U106 explored: its relationships, geography and history

Report to the U106 group, Mar 2016 edition

Principal investigator: Iain McDonald

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Methodology & Background

WARNING!

Everything presented in this document is wrong. The question is: how wrong? It is my hope that there is enough in here that is only a *little bit* wrong for it to be useful.

The dates I compute here were outdated before I finished compiling the information. More tests have resolved more branches of our family tree. Further archaeological DNA was published that altered the subtleties of the migrations portrayed at the end of this document. This is a very active field of research, advancing at an impressive rate. So please take everything in this document with a hefty pinch of salt and note of skepticism.

FOREWORD

The decisions of our ancestors have shaped the world we live in today, whether that decision was to fight or flee, what to hunt, who one should marry, or simply what to get for breakfast. Those countless decisions became manifest in their successes or failures in life, whether they survived to produce a family, and ultimately the fact that some of those families prospered and eventually produced you and I. In this work, we attempt to re-trace some of those decisions back in time to find who our ancestors were, and what decisions were made that allowed them to play a critical role in the history of Europe and the wider world.

Our particular story here concerns a family whose descendants carry a particular genetic mutation - worn like a molecular badge - which allows us to identify them as sharing a single common ancestor in which this mutation first arose. That mutation is named U106, or alternatively S21, and is the result of a simple typographical error that happened around 4500 years ago, where one encoding molecule was misread among the 59 million that made up the Y-chromosome of a particular cell. That cell grew into a man, and that man is the 125-times great-grandfather (or thereabouts) of about one in eight men of European descent today.

This document attentions to trace his descendants and the paths they took throughout history, and maps their distribution throughout Europe close to the present day.

METHODS: DNA TESTING BASICS

This report is an analysis of the Y chromosome, which is passed from father to son. It is therefore only a study of male lines: a person's father's, father's, father's, … father. It can therefore be used to trace the history of a surname, and uncover "superfamilies" which were founded before the age of surnames.

DNA is made up of four bases: A, C, G and T, and can be read out as a string of these letters. A single person's DNA means nothing. Genetic genealogy relies on a comparison between two or more people's DNA. The differences between them identify mutations that have happened in the transfer of the genetic code from parent to child. These mutations can be used to work out relationships, and the time since their most-recent common ancestor (TMRCA).

METHODS: Y-STR TESTING

There are two different kinds of DNA that are used to determine people's relationships to each other. The most commonly taken is an STR (Short Tandem Repeat) test, advertised at Family Tree DNA as a series of 12, 25, 37, 67 or 111 markers. These markers take the form of a short section of DNA that repeats a certain number of times. Mutations can cause this number to increase or decrease. A hypothetical example would be:

`DYS1234 = 4 TACATACATACATACA`

which could mutate to:

`DYS1234 = 5 TACATACATACATACATACA`

by gaining a repeat.

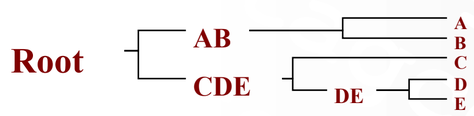
If most people have DYS1234=4 and some people have DYS1234=5, we presume that "4" is the ancestral value and that the people with "5" are more closely related.

Things are rarely that simple, as the same mutation can happen in different branches, STR markers can mutate back to their ancestral values, and a lot of poorly understood factors make them prefer certain values over others. For these reasons, they stop being very accurate tools on long timescales, and are not absolutely foolproof for creating these family groups. We tend to need two or more shared mutations to ensure a person belongs to a specific group.

Using a series of these mutations, we can build a relationship tree for families, e.g., for:

DYS	393	390	19	391	385	426	388	439	389i	392	389ii
A:	13	24	14	11	11-15	12	12	12	12	13	29
B:	13	24	14	10	11-15	12	12	12	13	13	29
C:	13	24	14	11	11-14	12	12	13	13	13	29
D:	13	23	14	11	11-14	12	12	13	13	13	29
E:	13	23	14	11	11-14	12	12	13	13	13	29

we presume the group CDE are more closely related because of the DYS439=13 mutation, with DYS390=23 defines are group within this (DE). DYS385=11-15 defines another group (AB). Thus:



METHODS: Y-SNP TESTING

The second test we perform is Y-SNP testing. Outside of the repeating STR regions, DNA is more of a genetic jumble. As material is passed down, parts of the code can be inserted:

`ATGCTGATCGC → ATGCTGATAGATCGC`,

deleted:

`ATGCTGATAGATCGC → ATGCTGATCGC`,

or mutated:

`ATGCAGATCGC → ATGCTGATCGC`.

Sites of these latter mutations are known as a single nucleotide polymorphisms, or SNPs. These SNPs are very reliably passed on from father to son, so they can clearly identify a family branch without the ambiguity than STRs provide.

SNPs can be tested individually through Sanger sequencing, as used conventionally by Family Tree DNA and YSeq. They can also be tested *en masse* and new SNPs discovered through 'second-generation' tests such as the Illumina dye sequencing used in Family Tree DNA's BigY or Full Genome Company's Y Elite and Y Prime.

We use these SNP tests to create the backbones structure of the human Y-DNA tree, draping over it the STR results of all testers to flesh out the branches. For a full understanding of the human male-line family tree, we require comprehensive SNP testing of every branch, backed by STR results to compare with the larger STR databases.

FUNCTION OF THE U106 GROUP

The U106 group facilitates these comparisons by providing a place where individual testers can share their data, regardless of the company and country of origin. The group provides expertise to analyse that data, and can make recommendations for people to get the greatest return from each test. By collecting this data together, we provide a sample size greater than almost every professional study (even though it is not so homogeneously sampled as such studies).

Although U106 encompases a lot of people, perhaps 3% of human male lines, it is a comparatively small twig of the human Y-DNA tree. By focussing on this single twig, we can provide a greater depth of analysis and understanding than broader-ranging professional scientific studies are able to, and drill deeply into the recent history of individual families.

This approach relies on the generosity of individuals who are willing to share the details of their genome with the community. In return, they get to learn more about their family history. This work would not have been possible without them. Thanks are also due to the rest of the U106 team - primarily Charles Moore and Raymond Wing - and David Carlisle and Andrew Booth for sorting the details of BigY. Kudos also goes to Dr. Tim Janzen and Prof. Ken Nordtvelt for detailed help with different aspects of STR age analysis, and to John Sloan of the U198 project and James Fox of the L1 project for collaborative help with their data. Finally, thanks to the innumerable members of the U106 Yahoo forum who have contributed in many different ways to the success of this project.

General testing advice

Everyone's situation is different. The best testing route depends on your budget, on your DNA matches, on their budgets (or your ability/willingness to pay for them), on what you are hoping to get out at the end, and what you can realistically achieve given the limits of money, people and technology. Everyone's journey is different.

What do you want to do with DNA testing? Find the origin of your immigrant ancestor? The origin of their surname? Find out when and how their ancestors arrived in Britain (or any other country)? Or perhaps find out what yur deep prehistoric roots are. I'll address these points below, but please bear in mind that this testing advice is general: use your judgement to determine whether this applies to your particular situation.

The testing advice to most people is fairly similar: maximise what you can find out about your own DNA, then carefully select people around you to upgrade – either at their expense or yours. This strategy stems from the basic principle that DNA is a comparative science: your results only mean something if you have someone else to compare them to. You will make the best progress by taking charge and directly engaging with the people to whom you are most closely related. The following testing advice is written from the point of view of U106, but it can readily be translated to any other haplogroup.

PREHISTORIC AND EARLY HISTORIC ROOTS

Are you Celtic or German, or even Saxon, Norman, Viking, Flemish, Angle, Jute, or maybe something else? Obviously, your Y-DNA is only a very small part of that story. Even only 600 years ago, your Y-DNA will account for less than 0.001% of your ancestry. But we are beginning to unravel the ancient roots for many Y-DNA groups. The Family Tree DNA haplogroup projects operate at the interface between amateur genetic genealogy and professional genetic anthropology. Both of us are trying to uncover the (pre-)historical migrations of the world: the professionals focus on the large-scale structure, whereas our interests are typically more specialised.

Step 1: SNP testing. The process of discovering your prehistoric ethnic Y-DNA ancestry relies on grouping together people with the same SNP mutations to form new branches of the human haplotree. The best thing you can do here is take a next-generation test like BigY or YElite to uncover the SNPs in your line. If you can't afford these tests, test the appropriate SNP pack(s) at Family Tree DNA or Yseq, or take a Chromo2 test. YSeq is generally cheaper: this is especially true for people who are probably or confirmed to be U106+, but who don't know which part of U106 they belong to. The U106+L48 pack covers all of U106, and only one pack needs tested, rather than two. For tests with companies other than Family Tree DNA, you should report your results to your haplogroup administrator(s). ***You should only order an SNP pack if you do not intend to take a next-generation test, or you will be paying for the same thing twice.***

For anyone in R-M269, any of these tests will probably take you what is happening in the period between 5000 years ago and about 2000 years ago. Some people are lucky, and have well-populated SNPs that are less than 1000 years old; some people are stuck in rare clades that are over 4000 years old. Either way, you will find yourself related to one or more people on your "terminal"* SNP. Your goal is to find people who are related to you more closely.

(* Terminal is a bad word to use here, but common parlance. It refers to the most-recent SNP you share with someone else. You will also find a lot of SNPs that are discovered in only your test, which we term "singletons". You want to find people who share some of these singletons with you.)

