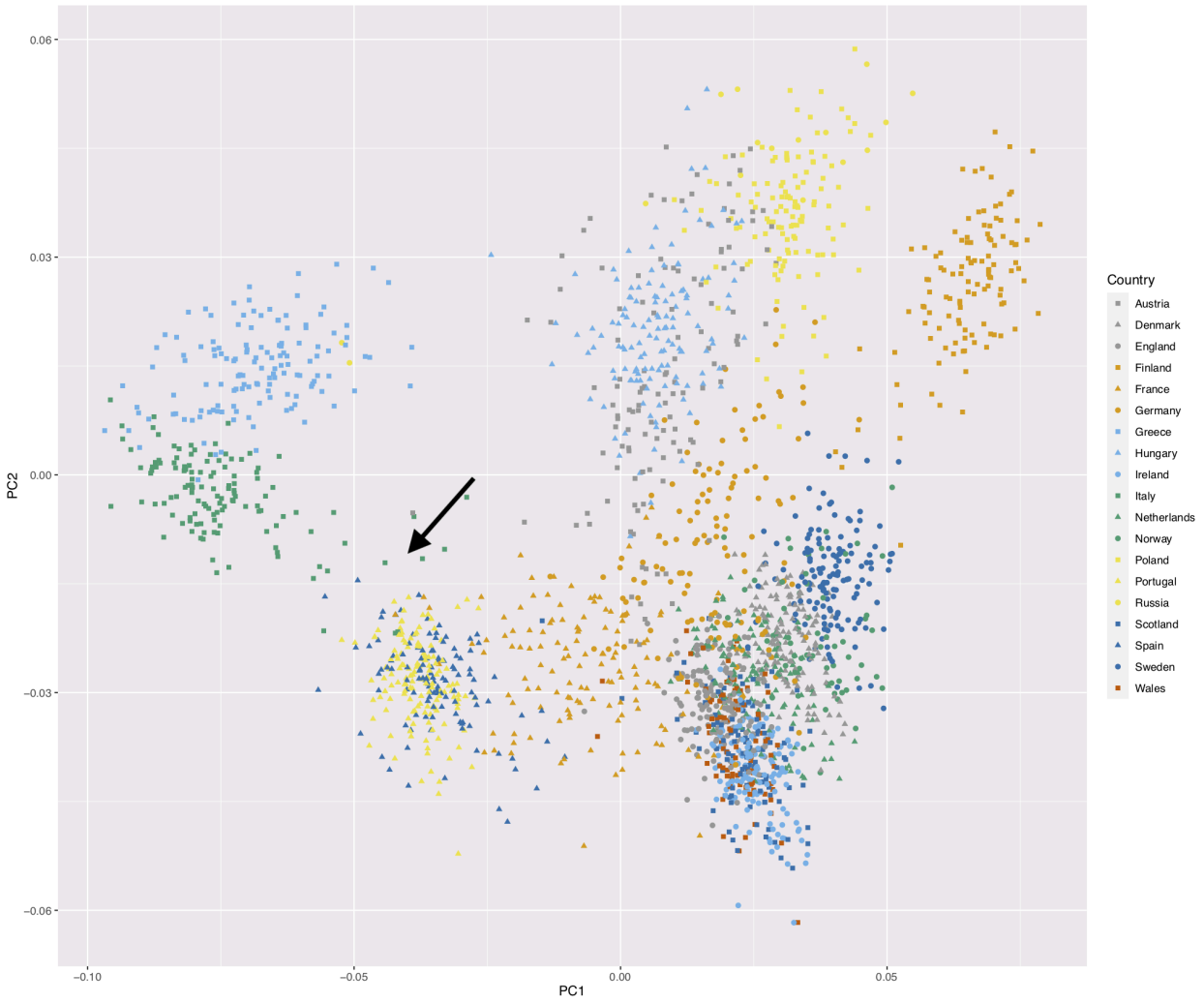


Figure 2.1: PCA Analysis on European Panel Candidates. *Scatter plot of the first two components from a principal component analysis (PCA) of candidate European samples for the AncestryDNA reference panel. Visual inspection of PCA is useful for numerous aspects of data QC. First, it can be used to identify individual outliers, such as the Italian samples (green squares) that appear near the Portugal and Spain (yellow and blue triangles, respectively) cluster. It can also be useful for identifying poor sample grouping. Finally, it can reveal regions where there is limited genetic separation and clusters overlap (e.g., England, Ireland, Wales, and Scotland clusters) and regions that can be further subdivided.*

2.5 Updated Reference Panel

The updated AncestryDNA ancestral regions reference panel contains 185,063 samples carefully selected as described above to represent 146 global regions (Table 2.1), each with a unique genetic profile. As a comparison, our previous panel of 116,830 samples represented 107 distinct global regions.

Table 2.1: The AncestryDNA Regions Reference Panel

Region
Senegal
Mali
Ivory Coast & Ghana
Benin & Togo
Yorubaland
Central West Africa
Central Nigeria
North-Central Nigeria
Nigeria
Nigerian Woodlands
Cameroon
Western Bantu Peoples
Twa
Southern Bantu Peoples
Eastern Bantu Peoples
Nilotic Peoples
Ethiopia & Eritrea
Somalia
Khoisan, Aka & Mbuti Peoples
North Africa
Egypt
Arabian Peninsula
Levant
Cyprus
Anatolia & the Caucasus
Iran/Persia
Lower Central Asia
Northern Iraq & Northern Iran
Burusho
Indo-Gangetic Plain
Western Himalayas & the Hindu Kush

Gujarat
Gulf of Khambhat
Southern India
Southwest India
The Deccan & the Gulf of Mannar
Bengal
Nepal & the Himalayan Foothills
Tibetan Peoples
Northern Asia
Mongolia & Upper Central Asia
Korea
Japan
Southern Japanese Islands
Northern China
Western China
Southwestern China
Central & Eastern China & Taiwan
Southern China
Dai
Mainland Southeast Asia
Maritime Southeast Asia
Vietnam
Northern & Central Philippines
Central & Southern Philippines
Luzon
Western Visayas
Guam
Melanesia
Aboriginal and/or Torres Strait Islander Peoples
Tonga
Samoa
Hawaii
New Zealand Māori
Arctic