Step 2: fully upgrade your STR markers, and find your close matches. To find people, you need close matches. Most people have only taken STR tests, not SNP tests. If you have not done so already, upgrade to 67 or (preferably) 111 STR markers. This will help you identify the maximum number of matches. Don't worry if you don't match anyone at 37 or 67 markers, you will match someone somewhere, and the more markers you test, the more we can beat down the random noise in the mutations to see who you match. Matches beyond the Family Tree DNA system can be found using YSearch, or Semargl.me.

If you feel capable, try to identify the STR mutations you have in common from an older modal (e.g. the U106 modal). You can then use this template on YSearch.org, semargl.me or simply the table of results from Family Tree DNA projects, to identify those people who also share some or all of these mutations. If you aren't happy doing this, identify the genetic distances of the people who are positive for your "terminal" SNP (or upstream SNP if you have none), then look for people who have not tested SNPs who match you with a smaller genetic distance.

Step 3: encourage your close matches to upgrade. If you have taken a next-generation test (BigY/YElite/etc.), you should also ensure your closest match has upgraded to the same number of STR markers as you. If you have taken an SNP pack, encourage your STR matches to take the same SNP pack or test your individual "terminal" SNP. From hereon, I shall assume you have taken a next-generation test, as the following steps otherwise don't make much sense.

You should then encourage your close STR matches to upgrade to BigY or YElite. They will hopefully share some of your singletons, and give you a new "terminal" SNP. If you can't cajole them into taking these tests, or pay for them, then you should encourage them to test your singleton SNPs at YSeq.net. You should particularly concentrate on those people who have European ancestry, as they can tell you where your "terminal" SNP originated. Devote even more energy to those from continental Europe, where our coverage is poorer, and especially places like France and eastern Europe where legal or economical reasons mean we have very few testers.

In this way, you will gradually bring what is known about your history forwards towards the present day. How far forward this will bring you depends on the number of matches you have, and how willing they are to upgrade.

HOW DID YOUR ANCESTORS ARRIVE IN BRITAIN?

The testing advice for this is goal pretty much the same as that above. Crucially, however, you need to target the period when your ancestors are likely to arrive in Britain. This will depend on your haplogroup. For most U106, particularly those in Z8, it will typically be in the range 800-2100 years.

You need to start by working out your closest STR and SNP matches from continental Europe. You need to find phylogenically when you were last related, by testing yourself and them with either a next-generation sequencing test (BigY/Yelite) or SNP packs, and get them to upgrade their STR results to match your own if necessary.

This gives you an upper limit for the length of time your family has been in Britain, though it is only valid if your continental matches did not migrate back to Europe. This seems comparatively rare but obviously did happen.

Generally speaking, we don't yet have enough data to determine that SNPs are specifically British, although there are a few recent cases (within last 1000 years or so for U106) where we can be quite sure. The biases in our samples mean that it only takes one distantly related tester to cast significant doubt on the "Britishness" of any SNP.

For U106, this is something we are actively researching as a group, where can apply statistical techniques across U106 to say groups are likely to be related on a particular timescale.

RECENT RELATIONSHIPS: SURNAME ORIGINS & ANCESTORS OF AMERICAN IMMIGRANTS

These problems are very much related (no pun intended). If you want to origins this recent, you obviously have one more piece of information: your surname. This means you can think a bit differently. You might also want to consider anyone who is expected to match you within the last 1000 years (refer to comparison tools like http://www.mymcgee.com/tools/yutility111.html or ages estimates like those from YFull or (for U106) my analyses). I will presume that your ancestors originate from the British Isles, but the same basic principles can be applied to other locations.

Step 1: upgrade your own DNA. The first thing to do is see what you can learn from your own DNA. Upgrade your STR markers to a reasonable level (67 or 111). This will let you accurately see who you match. Matches at lower levels can be spurious, unless you are lucky enough to have significant numbers of historical STR mutations in your first 37 markers. It is also impossible to get a sufficiently accurate TMRCA (time to most-recent common ancestor) using less than 67 markers. Even at 67 markers, the age of relationships will be uncertain by about a factor of two.

You should also consider at least an SNP pack to see which rough population you belong to, although BigY or YElite would be preferable. Both types of SNP tests may help identify clues at the 1000-2000-year-old level that can inform later discussion, even if we don't fully appreciate them at present. These tests can let you design a bespoke testing strategy that may end up cheaper in the long run.

While these SNP tests are useful, they are generally less important. If money is an issue and you have a good idea of where to look don't feel pressured to upgrade to BigY: it's a balance between the cost of one expensive SNP test, and the eventual cost of later steps.

Step 2: find your STR matches. The next thing to do is to find matches. If Family Tree DNA do not provide any close matches, you may have to look to external sites, such as YSearch.org or Semargl.me. Typical maximum genetic distances you should look for are less than 10@111 markers, 6@67 markers and 2@37 markers, but those appropriate to your particular situation will depend on the number of mutations your line has acquired in the last 1000 years.

If you are fortunate enough to have existing matches at 37, 67 or 111 markers who share your surname, you should encourage your existing matches to upgrade to the level of your own DNA tests. You should also do this for very close matches who do not share your surname, but are predicted to be related to you within the last 1000 years. Do not go beyond 1000 years without SNP testing to confirm that your relationship really is that recent, as the STR genetic distance calculators typically start to fail at around this age.

Step 3: identifying likely geographical origins. The next steps will pin down the geography of your surname. This is the most difficult, and perhaps the most expensive bit. Your success, and the order you do things in, depends on the frequency of your surname, whether you have any additional information (like a surname which may by topographic in origin), the number of existing matches you have, and how many people you can convince to pay for themselves. The rest of this is therefore even more generalised advice.

If you genuinely have no idea where in Britain to start, search for your surname here:

http://gbnames.publicprofiler.org/Surnames.aspx

and identify the regions where it is most common. If there is only one region, that gives you a good idea of where to start. If not, you will have to pick off these regions one by one.

Once you've found a region, you need to narrow things down by parish. You can also use resources like FreeCen, the IGI and Scotland's People to map the number of people sharing your surname by parish. On some pay-per-view sites like Scotland's People, searching is still free, so it is still possible, if laborious, to obtain numbers. Old historical records can be very useful too, such as pre-1841 tax registers. Mapping the number of people with your surname (or its variants) in these will typically show one or more "hot spots" on a parish level that you can work with further.

Common surnames pose more of a problem. A case study in this is my own historic surname, Donald, which is quite common in Scotland. There are at least 11 different origins for the Donald surname in Scotland. Mapping all of Scotland at the parish level has also identified at least 11 distinct "hot spots". My family traces to Aberdeenshire, as does that of my closest non-Donald match. Even within a few miles of Aberdeen, there are three distinct geographical groups that show up, that resolve into at least four different origins for the surname. In such cases, a lot of testing needs to be done to identify the relationship between geographic and genetic groups.

Step 4: probing "hot spots". The next step is to get DNA from people in these hot spots. If you are lucky, some people will already have taken DNA tests which will let you see if people from these regions are related to you or not. If you are unlucky, which will be most of the time, you will have to go and encourage people to take up DNA testing who haven't done already.

A good source of such people is the wider genealogy community. The important thing is to target people who have long, secure and unbroken lineages who share your family surname and who either still live in Britain or can trace their ancestry back to a particular place in Britain. Sites like Ancestry and other genealogy websites are useful for identifying such people.

You will then need them to test their Y-DNA, and convincing them to do so means you need to formulate the cheapest, most reliable test for them. This will depend on whether you have any mutations that uniquely identify your family in the first few markers. If you do, a 12-marker test and a single SNP test might be the cheapest way. Otherwise a 37-marker test might be preferable. (NB: you now have to ask for 12-marker tests specially.) You could even just get them to test a single SNP test for your "terminal" SNP at YSeq.net, but this will simply give you a binary indicator of whether or not they are related to you. A wider test will give them some extra information about their origins regardless of whether they are related to you or not. Remember that if they are paying, they are relying on you to provide good advice, so remember to act in their interests as well as your own!

Step 5: detailed exploration. Once you have covered all the "hot spots", you should find out where people are related to you, so roughly where your ancestors came from. You should then focus on this region, and get a few people with your surname to test (and surnames of your closest STR matches if applicable).

This will give you some idea of the breadth of variation in STR types, and which STR mutations happened when in your family. You should then construct a phylogenic family tree, showing how your lines fit together and when each mutation is likely to have occurred. Note that this may require SNP testing to accomplish: first a next-generation test (BigY/YElite) to identify novel variants, then single SNP testing at YSeq.net to identify who you share those variants with. Try to reconstruct family trees of everyone with your surname from these regions as best you can, and identify lines to research further. Very few people can expect to extend their paper trail via this method, but it should give you a good idea of where your ancestors came from and some key local figures to whom you may be related. In my own case, this has proved my ancestry comes from within about 10 miles of the City of Aberdeen, where it has been since at least 1400 AD.

Once again, everyone's situation is different. One of the things that haplogroup projects, like the U106 group, are best at is individual recommendations for testing. However, it's likely that it will be based around this kind of procedure. If you are considering further testing at this point, see if these situations can be adapted to what you want to accomplish, and your haplogroup project should be able to you more individualised advice.

BIGY OR YELITE?

The choice of next-generation sequencing is not so clear cut in today's market. There is not a one-size-fits all solution to everyone's problem. What you do depends on your particular problem and how best it is solved.

Coverage: The Y chromosome is about 60 million base pairs long. BigY sequences 8-10 million of these (see later pages). YElite sequences about 14 million of these. Read quality is generally better for YElite than BigY: the number of SNPs found in a YElite test is 40-60% greater than in BigY. Neither test covers every important SNP, but both cover most of them. E.g., in U106, YElite covers Z301 and DF98 but BigY doesn't. Generally speaking we will know whether you are positive for most of these kind of SNPs, but some people will be exceptions.