Canada & United States
Mexico
Yucatan Peninsula
Central America
Panama & Costa Rica
Cuba
Haiti & Dominican Republic
Puerto Rico
Colombia & Venezuela
Ecuador
Bolivia & Peru
Chile
Eastern South America
Ashkenazi Jews in Eastern Europe & Russia
Ashkenazi Jews in Central & Southeastern Europe
Sephardic Jews in the Eastern Mediterranean
Sephardic Jews in Northern Africa
Finland
Sweden
Denmark
Norway
Iceland
Estonia & Latvia
Lithuania
Russia
Northeastern Poland
North Central Europe
Southern Poland
Eastern Czechia
Slovakia
Slovenia
Western Ukraine
Western Balkans
Northwestern Balkans

Romania
Southwestern Balkans
Eastern European Roma
Ionian Islands
Northern & Central Greece
Southern Greece
Albania
Aegean Islands
Crete
Malta
Sardinia
Southern Italy
Sicily
Northwestern Italy
Northeastern Italy
Central Italy
Acadia
France
Brittany, France
Quebec
The Netherlands
Northwestern Germany
Southern Germany
Russian Germans
Basque
Canary Islands
Northern Spain
Spain
Portugal
Azores
Madeira
Cornwall
West Midlands
Devon & Somerset

North East England
Southeastern England & Northwestern Europe
East Midlands
Southern Wales
Northern Wales & North West England
Isle of Man
Central Scotland & Northern Ireland
North East Scotland
Hebrides & Western Highlands, Scotland
Connacht, Ireland
Donegal, Ireland
Leinster, Ireland
Munster, Ireland
Total

3. AncestryDNA Ancestral Regions Algorithm

3.1 Algorithm Intuition and Assumptions

After establishing the reference panels, the next step is to train and tune the algorithm that infers a customer's ancestral regions by comparing nearly 300,000 selected single nucleotide polymorphisms (SNPs) from their DNA to those of the reference panel. In this comparison algorithm, we assume that an individual's DNA is a mixture of DNA from some combination of the 146 identified populations. To illustrate this concept, we show a cartoon example in Figure 3.1, where, because of recombination, a customer inherits stretches of DNA from her four grandparents who, in this example, each come from four "single source" reference populations.

Because DNA is passed down from one generation to the next in long segments, it is likely that the DNA at two nearby loci in the genome were inherited from the same person and therefore the same population (for more details on DNA inheritance see our matching white paper). This means we can get more

accurate results by looking at multiple nearby SNPs together as a haplotype, instead of looking at each SNP in isolation. Our algorithm takes advantage of this to greatly improve our estimates.

Our approach divides the customer's genome into 1,001 windows and assumes that the DNA inherited from each parent in each window comes from exactly one population (the windows are small enough that this will almost always be true). We compare the customer's DNA to the reference panels for each window, and combine information from all the windows to estimate what overall portion of the customer's genome came from each population using a hidden Markov model (HMM), described in Sections 3.3-3.5 below.

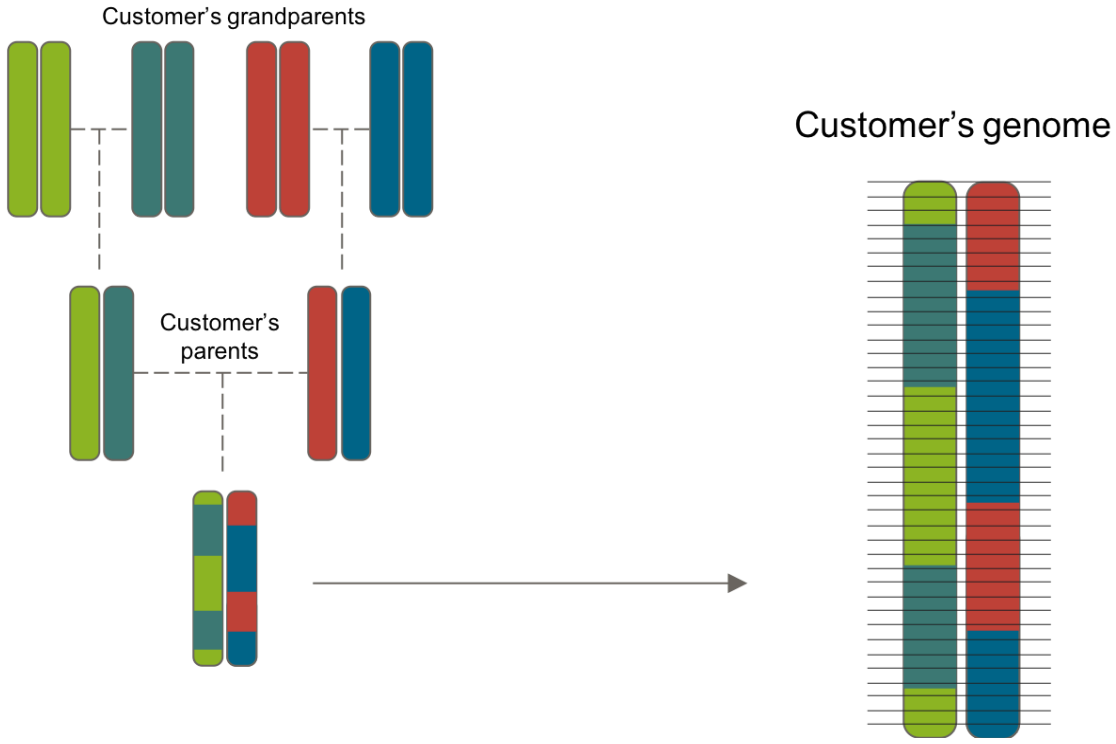


Figure 3.1: Inheritance of DNA from different populations. *On the left, we present a three-generation genetic family tree. For each individual, we show two vertical bars representing the two copies of a single chromosome present in each individual. These bars are colored to show the reference population from which they inherited their DNA. Each of the four grandparents (solid bars, top row) has inherited 100% of their DNA from a single population that is different from the other three. The DNA is passed to the parents and finally to the customer, who, through the process of recombination and assortment, ends up inheriting a shuffled set of chromosomes from each parent. The colors show that the customer's DNA is a mixture of the DNA inherited from their four grandparents, with long stretches inherited from the same grandparent. On the right, we show that to obtain a customer's ancestral regions, we divide the customer's genome into small windows (illustrated with dividing black horizontal lines). For each window we*

assign a single population to the DNA within that window inherited from each parent, one population for each parental haplotype. Our algorithm will assign a population to each window based on how well it matches genomes in the reference panel.