Value for money: YElite is about 35% greater than BigY ($775 versus $575: both prices are subject to change and discount). Since YElite has greater coverage, it is better in terms of SNPs per dollar.

Analysis: BigY will give you a list of known SNPs, which include most of those on their haplotree and plenty more besides. They will also give you a list of "novel variants" – some of these are SNPs we've known about for some time, some will be new to your test, and some will be bad data (no test is perfect). Almost everyone will need some help in analysing the results from BigY beyond what FTDNA will give you. Your haplogroup project (e.g. the U106 group) can help.

Full Genomes Corp. gives you a set of raw results, with limited interpretation on top of that. These results are better quality controlled. However, they will still require a modest technical knowledge to understand. I would anticipate that most people will need help in analysing the results. Again, your haplogroup project can help.

In many ways, this is similar to receiving your STR results for the first time. You will need to interpret a series of otherwise meaningless numbers.

Analysis by a group: Different groups have different ways of dealing with data. In the U106 group, we are well set up to rapidly analyse the results of BigY tests and report back to individuals. We can tell you how and when you relate to other testers. We are starting to offer suggested origins in a few cases where these are becoming clear.

In U106, we are not yet at that stage with YElite. Individually, I am not yet able to provide ages for YElite tests, as the systems are sufficiently different that I need to characterise the test results and work out how the differences between the different WGS/YElite tests affect the age estimates. This is work in progress: we intend to have something working soon.

Incorporation of data: YElite tests obviously will not be incorporated into the Family Tree DNA database. You will have to keep track of more things yourself, e.g. how your haplogroup relates to those of the people around you, and making sure all your project administrators know you have YElite results. It's a small thing, but one to be considered. For U106, our Yahoo forum acts as a repository for this information, where we can process everyone's tests together.

Conversely, FGC will name your SNPs and officially "register" them, which FTDNA is not yet doing. If you have a BigY test, you can pay FGC/YFull $49 for this privilege. FGC will also put you into their tree and let you know which singletons you truly share with other people (the U106 group does this for free!). YFull will also give you an age (although for U106 they have fewer testers so it will be less accurate).

Overview of the Y chromosome

Compiled by: Dr. Iain McDonald; updated: 2 Dec 2015

Source material:
H. Skaletsky, et al., 2003, *Nature*, **423**, 825
P. Francalacci, et al., 2013, *Science*, **341**, 565
B. Trombetta, et al., 2014, *Mol. Biol. Evol.*, **31**, 2108
Selected loci from: ybrowse.org

THE Y CHROMOSOME

The Y chromosome is the shorter of the human sex specific chromosomes. The reference sequence we use in this document, Build 37, stretches it out to 59,373,566 base pairs. (The newer Build 38 is slightly shorter, as several gaps have had their sizes changed.)

When a genome is sequenced, the DNA is broken up into bite-sized pieces of tens or hundreds of base pairs long. These pieces contain genetic code in the form GATACTGA… They run like stretches of tape. The reference sequence is a bunch of these, stitched together where they overlap. However, there are gaps in this sequence, so we do not even know exactly how long the Y chromosome is, never mind what is in it.

In reconstructing a genome from a new test (e.g. BigY/YElite), fragments are compared to this reference sequence, and pasted in where they best fit. New SNPs are discovered by looking for differences from the reference sequence.

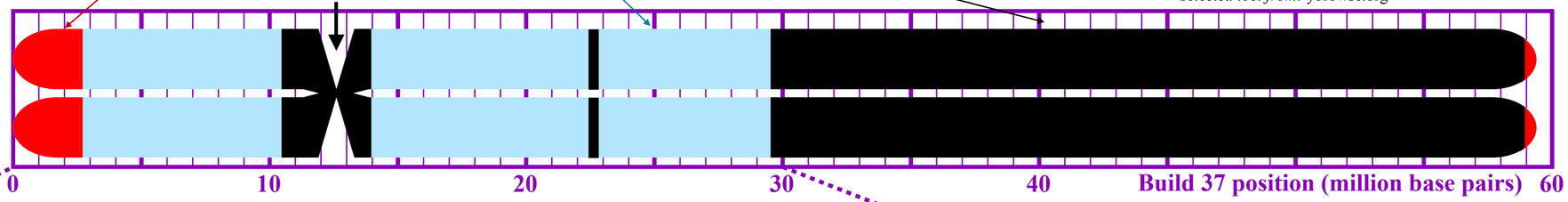
This process requires knowing: (a) which chromosome you are looking at and (b) which place on the chromosome you are looking at, so regions that look like other chromosomes and very repetitive regions usually cannot be read accurately. Only the *euchromatic* regions can.

PSEUDO-AUTOSOMAL REGIONS
These regions are very difficult to read, as their coding strongly overlaps with those of the other (autosomal) chromosomes. These regions are not inherited strictly via the male line, so are not of use to general Y chromosome studies.

CENTOMERE
The crossing point of the chromosome, where the two arms join together.

EUCHROMATIC REGIONS
These are the easily read regions of the Y chromosome where most SNPs and STRs are found.

HETEROCHROMATIC REGIONS
These are regions of very repetitive encoding. This makes these regions very difficult to sequence. 65000 bp of useful information can be found at the end of the long Yp12 heterochromatic region, before the pseudo-autosomal region PAR2. This is not shown on the plots below.



COVERAGE: The claimed coverage of 510 Family Tree DNA's BigY and 8 Full Genome Corp.'s YElite 1.0 tests are shown on the plots below. The companies differ in the quality needed to claim an SNP is called accurately. An intermediate trace is included, showing coverage of 7 BigY raw data (BAM files) as analysed at Full Genome Corp. by Vince Tilroe, using the same procedure as YElite. This allows the two tests to be compared to each other fairly.

There are a number of regions in BigY where a large fraction of the SNP calls are later found to be problematic, an called inconsistently with the structure we find in the rest of the tree. These are labelled. Ignoring these regions increases the fraction of repeatable SNPs from 76% to 87%. Similar regions are not shown for FGC as there are insufficient tests to make a consistency call in a manner that can be fairly compared against BigY.

TYPES OF EUCHROMATIC DNA:

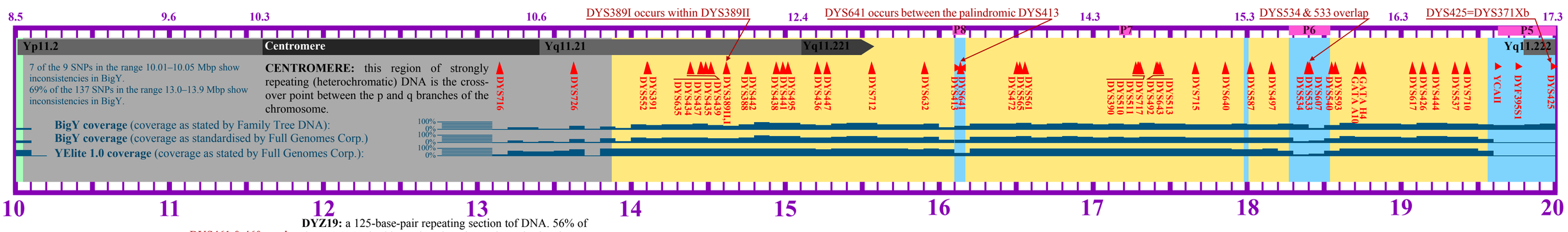
OTHER: Other regions not fitting the above categories.

X-DEGENERATE
Regions with origins predating the split between X and Y chromosomes (~166 million years ago). These regions are largely similar between X and Y, but the accumulated mutations mean they are generally separable.

X-TRANSPOSED
These regions are copies of regions of the X chromosome that are reproduced in the Y chromosome with ~99% repeatability. They are very difficult to sequence due to this similarity.

AMPLICYCLONIC
Regions of DNA that are largely unique to the Y chromosome. These are the easiest to sequence, but contain palindromic regions which are more difficult.

PALINDROMES
Regions of repeating DNA of the form (e.g.) AGCT…TCGA, many of which form palindromic loops, or extensions of the DNA out of the main strands. The multicopy markers lie on these arms, with one marker on each side of the palindrome.



NEXT-GENERATION TESTING

"Next-generation" tests like Family Tree DNA's BigY and Full Genome Company's Y-Prime and Y-Elite products offer an unparalleled chance to uncover new SNP mutations, insertions and deletions (indels) in your DNA. These are the only reliable tool we have for determining new structures within the Y-DNA tree (clades) and the only accurate way of obtaining dates we have. Of the 59 million base pairs in the human Y chromosome, these tests only cover between about 8 and 14 million.