3.2 Phasing SNP Data

At AncestryDNA, we use microarrays to obtain DNA data from customer samples. We look at approximately 700,000 individual locations of DNA (SNPs) on chromosomes 1-22 and the X chromosome. It is important to understand that every person inherits *two* alleles, one from each parent, at each of these 700,000 sites, and that we read these sites independently. For example, we may see an A and a T at position 1, a G and a G at position 2, and so on. A crucial step in region inference is to separate which letters were inherited from different parents—a process called **phasing**. Our cutting-edge technology SideView separates DNA inherited from each parent across the entire genome. Once separated, we infer the ancestral regions inherited from each parent using the approximately 300,000 SNPs that are shared with all members of the reference panel.

SideView uses DNA shared with distant relatives across the genome to aid in the phasing. The correctness of the DNA phasing for an individual therefore relies, in part, on that person sharing enough DNA with other people in our database. Since this is not always the case, we design the hidden Markov model (HMM) we use for region inference to allow for incorrect phasing. In the next section, we explain how an HMM is useful in region inference, first with a model to analyze one parent individually, and then we show how we extend that model to account for phase error.

3.3 Principles of a Hidden Markov Model

Our goal is to assign each window of the genome to two of the 146 reference panels (one for each parent). A hidden Markov model is well-suited for this task because it can represent thousands of interrelated variables but still perform efficient inference—using a technique called dynamic programming—as each variable depends on only a few others. An HMM is a set of *states* and *transitions* connected as a directed acyclic graph (the transitions move forward along the genome and never cycle back). Each transition is associated with a probability, and each state has an emission probability, which allows the HMM to compute the *posterior* probability (i.e., taking all populations and windows into account) of individual states, individual transitions, and *paths* through the model. Figure 3.2 illustrates an HMM representing the DNA inherited from one parent for three reference populations (represented by green, yellow, and red) and six windows (our complete analysis uses 146 populations and 1,001 windows). It also shows a *path* through the model (the thick blue transitions). We use HMMs to infer the

most likely path (called the *Viterbi* path), which assigns exactly one population to each window of the genome. We also use HMMs to take *path samples*—alternative paths that are also likely—to get a better idea of how much the assignment to each population might vary according to the model. A summation of these alternative paths is reported to the customer as a set of ranges for each of these region results.

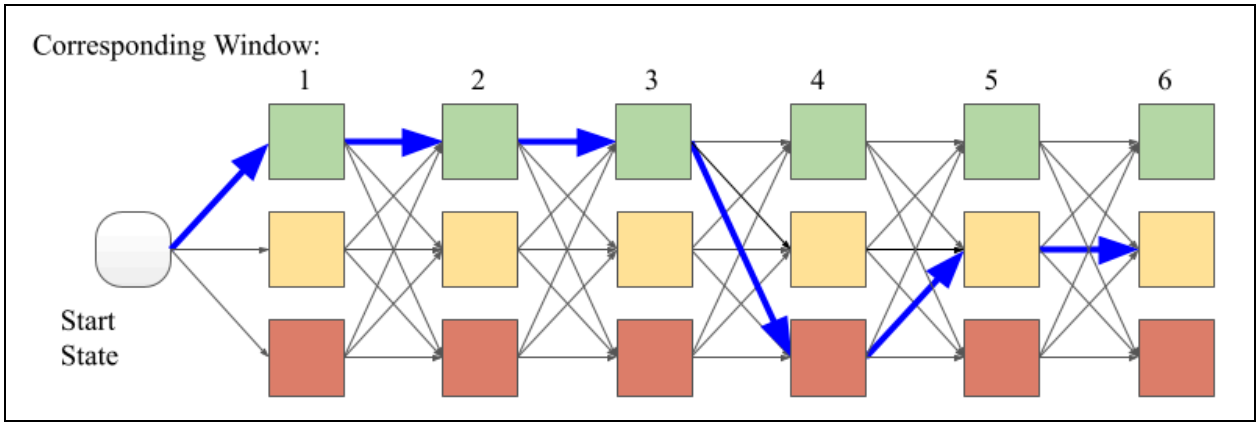


Figure 3.2: The *states* and *transitions* of an HMM representing the possible populations that explain the DNA inherited from one parent in each of several windows. *This illustration includes three populations (green, yellow, and red), and six windows. The arrows represent transitions between states, and each transition will have an associated probability. By using the transition probabilities, an HMM can compute the likelihood of each of these states and determine the most likely path through the model (illustrated by the bold blue arrows), which assigns one population to each window across the genome.*

The transition probabilities in this HMM depend on how often a population assignment should change, and, when they do change, how likely the new population is to be chosen. A transition to the same population is generally more probable in our model because the population that explains the DNA inherited from a parent is likely to be the same for several consecutive windows. However, the number of populations varies from person to person. Our HMM learns the probability of changing population states from the genotype data. When a transition does change populations, the transition probability depends also on the proportion throughout the genome of the population being transitioned to, which our approach also learns for each individual person.

The state emission probabilities in this HMM depend on the similarity between the DNA inherited from the parent and that of a reference panel corresponding to the population the state represents. We describe how we measure this similarity in Section 3.4 below.

3.4 Emission Probabilities

Determining how likely the DNA in a window came from a population (the emission probability) is described in more detail in our paper Ancestry Inference Using Reference Labeled Clusters of Haplotypes.

Briefly, our approach includes the following steps:

I. **Create haplotype models for each window.** Using a set of about 50,000 individuals representing diverse populations, we infer *BEAGLE* (Browning 2007) haplotype cluster models for each window.
II. **Annotate the reference panel.** The states in the BEAGLE models represent clusters of similar haplotypes. Because we are confident in the genetic separation of members of the reference panel, we are able to calculate the probability that a haplotype from a given population is represented by a particular haplotype cluster.
III. **Assign haplotype clusters to the test sample and aggregate the annotations.** Given a phased genotype, we observe which haplotype clusters the genotype belongs to and base the emission probabilities for a population on the weighted average annotation (how often the population reference panel belongs to the haplotype cluster, weighted so that each SNP in the window contributes equally).
IV. **Weight the emission probabilities by population.** We use results from our held-out testing data set to tune the emission probabilities so that the model produces the most accurate results possible for each population.

HMMs are used in a number of existing approaches for estimating ancestral proportions (Maples 2013). The key part of our method is step III, where we use rich haplotype models in each window, annotated with population labels from the haplotypes in our reference panel, to assign a likelihood over all population labels to the haplotypes in our test sample. It is worth noting that our method lends itself to high-throughput region inference, as steps I and II above—learning the haplotype models from a large training set and then annotating them with the reference panel populations—need only be carried out once.

3.5 Accounting for Phase Error

We use the HMM described above (Figure 3.2) to identify populations whose probability of assignment is virtually zero for one parent or the other, and we remove those from further consideration, but our final estimates are based on a more complicated HMM that simultaneously explains *both* haplotypes inherited from the parents. We need this more complicated model because we cannot be certain that every genome is completely separated into DNA inherited from each parent, since SideView cannot phase in places where an individual has no DNA matches.

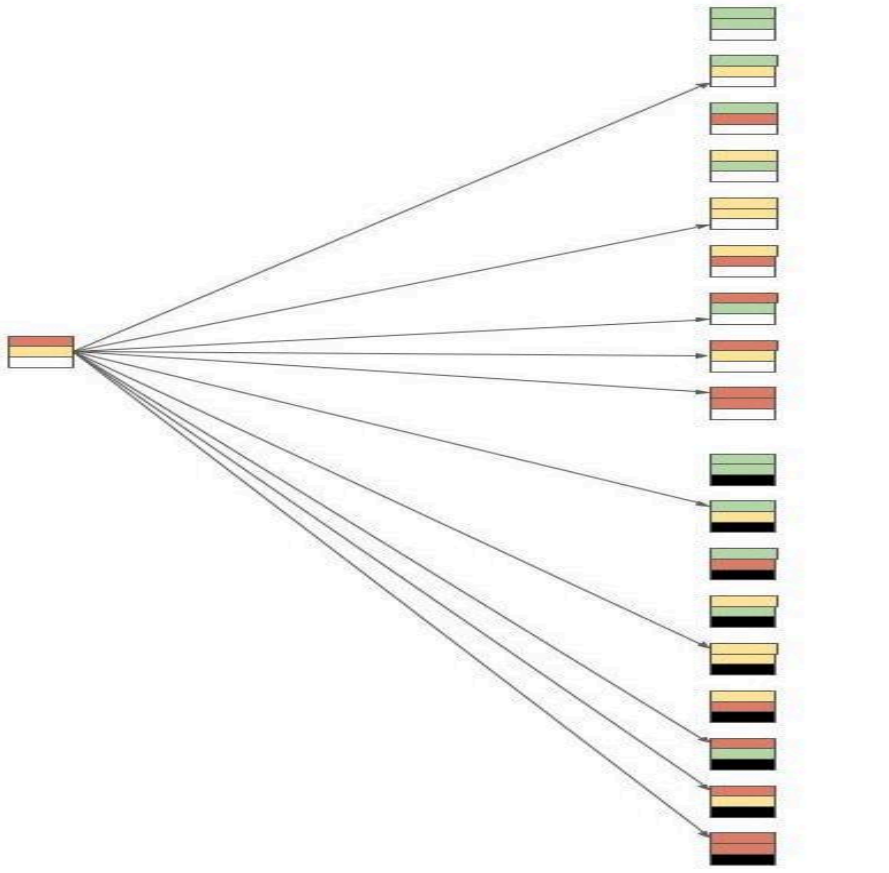


Figure 3.3: State transitions in an HMM representing *K*=3 populations. *The HMM we use in practice explains the DNA inherited from both parents simultaneously. This figure illustrates the states in a model with the same three (green, yellow, red) populations as the HMM in Figure 3.2. There are $K \times K \times 2$ states in each window. Each state represents the population inherited from parent 1 (top color of each state), parent 2 (middle), and whether or not parent 1 corresponds to haplotype 1 (bottom). Only one state is shown on the left, and possible transitions to all states in the next window (right). We only consider states such that the DNA inherited from at least one parent keeps the same population assignment.*

Figure 3.3 shows the set of states necessary for the HMM we use. Each state represents the population that explains the DNA inherited from *both* parents, and we also assign one parent to haplotype 1 in the phased data and the other parent to haplotype 2 and allow those phase assignments to change from window to window. The resulting HMM has many more states, and each state represents the population that explains parent 1's DNA (*K* possible values, if there are *K* populations), the population that explains parent 2's DNA (*K* possible values), and which haplotype corresponds to which parent (2 possible values). The HMM has $K \times K \times 2$ states for each genomic window and all possible transitions between them such that, at most, one parent's state changes population. While the constraint to one parent changing populations is consistent with biology—recombination events in different parents are independent—it is put in place mostly for practical reasons of efficient inference. The transition probability in this HMM (Figure 3.3) depends on two additional variables: the probability of changing phase from window to window and the probability of changing back. These values are also learned for each individual.

The parameters of the HMM are set based on several iterations of an expectation-maximization algorithm based on a standard HMM learning approach called Baum-Welch. For each individual, the algorithm learns (i) the probability of changing populations (for each parent), (ii) the overall distribution of population assignments (for each parent), (iii) the probability of changing phase (and changing back). The emission probabilities for each state are fixed throughout the process. Although the model allows for phase error, the model most often learns that the optimal estimate includes no phase corrections, and therefore the estimates for most Ancestry DNA customers are based on the SideView phase and parent assignments exactly.

After learning, we are able to compute through our HMM model:

1. The *Viterbi* path through the model. This is the single most likely path, according to the parameters of the model, which assigns one population to the DNA inherited from each parent in each window of the genome.
2. Probabilistic path samples through the model. These paths also assign one population to each parent in each window, and they are only slightly less likely (according to the model) than the Viterbi path, so they help describe how much or how little of a given population may still be consistent with the individual's DNA.