CLADE IDENTIFICATION

Clade identification typically progresses as follows. When a new test arrives, we get two sets of summary data: the coverage of the test, and the differences that test has from a known sequence. These are reported as positions along the chromosome, e.g.:

```
chrY   2660548   2665410
```

means the test covers all base pairs between these two positions, and:

```
chrY 2661694   .   A   G   1484.13   PASS .   GT   1
```

means that a mutation from A to G has occurred at position 2661694. In this case, this SNP has been later given a name, L311, which typically replaces this number. Occasionally, SNPs may be rejected if they have a low quality score:

```
chrY 2649856   .   G   .   150.356   REJECTED   .   GT   0
```

SNPs from each test are compared to each other, e.g.:

```
7246726  7246726          7246726  7246726  7246726  7246726
23612197          23612197 23612197
                           19047132 19047132
6788390  6788390
                 22178569 22178569
13494176                  13494176
22191144
         7906217
                 22758149
                                  17735808
                                           23165645
                                                    19035709
                                                             14991735
```

Names of known SNPs are filled in and singletons are put together:

```
Z381   Z381         Z381   Z381   Z381    Z381
L48           L48   L48
                           Z307   Z307
Z9     Z9
                 L47   L47
Z2001            Z2001
SINGLETONS:
Z8     S6909  Z159         DF96   S1911   S5520  Z18
```

Comparisons to the coverage file let us fill in "no calls" (nc) and most false negatives (+?):

```
Z381   Z381   (+?)  Z381   Z381   Z381    Z381
L48    nc     L48   L48
                           Z307   Z307
Z9     Z9
                 L47   L47
Z2001            Z2001
SINGLETONS:
Z8     S6909  Z159         DF96   S1911   S5520  Z18
```

Inconsistent SNPs (Z2001) are treated as false positives and removed:

```
Z381   Z381   (+?)  Z381   Z381   Z381    Z381
L48    nc     L48   L48
                           Z307   Z307
Z9     Z9
                 L47   L47
SINGLETONS:
Z8     S6909  Z159         DF96   S1911   S5520  Z18
```

Providing a tree like that produced for U106 by Andrew Booth.

CHARACTERISATION OF FTDNA BIGY

We now have sufficient tests that we can perform a fairly rigorous characterisation of BigY. The analysis presented below is based on 510 BigY tests: the entire analysed sample as of 17 November 2015.

The average BigY test comprises of 10,613,667 callable base pairs (standard deviation 312,666) over 11,358 regions (st.dev. 2391). Typically 131 SNPs are called in each file including 14 novel variants (new SNPs private to this test).

A problematic region exists around position 22,400,000. Many SNPs are correctly called in this region, but there are a lot of falsely called SNPs too. This region of the Y chromosome is very similar to one on the X chromosome, and coverage of this region is very low. Many larger indels in this region are falsely reported as a series of SNPs. These often show up as singletons and confound later dating operations. For many applications, include dating of SNP ages, I have removed the entire DYZ19 region between positions 22216800 and 22512940 and do not use any SNPs found here. Typically 102,700 base pairs are called in this region. Other problematic regions exist, but they are less significant, and do not greatly affect the overall results presented here.

The typical overlap between two tests (excluding DYZ19) is 10,504,207 base pairs (st.dev. 308,548), or 97.5% overlap. For two given tests, 2.5% of SNPs are not be called in the matching test.

Boundaries of declared coverage are also a problem for BigY. From the first 319 BigY tests, of 6974 SNPs expected to be common to five or more people, 276 (4.0%) are not correctly called. Of these, 51 (0.73%) are "no calls", 220 (3.2%) are false negatives on the lower end of coverage boundaries, and 5 (0.07%) are false negatives in the main body of coverage. In total, 370 SNP calls are made on lower coverage boundaries, resulting in a 59.5% false negative rate.

A total of 540 SNPs were listed as having incorrect calls in BigY tests. Of these, 177 are "correctly" called SNPs shared by all testers but are listed as inconsistent as they have gaps for no calls and coverage boundaries, leaving 363 SNPs which are sporadically called and ignored (e.g. L128). A total of 3553 false positives are counted, at a rate of one per 1.216 million calls (0.000 082%).

A typical test has 33.13 (st.dev. 6.79) SNPs underneath U106 once all these factors are taken into account, or one per 307,162 base pairs. Including the DYZ19 region would give 36.33 SNPs, or one per 291,393 base pairs, or 5.2% more. An average of 8.71 SNPs per test are estimated to be called sporadically, leading to one SNP per 243,227 base pairs, or 26.3% more SNPs in the raw results as received from Family Tree DNA compared to the final cleaned results.

Of these, 164 SNPs are incoherently called twice, and could represent two instances of a new novel variant. With 33 SNPs per test, lottery mathematics expects one in every 9346 SNPs* to overlap. For 7212 unique SNPs, the probability of this happening once is 54%**. The likelihood is therefore that only one out of these 164 SNPs is actually two instances of a novel variant.

```
*=COMBIN(101762179;33)/COMBIN(33,1)/COMBIN(101762719-33;33-1)
**=1-(1-1/[*])^7212
```

CHARACTERISATION OF FGC Y-ELITE

Eight Full Genomes Corp. YElite 1.0 tests were analysed by Vince Tilroe during December 2015. YElite 2.0 tests were not available for comparison at this time, but reports from early batches indicate a similar number of callable base pairs for the YElite 1.0, 2.0 and 30x Full Genome tests.

The average YElite test comprises of 14,073,254 callable base pairs (standard deviation 178,471). This gives it considerably higher coverage than BigY (33% more, according to the respective companies' coverage — this is explored more later). It also gives it much higher repeatability between tests.

Rather than a simple yes/no flag, FGC assigns quality codes to each SNP: +, *, ** and *** in decreasing order of quality. Taking an average among the 27 existing YElite 1.0 tests registered with the U106 group, there are an average of 559 SNPs of all qualities called in each test. Of these, 170 are common to all U106 testers. Typically each test will have 29.9 shared SNPs below U106 (at all quality levels), and 34.0 singletons will be called (at the + and * quality levels), making 63.9 SNPs below the U106 level.

At face value, this gives 1.75x more SNPs under U106 than BigY. Note that this is obviously inconsistent with the 1.33x greater coverage. Some of the difference will be because not all the inconsistent SNPs have been found in YElite, as not enough tests have been taken at this stage. Some of the difference will be due to intrinsic differences in the rate of SNP formation in different regions of the Y chromosome. However, a more significant difference will be due to differences in what each company claims to be a callable base pair.

CHARACTERISATION OF BIGY USING THE FGC PIPELINE

Vince Tilroe also characterised the raw data (BAM files) of seven BigY tests using the Full Genomes Corp. data reduction pipeline. This provides a consistent basis on which to assign coverage between the YElite and BigY tests, allowing for direct comparison between the two. The results by Y-chromosome region are presented in the chart on the next page.

This analysis highlighted a much reduced number of callable base pairs using the FGC pipeline than the reported coverage by Family Tree DNA. In the following pages, I discuss the differences between the Family Tree DNA and FGC coverage statistics of the BigY and determine which is more appropriate for assigning the number of base pairs called in any particular test.

COMPARING BIGY COVERAGE

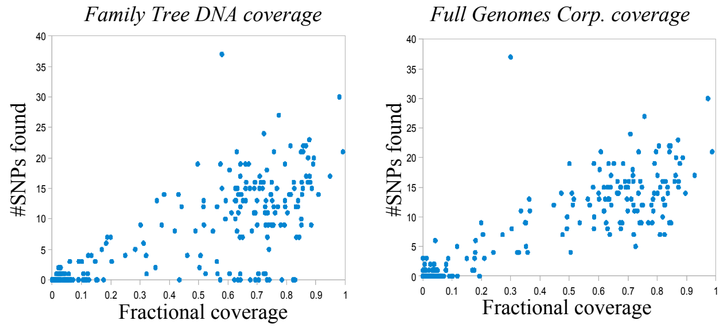


Figure: *Coverage of 100,000-base-pair regions from samples of BigY tests, as reported by BAM analysis from Family Tree DNA and Full Genomes Corp., along with the number of SNPs found in each region. For accurate coverage, a fairly close relation is expected, although natural variation is known to exist.*

As mentioned in the previous sections, notable differences exist between the coverage reported by Family Tree DNA of BigY tests, and that reported from analysis of the raw data by a third party, Full Genomes Corp. To investigate which coverage statistics are more accurate, we take each firm's coverage with the number of repeatable SNPs in that region. These repeatable SNPs are selected and defined by the following steps: (1) they are called by Family Tree DNA in the test results of our 516 BigY testers (the full sample at the time of this analysis); (2) they must be called "PASSED" in the Family Tree DNA variant call file (VCF); (3) they must be found in at least two closely related tests to demonstrate they are accurately called and (4) they must not be called inconsistently with the rest of the tree phylogeny, in order to ensure that false positives are removed.

The more accurate the coverage, the closer the relation we can expect between coverage and number of SNPs called. We take the square of the Pearson correlation co-efficient to determine the relationship between the two sets of data: for a perfectly correlated dataset, R^2=1, while for a perfectly uncorrelated dataset, R^2=0. Some natural variation exists within different regions of the Y chromosome (~20%; Helgason et al. 2015), but this is small compared to the scatter in the above figure. A significant scatter is expected due to the finite number of SNPs called in each region, through the mechanism of small-number (Poisson) statistics. Overall, this combination of factors accounts for the scatter in the FGC analysis, which has R^2=0.411, compared to R^2=0.320 for Family Tree DNA's analysis.

In the Family Tree DNA analysis, there are a number of regions of reported high coverage (60-90%) with very few SNPs (0 or 1). This cannot be accounted for by random processes or natural variations in the data, and must reflect an inaccurately called coverage. The accuracy of coverage becomes important later when determining the ages of clades. In order to accurately determine the number of base pairs accurately called in a BigY test, we must determine in which regions the coverage claimed by Family Tree DNA is accurate.

EXCLUDING REGIONS FROM BIGY

The regions where Family Tree DNA and Full Genomes Corp. have the greatest discrepancy in their coverage statistics are in the heterochromatic regions (centromere region and DYZ19), the palindromic arms and the inverted repeats. A large fraction of SNPs in these regions do not match our consistency criterion applied earlier (i.e. they are not found in people of the same clade, but are found in people of different clades). This is very strong evidence that many or all instances of these SNPs are false positive calls. DYZ19 is a particularly problematic region, as not only are a large fraction of SNPs called in this region problematic, but it is also a large region with good claimed coverage.