We report the sum population assignment for each parent according to the most likely path and report a most probable range based on 1,000 path samples taken from the model (see Section 4.5).

4. Assessing Ancestral Regions Performance

While we are developing and optimizing the estimation process, and after we finish, we repeatedly measure how well our method performs. Basically, we want to measure how close our process gets to the right answer through rigorous evaluation using a wide variety of test cases with known origins.

We use four different approaches to validate our models: 1) customer-focused simulations, 2) single-origin customers from our testing and validation sets, 3) tree-based validation, and 4) polygon creation. Each of these are described below.

4.1 Customer-focused Simulations

In any data science application, how performance is measured is the key to the algorithm's success. We use a data-centric approach to construct our testing and validation data to match the customer experience in our database.

We leverage ancestral journeys (for more information see the Genetic Communities white paper), to identify customers who share similar family histories. By aggregating 10s to 100s of thousands of family trees, we are able to identify accurate patterns of admixture between populations that differ for each group. We can then simulate separate test and evaluation data sets of genotype information based on these admixture patterns, where we will also know the region results. For populations with admixture patterns that are not captured in the pedigrees, such as African American or Latin American populations, we use historical information to guide the simulations, such as in *Mooney et al.* (2023).

After analyzing the simulated data with our model, we compare the output from our model to the expected, ground-truth results. We measure three different statistics in aggregate and per population:

1) **Overlap** – the observed percentage for a region divided by the expected percentage for a region. Note that if the observed percentage exceeds the expected, the overlap will be above 100%.
2) **Recall** – the proportion of expected regions that are observed in the output. E.g., For 100 simulated people, their pedigrees include an ancestor from Norway. Only 75 of these people have Norway in their results. The recall for Norway would be $\frac{75\ observed}{100\ expected} = 75\%$.

3) **Precision** – the proportion of observed regions that are expected. E.g., For 100 simulated people, their results include Leinster, Ireland. Only 90 of these people have an ancestor from Leinster in their pedigree. The precision for Leinster, Ireland would be $\frac{90\ expected}{100\ observed} = 90\%$.

As we tune our models, we balance the performance of the overlap, recall, and precision statistics overall and per population. For example, as we increase the recall and overlap for one region, we often see a decrease in the precision at the same time. Our goal is to maintain as high recall as possible, while not sacrificing precision.

We note that recall and precision behave differently for regions that are assigned at a very low percentage. Therefore, we use a cut-off of 5% assignment to a region to report on performance. Expected and observed values that include regions assigned below 5% have a much higher error and missing rate than those that include regions assigned above 5%.

Here, we report a few numbers from a handful of our simulations from our final evaluation:

Table 4.1: Results from a simulation of 30,249 admixed European and African American individuals.

Region	Mean Overlap	Recall	Precision
Leinster, Ireland	67.76%	75.84%	78.85%
Donegal, Ireland	120.27%	98.90%	79.82%
Connacht, Ireland	112.26%	95.15%	75.35%
Munster, Ireland	121.41%	95.48%	86.31%
North East Scotland	70.09%	65.64%	68.21%
Hebrides & Western Highlands, Scotland	75.31%	78.51%	83.51%
Central Scotland & Northern Ireland	126.73%	89.04%	88.81%
Southern Wales	116.50%	97.42%	83.47%
Isle of Man	50.98%	93.98%	98.73%
West Midlands	115.97%	79.98%	66.86%
East Midlands	96.64%	79.03%	92.73%
Southeastern England & Northwestern Europe	98.71%	82.38%	96.75%
Northern Wales & North West England	115.84%	88.01%	87.06%
North East England	88.52%	84.02%	37.44%
Devon & Somerset	67.69%	59.68%	78.66%

Cornwall	68.34%	72.24%	70.18%
Madeira	75.86%	98.97%	99.31%
Azores	82.52%	97.19%	97.67%
Portugal	126.44%	98.99%	64.20%
Spain	70.33%	98.36%	93.22%
Canary Islands	51.49%	96.81%	100.00%
Northern Spain	86.86%	97.87%	82.51%
Basque	94.62%	100.00%	98.83%
Southern Germany	79.47%	77.38%	91.28%
Northwestern Germany	109.74%	84.13%	70.03%
The Netherlands	101.12%	97.48%	35.80%
Quebec	71.18%	83.93%	98.13%
Acadia	105.82%	99.85%	99.96%
Brittany, France	32.50%	46.58%	97.14%
France	39.49%	71.51%	88.57%
Northwestern Italy	102.70%	96.00%	89.78%
Northeastern Italy	92.87%	96.26%	82.76%
Southern Italy	84.97%	86.30%	98.41%
Central Italy	113.71%	93.41%	86.61%
Sicily	68.75%	79.25%	99.92%
Sardinia	105.17%	100.00%	81.82%
Malta	116.30%	99.43%	100.00%
Ionian Islands	79.34%	100.00%	97.73%
Crete	93.45%	98.96%	98.96%
Aegean Islands	93.67%	98.69%	91.52%
Southern Greece	92.62%	95.29%	75.83%
Northern & Central Greece	87.83%	98.33%	97.51%
Albania	78.26%	99.38%	96.99%
Southwestern Balkans	120.57%	100.00%	58.72%
Romania	68.36%	98.81%	88.30%
Slovenia	90.64%	100.00%	64.29%
Northwestern Balkans	95.67%	97.07%	80.57%
Western Balkans	68.53%	96.15%	98.04%
Southern Poland	93.31%	90.20%	84.59%

North Central Europe	117.43%	94.19%	44.90%
Northeastern Poland	89.67%	88.04%	58.70%
Eastern Czechia	70.43%	100.00%	7.06%
Slovakia	77.84%	94.64%	44.92%
Western Ukraine	78.18%	94.85%	53.18%
Russia	68.40%	94.82%	99.80%
Lithuania	96.83%	99.55%	66.27%
Estonia & Latvia	100.55%	99.34%	87.79%
Iceland	108.82%	100.00%	97.24%
Norway	125.08%	97.39%	95.00%
Denmark	78.99%	84.44%	63.96%
Sweden	115.62%	95.61%	95.85%
Finland	114.19%	100.00%	98.11%
Khoisan, Aka & Mbuti Peoples	101.45%	100.00%	100.00%
Eastern Bantu Peoples	63.43%	97.22%	100.00%
Southern Bantu Peoples	105.20%	100.00%	100.00%
Twa	119.69%	100.00%	100.00%
Western Bantu Peoples	57.49%	97.63%	100.00%
Cameroon	115.18%	100.00%	99.32%
Nigerian Woodlands	104.20%	99.48%	100.00%
Nigeria	109.87%	97.06%	100.00%
North-Central Nigeria	98.64%	96.77%	100.00%
Central Nigeria	115.99%	99.15%	99.15%
Central West Africa	126.26%	100.00%	100.00%
Yorubaland	125.52%	100.00%	100.00%
Benin & Togo	129.29%	100.00%	97.18%
Ivory Coast & Ghana	75.33%	88.10%	100.00%
Mali	87.96%	97.31%	100.00%
Senegal	90.96%	100.00%	100.00%
Per Individual (mean)	73.30%	88.17%	88.30%

Overall, we see very strong performance across all regions, with most having precision values greater than 90% and overlap between 80% and 120%. In only a few regions, like *Slovakia* and *Eastern Czechia*, do we see precision values below 50%, demonstrating that the majority of the time we report the correct

regions to users based on their origins. We also see very strong recall and precision values across the Africa regions, suggesting that assignments greater than 5% to these regions indicate a very confident link between a customer and that population.

We see that, on an individual level, the average expected percentage overlap is 73.03%, which is consistent with the performance of our 2024 model.

4.2 Single-origin evaluation

Another way to access the performance of our model is through our evaluation dataset. For each of our reference panels, we create a testing dataset of up to 500 people to train the model weights, and a validation dataset of up to 500 people to evaluate the final model. Like the individuals used to create our reference panels, the people included in the testing and validation datasets are believed to be of a single origin, and are expected to receive 100% assignment to a specific region. We can assess each region for overlap, precision, and recall as before. After fully tuning our model, we measured the following performance metrics (Table 4.3). Regions not updated in the 2025 model are not shown.

Table 4.3: Results from 17,625 single-origin evaluation individuals.

Region	Overlap	Precision
Leinster, Ireland	57.57%	39.01%
Donegal, Ireland	92.77%	33.78%
Connacht, Ireland	83.17%	44.79%
Munster, Ireland	95.45%	43.85%
North East Scotland	72.80%	82.75%
Hebrides & Western Highlands, Scotland	64.00%	37.43%
Central Scotland & Northern Ireland	87.09%	30.36%
Southern Wales	94.29%	69.68%
Isle of Man	58.44%	100.00%
West Midlands	72.69%	37.68%
East Midlands	76.69%	50.25%
Southeastern England & Northwestern Europe	73.57%	23.04%
Northern Wales & North West England	72.00%	47.62%
North East England	74.89%	54.35%
Devon & Somerset	59.43%	55.95%

Cornwall	71.17%	61.10%
Madeira	81.90%	89.66%
Azores	91.34%	91.69%
Portugal	92.43%	45.62%
Spain	57.27%	33.12%
Canary Islands	54.42%	96.72%
Northern Spain	66.89%	29.07%
Basque	94.46%	85.14%
Southern Germany	71.53%	35.19%
Northwestern Germany	79.82%	36.46%
Russian Germans	96.95%	100.00%
The Netherlands	89.35%	42.60%
Quebec	86.99%	96.71%
Acadia	87.02%	89.50%
Brittany, France	17.15%	100.00%
France	45.70%	34.12%
Northwestern Italy	94.36%	80.23%
Northeastern Italy	91.42%	83.91%
Southern Italy	89.53%	55.87%
Central Italy	91.43%	72.14%
Sicily	78.01%	95.06%
Sardinia	98.44%	97.30%
Malta	97.90%	97.25%
Ionian Islands	77.48%	85.71%
Crete	92.28%	92.00%
Aegean Islands	87.87%	87.44%
Southern Greece	88.54%	64.02%
Northern & Central Greece	81.60%	51.85%
Albania	74.40%	36.78%
Eastern European Roma	97.15%	100.00%
Southwestern Balkans	97.18%	45.90%
Romania	78.64%	90.91%
Slovenia	79.07%	54.05%
Northwestern Balkans	71.49%	71.46%

Western Balkans	72.07%	37.87%
Southern Poland	85.06%	34.18%
North Central Europe	62.85%	46.76%
Northeastern Poland	70.28%	25.90%
Eastern Czechia	74.53%	70.18%
Slovakia	66.23%	60.42%
Western Ukraine	74.13%	49.70%
Russia	73.90%	94.96%
Lithuania	89.20%	46.67%
Estonia & Latvia	94.76%	23.12%
Iceland	96.93%	93.59%
Norway	95.74%	72.99%
Denmark	72.59%	48.94%
Sweden	92.56%	64.77%
Finland	98.48%	75.12%
Sephardic Jews in Northern Africa	93.74%	70.87%
Sephardic Jews in the Eastern Mediterranean	72.77%	86.67%
Ashkenazi Jews in Central & Southeastern Europe	90.22%	47.39%
Ashkenazi Jews in Eastern Europe & Russia	65.44%	63.53%
Per Individual (Mean)	84.29%	72.01%