Discussion with several experts on this topic (including Justin Loe, Vince Tilroe, Greg Magoon and James Kane) suggests the problem may lie in particular details of the algorithm used to assign whether a call can be made. While the details of how Family Tree DNA defines a callable base pair are unknown at this time, FGC assigns quality based on the GATK (Harvard – MIT Broad Institute) criterion, which takes into account the number of reads of that base pair, and both the quality of each read and the quality with the fragment it is on is matched.

The suggestion is that Family Tree DNA's algorithm allows a lower mapping quality criterion. This means that regions which closely duplicate other regions of the chromosome (e.g. palindromic and inverted repeats) or are strongly repetitive (e.g. heterochromatic regions) are marked "callable", even though there is increased ambiguity over whether each segment is correctly aligned.

For this reason, analysis of BigY VCF/BED files which depend on coverage should restrict themselves to euchromatic regions that are not part of palindromic arms. It is also advisable in many cases to remove regions of very low coverage, where many SNPs will be marked as singletons that are simply not called in other tests. The GrCh37 positions for the remaining regions are given below.

ACCEPTED BIGY REGIONS

The following table gives the start and end co-ordinates of the regions of BigY which should approximate accurate coverage in both the FGC and FTDNA analyses (repeats less than about 10000 bp are ignored).

START	END	INCLUDED?	DESCRIPTION
0	2649599	No	Non-euchromatic (PAR1)
2649600	**2917999**	**Yes**	**X-degenerate, non-palindromic**
2918000	6102799	No	X-transposed, very low coverage
6102800	6400699	No	Ampliconic, but inverted repeat (IR3)
6400700	6616499	No	X-transposed, very low coverage
6616500	**7446499**	**Yes**	**X-degenerate, non-palindromic**
7446500	7507249	No	Ampliconic, but inverted repeat (IR1)
7507250	8864102	Yes	Ampliconic, non-palindromic
8864103	9166089	No	Ampliconic, but inverted repeat (IR4)
9166090	**9466025**	**Yes**	**Ampliconic, non-palindromic**
9466026	9757201	No	Ampliconic, but inverted repeat (IR3)
9757202	**9968999**	**Yes**	**Ampliconic, non-palindromic**
9969000	**10034899**	**Yes***	**Non-euchromatic, mostly but correctly read**
10034900	13870499	No	Non-euchromatic: heterochromatic (centromere)
13870500	**16095999**	**Yes**	**X-degenerate, non-palindromic**
16096064	16167426	No	Ampliconic, but palindrome (P8)
16167427	**17986699**	**Yes**	**X-degenerate, non-palindromic**
17986700	**18016999**	**Yes***	**Ampliconic - unclear relation to P7**
18017000	**18217274**	**Yes**	**X-degenerate, non-palindromic**
18217275	18537844	No	Ampliconic, but palindrome (P6)
18537845	**19622082**	**Yes**	**X-degenerate, non-palindromic**
19622083	20557687	No	Ampliconic, but palindrome (P5)
20557688	20648536	No*	Ampliconic, non-palindromic, but poor and differing coverage
20648537	20995635	No	Ampliconic, but palindrome (P4)
20995636	**22216399**	**Yes**	**X-degenerate, non-palindromic**
22216400	22512899	No	Non-euchromatic, heterochromatic (DYZ19)
22512900	**23462399**	**Yes**	**X-degenerate, non-palindromic**
23462400	**23693155**	**Yes**	**Ampliconic, non-palindromic**
23693155	23996387	No	Ampliconic, but inverted repeat (IR4)
23996338	**24050183**	**Yes**	**Ampliconic, non-palindromic**
24050184	24872777	No	Ampliconic, but palindrome (P3)
24872777	25207472	No*	Ampliconic, non-palindromic, but poor and differing coverage
25207473	25501597	No	Ampliconic, but palindrome (P2)
25501598	25620314	No*	Ampliconic, non-palindromic, but poor and differing coverage
25620315	28475315	No	Ampliconic, but palindrome (P1)
28457316	28783999	No	Non-euchromatic, poor repeatability
28784000	59373566	No	Non-euchromatic, heterochromatic

A description of the inclusive regions in a C-based Boolean format is:

```
(x >= 2649600 && x <= 2917999) || (x >= 6616500 && x <= 7446499) || (x >= 9166090 && x <= 9466025) || (x >= 9757202 && x <= 10034899)  || (x >= 13870500 && x <= 16095999) || (x >= 16167427 && x <= 18217274) || (x >= 18537845 && x <= 19622082) || (x >= 20995636 && x <= 22216399) || (x >= 22512900 && x <= 23693155) || (x >= 23996338 && x <= 24050183)
```

or for use in spreadsheets (for a fictitious range A1:A999, check whether your system uses ";" or "," as a field separator):

```
=(COUNTIF(A1:A999;">=2649600")-COUNTIF(A1:A999;">2917999"))
+(COUNTIF(A1:A999;">=6616500")-COUNTIF(A1:A999;">7446499"))
+(COUNTIF(A1:A999;">=9166090")-COUNTIF(A1:A999;">9466025"))
+(COUNTIF(A1:A999;">=9757202")-COUNTIF(A1:A999;">10034899"))
+(COUNTIF(A1:A999;">=13870500")-COUNTIF(A1:A999;">16095999"))
+(COUNTIF(A1:A999;">=16167427")-COUNTIF(A1:A999;">18217274"))
+(COUNTIF(A1:A999;">=18537845")-COUNTIF(A1:A999;">19622082"))
+(COUNTIF(A1:A999;">=20995636")-COUNTIF(A1:A999;">22216399"))
+(COUNTIF(A1:A999;">=22512900")-COUNTIF(A1:A999;">23693155"))
+(COUNTIF(A1:A999;">=23996338")-COUNTIF(A1:A999;">24050183"))
```

The history of U106

(1) INTRODUCTION

This deep phylogenic tree of the human population represents our current understanding of the way the human family tree has divided along its male lines. This is a rapidly-evolving field, thus the information is subject to considerable change over time.

This tree summarises the extensive tree that lies above U106. This shows how U106, which now represents many tens of millions of men worldwide, branched off from the rest of the human Y-chromosome tree at different points in prehistory. A map of this tree is shown on the next page.

(2) OUT OF AFRICA

Ultimately, we all descend from the first life-forms, which lived approximately three billion years ago. Through a long and convoluted process, they evolved into *homo sapiens*. While *H. sapiens* has only been around for about half a million years, this is still older than the common ancestor of the male lines of every person alive today. We call this person Y-chromosomal Adam, because we all descend from him via our father's father's father's father's… etc. Recent estimates of his age vary widely from 120,000 to 380,000 years ago.

The vast majority of people descend through Haplogroup A. In fact, it's only recently that researchers discovered our most-distant relations hiding among remote Africa tribes. Haplogroup BT arose in Africa about 70,000 years ago, when the most of the human population consisted of a small number of tribes living in the Horn of Africa.

The human genetic tree continued to diversify and flourish as mankind expanded throughout Africa. Around 50,000 to 60,000 years ago, a small group of migrants is thought to have crossed the Red Sea into Arabia, starting the most important in a series of Out of Africa migrations.

Some time not too long after this point, a little over 45,000 years ago, we split from haplogroups G and I, which appear to form the original modern human population in Europe. This point is defined by the recently analysed 45,000-year-old remains from western Siberia (Ust-Ishim), from a man who was haplogroup K (but not haplogroup LT).

Our base haplogroup, R, arose from this migration between 24,000 and 34,000 years ago. This is again limited by the archeaological remains of Mal'ta Boy, who was buried 24,000 years ago in Siberia. By this time, our ancestors had probably expanded to across much of north-west Asia, where they existed as hunter gatherers.

(3) EXPANSION INTO EUROPE

Within haplogroup R, most people are part of R1, descended from an individual living 24,000 to 34,000 years ago. The majority of western Europe is descended from the R1 founder. Within R1, there is a bifurcation into two groups: R1a, or M420, and R1b, or M343. R1a is strongest in eastern populations, where it can exceed 60% of individuals in Poland and the south-west Russian states. Its British content is thought to be strongly Viking in origin.

R1b (M343) is thought to have arisen less than 18,500 years ago. In Europe, it is very much dominated by R1b1a2, or M269. This group alone makes up over half the population in Western Europe, and makes up over 90% of some populations. Despite this, its origins are still thought to have been in western Asian populations.

The date of this expansion into Europe can probably be tied to the sudden growth in the number of branches below M269, which can be very roughly dated to around 4000 BC. The origin of this migration and its route into Europe are not well determined at present. However, archeaological remains show that there was extremely few haplogroup R men in Europe before 2600 BC, when remains from both R1a and R1b are found in Corded Ware and Bell Beaker burials (respectively) in south-eastern Germany.

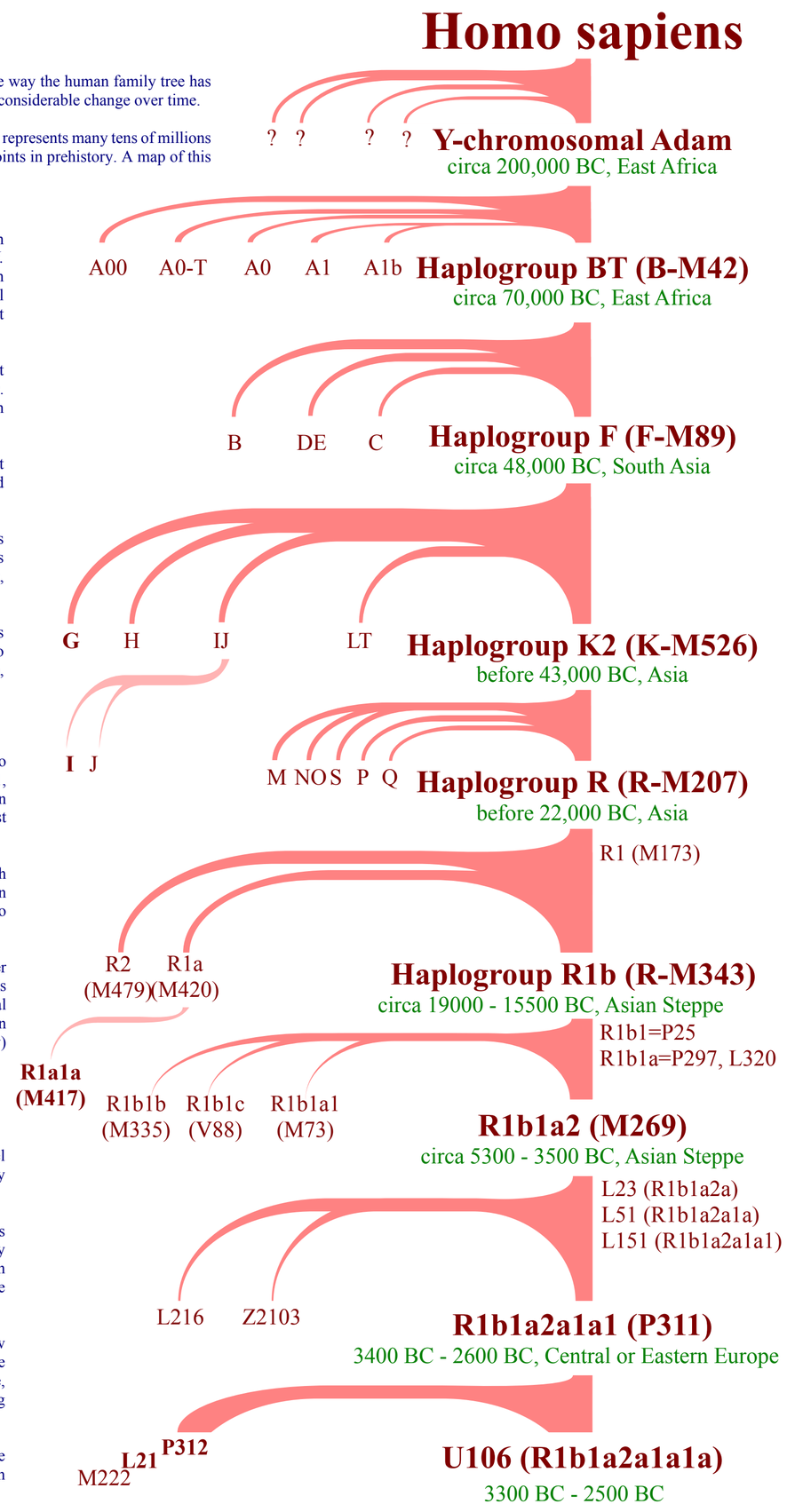
(4) FOUNDING A NEW EUROPEAN POPULATION

Most of the branches above U106 are minor, however there is one important branch at the level immediately above U106, signified by the mutation P311. A split exists at this point in our family tree between the larger P312 branch and the smaller U106 branch.

The P312 branch is generally found more on Europe's Atlantic Coast, while the U106 branch is generally found more in Europe's heartland. This has led to P312 being referred to synonymously with "Celtic" peoples while U106 is "Germanic". While there is clearly some overlap between membership of these SNPs and populations, both SNPs originate several thousand years before these terms are relevant.

Nevertheless, it is the last common ancestor of these two branches, "Mr. P311" whose clan is now represented by around half of western European men, with a third of a billion diaspora worldwide (see panel at right). The date of this man's birth is likely to be during the European Bronze Age, and the possible range of dates correspond to a series of archeaological horizons spreading eastwards over Europe at the same time.

Within P311, U106 represents about 1/8th of Europe, or 110 million men worldwide. We estimate its age to be between 2500 and 4600 years old. We trace what is known about the migrations from Asia to Europe on the next page.



Deep ancestry of U106

Acknowledgements

The information in this tree comes from a variety of sources, but I am most grateful to the International Society of Genetic Genealogy (ISOGG) for maintaining the underlying tree structure displayed here. The anthology of haplogroup statistics on eupedia.com has also been instrumental in creating these data.

Created by: Dr. Iain McDonald; updated: 17 Nov 2014

How to read this chart

This chart shows how the male-line genetic (phylogenic) tree splits from its foundation down to the U106 branch. Different ages and geographical origins distances are shown on the chart, which should be interpreted carefully.

Where quoted, ages are given as 95.5% confidence intervals, what we call "2-sigma". We are 95.5% sure that the real dates lie between these two boundaries. By dividing the uncertaintty in half, we can recover the 68% confidence interval, or "1-sigma" range. Dates are rounded to the nearest 50 years. For example, we are 95.5% sure that the U106 founder lived between 3260 BC and 1974 BC. We are 68% sure that he lived between 2938 and 2295 BC.

This date was calculated using SNP-counting methods which are detailed on later pages.

Haplogroup Frequencies in Europe

The following data give the number and percentage of various levels between R1b-M343 and U106 in different parts of Europe, as found by Myers et al. (2007) and selected other studies. These can be used to approximate correction factors to debias our statistics according to how many men of different ancestries have tested. These numbers are only very approximate in many cases and only represent first-order estimates of the underlying population.

COUNTRY	POPLN.	%M269	%U106	M269 & U106 POPLN.		#TESTERS	WEIGHT
British Isles							
Ireland	6429508	80%	6%	2571803	192885	99	4
Scotland	5327000	73%	12%	1931037	319620	132	5
England	53012456	57%	20%	15108549	5301245	317	33
Wales	3063456	84%	5%	1278992	76586	13	12
Total	**67836420**	**62%**	**19%**	**21029290**	**6444459**	**658**	**18**
Iberia							
Spain	47150800	42%	5%	9901668	1178770	6	629*
Portugal	10607995	56%	5%	2970238	275807	3	53*
Total	**57758795**	**45%**	**5%**	**12995728**	**1443969**	**9**	**323***
Scandinavia							
Norway	4930116	25%	15%	616264	369758	31	24
Sweden	9360113	15%	10%	702008	468005	29	32
Finland	5357537	2%	1%	53575	26787	8	7*
Total	**19647766**	**14%**	**9%**	**1375343**	**884149**	**68**	**25**
Central Europe							
Denmark	5568854	34%	17%	946705	473352	9	105*
Netherlands	16696700	54%	35%	4508109	2921922	32	183
Belgium	11198638	60%	25%	3331594	1399829	10	280
France	65460000	52%	7%	17019600	2291100	21	218
Germany	81757600	43%	19%	17577884	7766972	103	151
Switzerland	7785000	58%	13%	2257650	506025	13	78
Italy	60418711	37%	4%	11177461	1208374	14	173
Austria	8414638	27%	23%	1135976	967683	2	968*
Total	**257300141**	**45%**	**14%**	**57892531**	**18011009**	**204**	**172**
Eastern Europe							
Hungary	9979000	20%	4%	997900	199580	6	67*
Czech Rep.	10261320	28%	14%	1436584	718292	5	287*
Slovakia	5443386	25%	3%	680423	81650 1	327*	
Poland	38192000	23%	8%	4392080	1527680	19	161
Lat./Lit./Est.	6032500	10%	4%	301625	120650	12	20
Belarus	9503807	5%	1%	237595	23759 1	48*	
Ukraine	45939820	25%	9%	5742477	2067291	4	1034*
Romania	20121641	15%	2%	1509123	201216	1	402*
Bulgaria	7621337	10%	2%	381066	76213 0	-*	
Former Yugo.	20449929	5%	1%	511248	102249	1	204*
Slovenia	2012917	17%	4%	171097	40258 3	27*	
Greece	11645343	10%	1%	582267	58226 0	-*	
Russia	110000000	21%	5%	11550000	2970000	7	849*
Turkey	76667864	14%	0%	5366750	153335	0	-*
Total	**373870864**	**18%**	**5%**	**33648377**	**8412094**	**60**	**281**
European Colonies (estimated)							
United States	230000000	46%	15%	52900000	17250000	-	-
Australia	20000000	46%	15%	4600000	1500000	-	-
NZ	4000000	46%	15%	920000	300000	-	-
Canada	30000000	46%	15%	6900000	2250000	-	-
Total	**1041 million**	**N/A**	**N/A**	**193 million**	**56 million**	***(* Bias factor highly uncertain)***	

Origins of U106 clades: *Homo sapiens* to R1b-M269

Updated: 11 December 2015
Dr. Iain McDonald
on behalf of the U106/S21 group

(9) R1b
R1a and R1b now define substantial fractions of the populations west of the Ural mountains. The "original" R1b SNP, M343, probably arose less than 20000 years ago, still in the Russian steppe. Although they didn't begin to migrate westwards until much later, the destinies of both R1a and R1b in Europe were still intertwined. It is likely that these two populations were both contained by the glacial snowline, which they ultimately followed westwards.

Yamnaya culture

(10) M269
The story of R1b beyond the Urals is poorly known. What is clear is that there was a gradual movement westwards, probably to somewhere in the Dnieter-Don valley system, where it probably arrived in the post-glacial Holocene era. M269 defines the next major split in our ancestral population. It occurred around 6000 years ago, and has been linked to the Yamnaya culture and the expansion of Indo-European languages and culture to Europe around 5000 years ago (the Kurgan hypothesis). An alternative hypothesis, by which M269 originated in the Caucasus and spread via Anatolia with the first European farmers, appears discredited based on recent age estimation and ancient DNA testing. The details of the movements between M269 and U106 are explored later in this document.