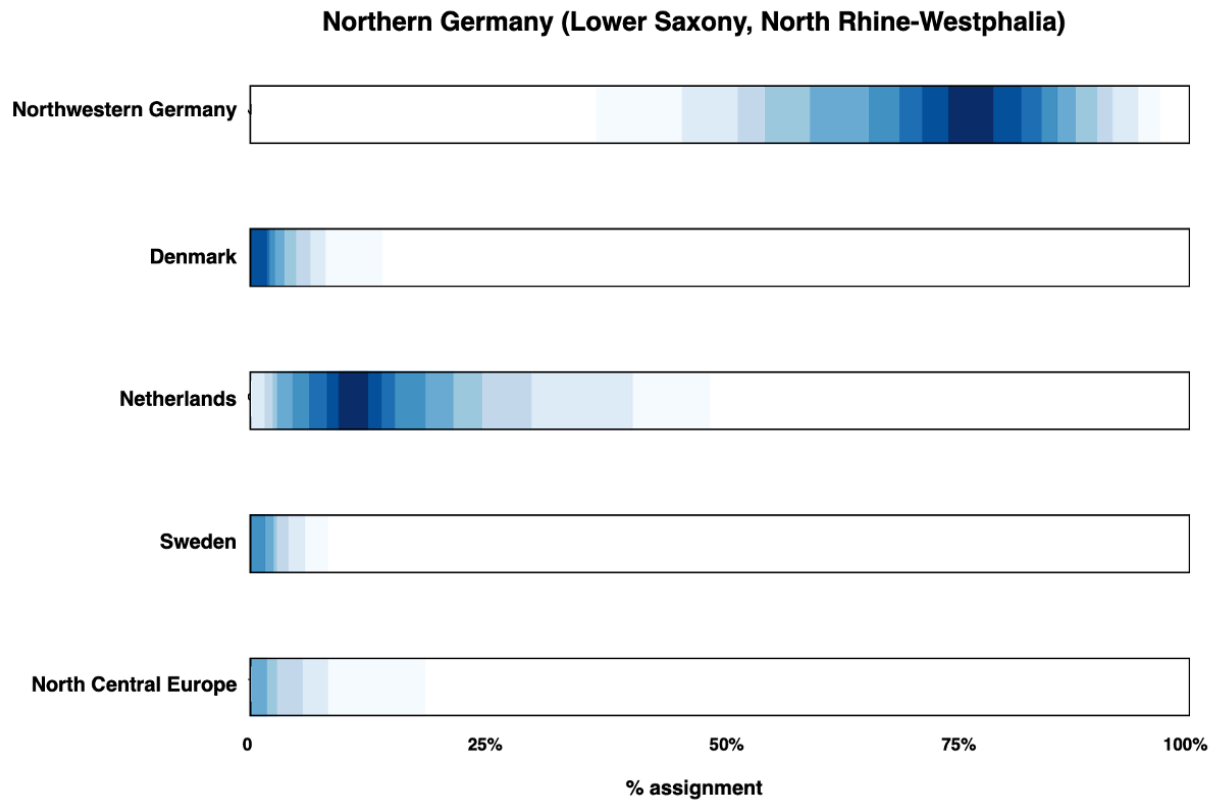
Overall, we found that the majority of our new and updated regions have precision greater than 60% and overlap greater than 80%. The per individual results are also comparable with values from our 2024 model, indicating a consistent performance year-to-year. In summary, although our models are tuned based on admixed samples, we are pleased to report a consistently strong experience for people of single-origin.

4.3 Tree-based validation

An independent way to validate our model is to look at the ancestral regions results of people with deep genealogical roots back to the same country or part of a country. To find these individuals, we use customer-created family trees and look for customers who have consented to research and have all of their ancestors from the same country. Ideally, we'd only look at people with all of their grandparents (or

older) from the same country, but due to low numbers for some countries we sometimes include people where only their parents are from the same country.

Customers who are not in the reference panel and have deep trees tracing back to a single country are expected to have high assignments to the regions associated with that country, and this is what we generally find for the more than 500 regions of the world that we considered. For example, Figure 4.1 shows the average assignments for approximately 200 customers with all four grandparents (or older) born in Northern Germany (top) and approximately 200 customers with all four grandparents born in Norway (bottom). As you can see, while most of their assignment is to the expected corresponding regions, *Northwestern Germany* and *Norway respectively*, other regions do appear in small but appreciable amounts. These analyses help ensure that results for people from a geographic area agree with expectations.



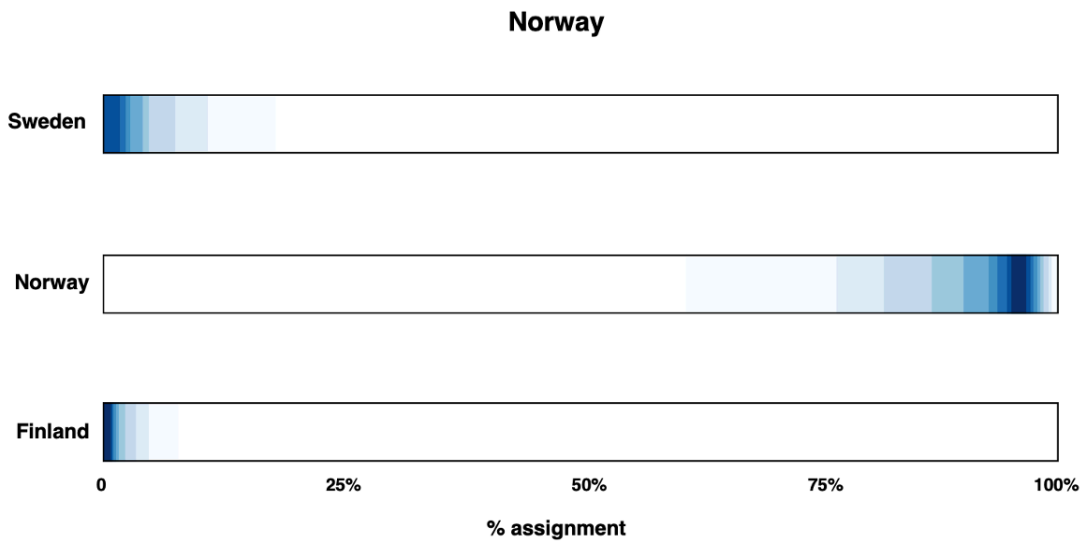
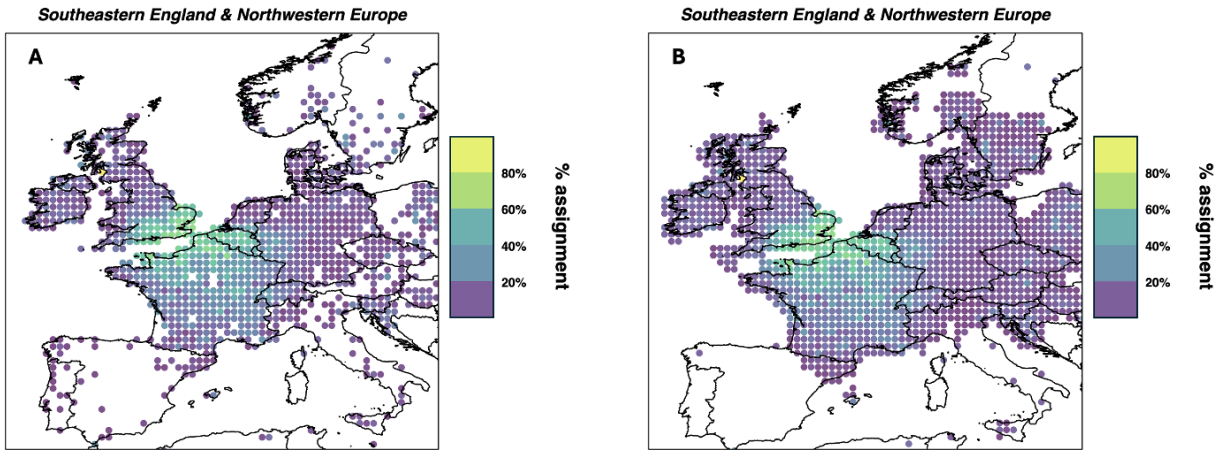