(4) Fertile Crescent
Before long, our ancestors had reached the Fertile Crescent. At this point it extended down into the Arabian Gulf (which was an isolated wetland until around 8000 BC). Haplogroup F was probably born somewhere in this region. It marks the split of haplogroups G, I and J, which went on to become the "native" *Homo sapiens* population in Europe.

(3) Out of Africa
Descendants of "Adam" spread throughout Africa. While the Neantherthals and Denisovans had spread out of Africa long before, it took our ancestors until around 47,000 to 55,000 years ago to make the move. Debate exists as to whether they left via a southern route, crossing the Straits of Aden, or a northern route via the Sinai peninsula (see, e.g., Pagani et al. 2015).

(2) Y-chromosomal Adam
In this study, we only trace male lineages. These converge in a more recent ancestor, who we call "Adam". Our most distant relations are a group of ancient African tribes, notably including some from the Kalahari, who share the most distant haplogroup, A00. Recent estimates of when "Adam" lived vary, but are typically 170,000-320,000 years ago.

(1) The Dawn of Man
The origin of man can be traced to the Horn of Africa, where *Homo sapiens*, *Homo neanderthalis* and *Homo denisovans* were last related by their common ancestor, *Homo heidelbergensis*. This relation occurred around 300,000 to 400,000 years ago. Interbreeding among these three groups means that we all share a little of that Neantherthalic and Denisovan DNA, so one interpretation is that dawn of man occurred at this point (see Karmin et al. 2015 for date estimates).

Ust'-Ishim

(7) Mal'ta & haplogroup R
The following few millenia are even more difficult to piece together. It appears that our ancestors survived in the Siberian tundra for many tens of thousands of years. An important split, between the Q and R haplogroups, occurred around 25000 years ago. Haplogroup Q went east to east Asia and Siberia. Haplogroup R is the lineage of our ancestors. The exact date isn't known, but an early haplogroup R burial has recently been sequenced in the region of Mal'ta, just west of Lake Baikal, which dates to 24000 years ago. This burial is probably less than 1000 years before the last common ancestor of all haplogroup R today.

Mal'ta

(8) R1
Haplogroup R1 is a few thousand years younger, yet our ancestors probably still lived in the Ice-Age Siberian tundra. R1 and R2, the two major branches of haplogroup R, separated at this time. Our branch, R1, went west, while R2 went south towards the Indian sub-continent.

(6) Ust'-Ishim
The origin of haplogroup K is debatable. Several scholars place it along a continuing progression across modern Iran, skirting the Tian Shan mountains towards Lake Baikal, though origins from the Caucasus to south-east Asia have been discussed. Recent hypothetical reconstructions have put the origin of haplogroup K as far south and east as the Indian sub-continent. However, DNA recovered from a skeleton in western Siberia has been shown to be haplogroup K, and has been dated to between 44000 and 46000 years ago, close to the time when haplogroup K is supposed to have been established. It may be that our lineage took a more northerly route. (See Fu et al. 2014 for the Ust'-Ishim burial.)

(5) North and East
Haplogroup K represents a large section of the descendents of the people that left Africa. It includes many Amerindian, Australasian, far Eastern and polar populations.

WARNING!
THE DETAILS OF THIS MAP CANNOT CURRENTLY BE PROVED WITH ANY SCIENTIFIC RIGOUR. THE EXACT PATH IS NOT MEANT TO BE A TRUE REPRESENTATION OF HISTORICAL MIGRATIONS: IT ADOPTS THE MINIMUM PATH LENGTH THAT ADEQUATELY DESCRIBES THE CURRENT DATA. DETAILS OF SOME ALTERNATIVE HYPOTHESES ARE GIVEN IN THE DESCRIPTIVE TEXT, AND NUMEROUS OTHER, MORE AUTHORITATIVE CHARTS ARE AVAILABLE ELSEWHERE.

Age estimation

AGE ESTIMATION FROM NEXT-GEN TESTS (BIG Y, ETC.)

The formation of SNPs is a largely random process. Many processes affect genetic integrity and structure, many carcinogens cause genetic mutations (harmful or otherwise), and many social and environmental factors affect the number of mutations passed from father to son. However, these largely cancel out when one considers a large population over a long time. Certainly, SNP creation seems like a random process within the errors of our observations.

SNP mutations can therefore act as a clock, albeit one that does not have a regular tick. SNP creation is a roll of the dice: sometimes you will get one, sometimes you won't. Sometimes it will be in your tested region, sometimes it won't. Over long timescales, and many lineages, these effects cancel out, so that there is a particular rate at which SNPs form. We can therefore expect that SNPs will build up in "next-generation" tests like BigY or YElite at the rate of a certain number of years per SNP, which we will call r.

At its simplest, the age of a clade (t) can be estimated by the taking the number of SNP mutations that are not shared by all the members of that clade (m), multiplying it by the timescale for SNP formation (r) and dividing it by the number of testers (n), thus:

$$t = m\,r\,/\,n$$

where m and n come from the BigY tests, and r comes from some nominal, independent measurement. For large clades, the rate r is the most uncertain parameter in this calculation: n is known precisely, and m is typically determined to much better than 3%. For small clades, the fact that SNP creation is a random process becomes important, and the small-number statistics of m are the dominant uncertainty.

ACCOUNTING FOR SMALL-NUMBER STATISTICS

Small-number statistics of SNP creation is governed by a branch of mathematics called Poisson statistics. Poisson statistics tells us the probability of observing any given number of mutations in a single lineage, compared to what a regular mutation rate "clock" would give. We reverse-engineer this calculation to find the uncertainty in the number of mutations we see (δm).

For large clades, calculating this uncertainty becomes technically impractical, so we use the Gaussian approximation that the 1-σ (68.3%) uncertainty in m is the square root of m, and that the 1.96-σ (95%) uncertainty is $1.96 \times \sqrt{m}$.

An additional uncertainty comes from the conversion of SNPs to years. This is because the mutation rate comes with its own uncertainty (δr). Since these uncertainties are uncorrelated, they are added in quadrature, such that:

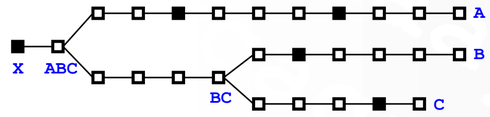
$$\delta t/t = \sqrt{([\delta m/m]^2+[\delta r/r]^2)}$$

This gives the age and its uncertainty listed in the final age products shown in this work, and is the final way in which the age of a mutation like U106 can be worked out.

TMRCA vs. BRANCH AGE vs. SNP AGE

What this calculation gives you is the time between the birth of the most-recent common ancestor and the average birth date of the n testers which have been tested. This is the "time to most-recent common ancestor" or **TMRCA**. This is subtly different from the **SNP age**: the actual age of the quoted SNP. In most cases, this distinction doesn't matter, but it can become important in some clades.

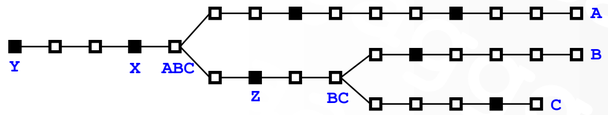
In the simplest case, we might have the following family tree, where every box represents the birth of a son and filled boxes represent the creation of a new SNP:



In this case, A, B and C share a most-recent common ancestor (ABC) and a terminal common SNP (X). The age of X is slightly older than that their TMRCA, but this can usually be ignored.

However, if only B & C take a next-generation test, and their common ancestor (BC) has not had any further mutations since the ABC ancestor, the TMRCA for B & C might be a century or two younger than either ABC or X.

This can become more serious if we have the following scenario:



Here, B & C share a set of SNPs (X,Y and Z). If only B & C test, we would get the following test results:

```
B: X+ Y+ Z+, 1 novel variant
C: X+ Y+ Z+, 1 novel variant
```

we have no idea which one out of X, Y or Z comes first. These lists of SNPs can become very long (30 or more SNPs), so they are often abbreviated by one of the SNPs, in this case "X". So what we write is the age of X (because we do not know any better), but what we calculate is the time since the birth of BC.

If tester A then comes along with the results:

```
A: X+ Y+ Z-, 2 novel variants
```

then we will know that X and Y come before Z. The recorded age of X will change, as the common ancestor ABC is much older than BC. It is therefore important to bear in mind the fact that what we are reporting is the time since the birth of the most-recent common ancestor of all people with the indicated SNP ***who have taken that next-generation test***.

Sometimes additional data is available (e.g. from a different next-generation test or individual SNP testing at YSeq or Family Tree DNA) that can split long chains of SNPs in this fashion. This data is not included when calculating the ages above, as it is not homogeneously reduced.

At the present time (Jan 2016), we are only calculating dates from BigY tests, as we have both a sufficient number of these, and a good enough idea of their calibration to do so. The calibration exists to do the same analysis with the FGC YElite tests, but this remains a future exercise.

A MORE ACCURATE AGE

Particularly in the case of small clade branching off from a much larger one (e.g. S5520 under Z156 or FGC396 under U106), a more accurate age can be derived by considering the time between the parent SNP and the target SNP.

This can be done in a similar manner, considering the number of SNPs between the parent and target SNP (m_p). This provides a more accurate answer when m/n is much larger than m_p. Excluding the DYZ19 region, for FGC396's two testers Lindemann and Kuykendall, $m_p = 7$ while $m/n = 17$. In practice, we can do this both from the U106 age and from the age of the immediate parent SNP, as sometimes one is more accurate than the other.