Figure 4.1 Average assignments based on grandparents' birth location. *Region assignment distribution for customers with all four grandparents born in the same country or region. Northern Germany (top), and Norway (bottom). Dark blue is the middle 50th percentile, with the distribution bucketed and colored by percentile. The analysis indicates that on average individuals with roots to northern Germany will see ~75% Northwestern Germany and ~10% Netherlands, and those with roots to Norway will see ~95% Norway. In both cases, individuals tend to see some small percentage assignment to other neighboring regions.*

4.4 Regional Polygon Construction

The process we use to create polygons for each of our 146 regions also helps to validate our model. Where possible, we use the known geographic locations of our samples to guide how we create the regions. Figure 4.2 shows an example of the results and geographic information used to define the polygon for our *Southeastern England & Northwestern Europe* region.



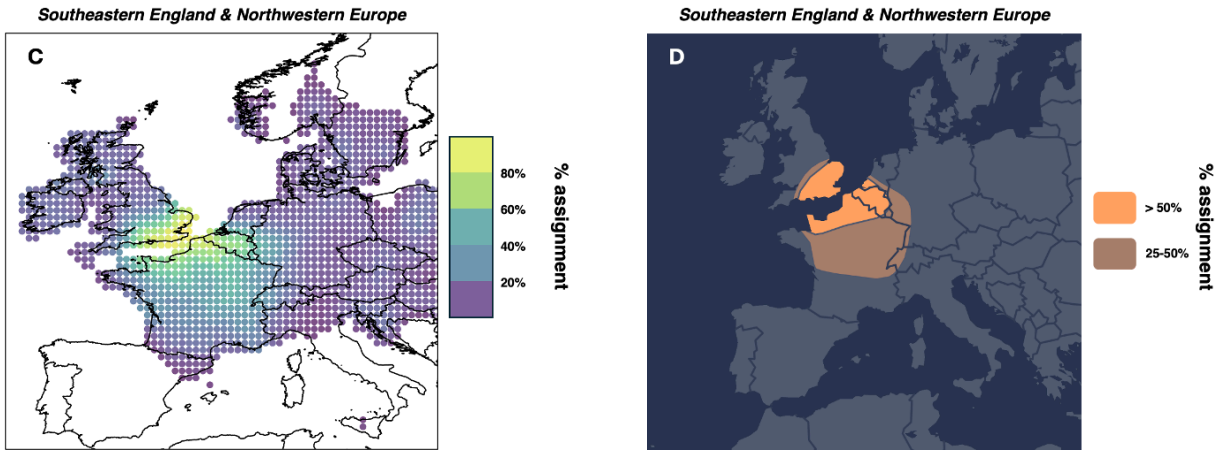


Figure 4.2: Using geographic sample locations to draw regional polygons. *Panel A shows the distribution of the Southeastern England & Northwestern Europe region predicted for a set of samples with geographic information. Samples are assigned to grids of 0.5 degrees longitude by 0.5 degrees latitude based on the average birth location of their grandparents. The color of each grid point on the map represents the average percentage of Southeastern England & Northwestern Europe for samples from each grid. Panel B shows the maps after filling in missing grids using an imputation method. Panel C shows the information processed with further smoothing, creating the outlines representing the ancestral regions shown to customers. Panel D shows the final polygon presented in a customer's results.*

In Figure 4.2A, we show the amount of our *Southeastern England & Northwestern Europe* region assigned to a combination of reference panel evaluation samples and customers with deep roots from the same country. Figure 4.2B and C show the results after imputing values to fill in gaps in our map grid and applying smoothing methods to make the plot less spotty. It is clear from the plot that there is a gradient of assignment in this area that is centered in England and quickly tapers off in surrounding areas. For example, the highest level of assignment, represented by a green yellow in Figure 4.2C, is in northeastern France and Belgium. The gradient continues to diminish as represented in purple, with the borders reaching as far away as Northern Italy, Norway, and Switzerland.

Manual edits are sometimes performed on polygons to better align them with geography like narrow peninsulas or when the polygons may imply finer-scale population structure than the underlying genetic data support. Additionally, polygons representing some of our regions have hand-drawn components to describe minority populations that may not be explicitly defined by geography. For regions which are data driven, these polygons are a powerful tool that we use to validate each one of our regions.

4.5 Reporting uncertainty of estimated values

As mentioned in Section 3.5, we report a range for each ancestral region that we deliver to customers. For example, we might report someone as 40% *Southeastern England & Northwestern Europe* with a range of 30-60%. This means that the model reports the most likely estimate of 40% *Southeastern England & Northwestern Europe*, but that our model also supports an estimate anywhere between 30% and 60% *Southeastern England & Northwestern Europe*. We run a separate analysis to validate the range results in our simulation datasets to ensure that the expected value is captured within the range most of the time.

5. Future Refinement

While AncestryDNA is extremely proud of the updates in this release, we plan to improve the product over time. The availability of new data, the development of new methodologies, and the discovery of new information relating to patterns of human genetic variation will all enable future improvements to the product. Each new release of genetic ancestral regions will represent a step forward in our ability to give our customers a complete description of their heritage and inform them about their genetic origins. We hope that, like the entire team at AncestryDNA, our customers will look forward to these future developments.

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