A final modification we can make is based on this method. If we fix the age of U106 using our original method, then we can adapt the ages for the fact that some lines (e.g. L48 averages 36.41) have more mutations than average, while some (e.g. Z18 averages 27.04) have fewer. This difference is expected, as larger clades will preferentially have more SNPs due to random sampling. This is exemplified in the two trees presented earlier, where the first tree produces three small clades, but the addition of SNP "Z" produces two clades, of which clade Z is larger. This is particularly effective during population expansion periods.

In this final method, we have a fixed age of U106 (let's say it's 4500 years). If we have a clade under U106 with an average of 45 SNPs, we can fix a mutation rate for this lineage of one SNP per 100 years. If it has an average of 22.5 SNPs, it will be one per 200 years. Naturally, our uncertainty measurement has to take this new mutation rate and its uncertainties into account.

Using these methods, we have a suspension-bridge-like design, whereby the origin of the tree, U106, is fixed from the present day. Clades are pinned to this tree both downwards from U106 via their parent lines, and up from the present day. The intersection of these two methods provides much more stable and self-consistent ages for each SNP than would be arrived at otherwise.

AGES OF INDIVIDUAL SNPS

Ages of the actual SNPs are more uncertain, given the processes described above. However, they will occur at a fixed time before the TMRCA or convergence age. This is given by:

$$t_s - t = r\,(n_r - 0.5) / 2$$

where n_r is the number of SNPs in an unbroken run (e.g. Z305, Z306, Z307, S1667 would give $n_r = 4$).

The 95% uncertainty on this is again computed from Poisson statistics, but asymptotes to +/- 0.475 $r\,n_r$ for large n_r.

FINAL AGE CALCULATION

The final age is determined from three numbers:
Firstly, from the number of SNPs beneath the target:

$$t = m\ r\ /\ n \qquad \textit{[T1]}$$

Secondly, from the number of SNPs between U106 and the target:

$$t_0 = t(\text{U106}) - m_0\ r_0 \qquad \textit{[T2]}$$

where t(U106) is the age of U106 from ***[T1]*** and m_0 is the number of mutations since U106. Here, r_0 is defined from the average number of mutations in that branch since U106 (m(U106)) as follows:

$$r_0 = m(\text{U106})\ /\ t(\text{U106}) \qquad \textit{[R2]}$$

Thirdly, from the number of SNPs since the parent clade:

$$t_p = t_0(p) - m_p\ r_0 \qquad \textit{[T3]}$$

where $t_0(p)$ is the age of the parent from ***[T2]*** and m_p is the number of mutations between the parent and the target SNP. ***[T1]*** can be adapted for a given clade such that:

$$t = m\ r_0\ /\ n \qquad \textit{[T4]}$$

which then gives the equality:

$$t = t_0 = t_p \qquad \textit{[T5]}$$

such that ages from the three estimates are consistent. A final modification to this age is made in the rare case that a sub-clade has a larger average number of SNPs beneath it than its parent ($m/n > m_p/n_p$). In this case, a hard limit are placed of at least 30 years after the parent clade's origin. A hard limit is also placed at 1950, representing an age of zero (see "Defining the Present Day", below).

FINAL AGE UNCERTAINTY

The uncertainty in the final age estimation is a combination of the uncertainties derived from equations ***[T2]***, ***[T3]*** and ***[T4]***. It therefore relies on the uncertainty in the U106 age. For a 95% confidence interval, this is the 1.96-σ uncertainty value, namely:

$$\delta t/t = 1.96\ \sqrt{([\delta m/m]^2+[\delta r/r]^2)} \qquad \textit{[ET1]}$$

where δr is derived from the literature or case studies. Similiarly, the uncertainty in ***[T4]*** can be derived as:

$$\delta t/t = 1.96\ \sqrt{([\delta m/m]^2+[\delta r_0/r_0]^2)} \qquad \textit{[ET2]}$$

where δr_0 is given from ***[R2]*** by:

$$\delta r_0 = 1.96\ [m(\text{U106}) -/+ \delta m(\text{U106})]\ /\ [t(\text{U106}) +/- \delta t(\text{U106})] \qquad \textit{[ER1]}$$

In both cases, $m-\delta m$ and $m+\delta m$ are given by the highest and lowest value of λ, respectively, for which:

$$\int_{k=0}^{k=m} \text{Pois}(\lambda,k) < 0.1585 \qquad \textit{[ER2]}$$

$$\int_{k=0}^{k=m} \text{Pois}(\lambda,k) > 0.8415 \qquad \textit{[ER3]}$$

at 1σ and:

$$\int_{k=0}^{k=m} \text{Pois}(\lambda,k) < 0.025 \qquad \textit{[ER4]}$$

$$\int_{k=0}^{k=m} \text{Pois}(\lambda,k) > 0.975 \qquad \textit{[ER5]}$$

at 95% confidence, where Pois() is the Poisson function, $(\lambda^k/k!)e^{-\lambda}$. For large m, where this value is computationally expensive to determine, the approximation $\delta m = 1.96\ \sqrt{m}$ is used for the 95% confidence interval.

The uncertainty in the other two age measurements follows similar principles, except that the uncertainty in m_0 and m_p replaces the uncertainty in m, and age is calculated in time since ($t\pm\delta t$) for U106 and the parent SNP, respectively, rather than from the present day.

If $\delta t_0 < \delta t_p$ (i.e. the age from U106 is more accurately determined than the age from the parent clade), then the U106-based age is used, otherwise the age is based on the parent SNP. This provides an age propagated forward in time, which we will call t_e (uncertainty δt_e). Note that as ***[ER4]*** and ***[ER5]*** provide asymmetric errors around m, the final uncertainty, δt_e, will be asymmetric around t as well.

Age uncertainties can be combined using a weighted average to produce a final uncertainty in the convergence age as follows:

$$\delta t_{\text{final}} = \frac{\left(\frac{t\pm\delta t}{w} + \frac{t_e\pm\delta t_e}{w_e}\right)}{\left(\frac{1}{w} + \frac{1}{w_e}\right)} \qquad \textit{[ET3]}$$

where the weights are set as follows:

$$w = (t/n\ .\ 2\delta t)^2 \qquad \textit{[ET4]}$$

$$w_e = ([t^* - t_e\ ].\ 2\delta t_e)^2 \qquad \textit{[ET5]}$$

where t^* is either t_p or t(U106), depending on which gives the more accurate age. The same limits are applied such that the cluster cannot be older than its parent and cannot be younger than the present day.

DEFINING THE PRESENT DAY

In this work, we use 1950 as being the present day, representing the average birth date in the testing population. This comes from an online survey of 98 DNA testers from the U106 group itself. The average birth year of these testers is 1950.3 with a standard deviation of 15.5 (i.e. a 1.96-σ uncertainty of 30.4 years for a single tester or 1.50 years for the total BigY testing population).

This estimate is likely to be slightly biased by those individuals who are active on the online forum compared to the underlying dataset, but overall this is expected to impart a relatively small uncertainty to the age of any particular SNP.

CHOOSING A MUTATION RATE

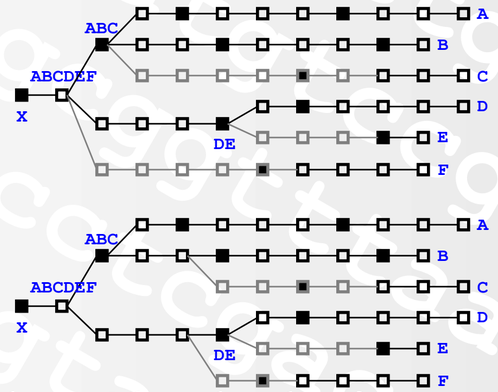
We have so far ignored how the choice of the underlying mutation rate, r, and its uncertainty, δr, are calculated. Ultimately, these come from three sources: (1) counting SNPs from known lineages among the BigY tests themselves, (2) literature studies which perform the same task of summing up a measured number of mutations which have occurred over a known period of time, (3) limits from DNA testing of archaeological remains.
In the next section, a selection of these are applied to the BigY tests. Full notes on their methods and homogenisation are detailed in the supplementary information in the associated file (snp-mutation-rate.xls) on deposit in the U106 forum or available on request.

(1) RATES FROM LINEAGES IN BIGY

As of Jan 2016, we have 103 BigY tests from lineages where we have a named individual who is very likely the common ancestor of at least two tests, where we also have positions of the mutations accumulated since that common ancestor. "Very likely" in this case is a judgement call made based on paper-trail and genetic evidence. In total, they represent around 46,000 years of lineages.

These are dominated by two Scottish families: the Clan Donald and the House of Stewart. In most cases with these lineages, we do not have the complete paper trail leading from the testing individual back to the common ancestor, but we have other BigY tests which show that they must come from this lineage.

These cases suffer from problems in accounting for the number of years in a lineage. In the following figure, we consider six testers (A through F) of which we have full paper trails from A, B and D. Paper trails from C, E and F are only partially known (shown in gray). We show two possible configurations for the family tree, depending on whether C, E and F branch earlier or later. As before, black squares denote generations in which SNPs occur. This figure is a simplification of the situation in the House of Stewart.



In either case, the total number of years can be found by summing the lengths ABCDEF→ABC + ABC→A + ABC→B + ABC→C + ABCDEF→D + ABCDEF→E + ABCDEF→F. However, the uncertainties are larger than for a family where we know the entire family tree. We can better account for structure we know (e.g. the relationship between D & E is fixed by the SNP at DE) than for structure we can't (e.g. the relationship between D & F). This can lead to a systematic bias towards a higher number of years/SNP for large families if there is a long period between ABCDEF and DE where no SNPs occur. It is suspected that this is the reason that the Clan Donald and House of Stewart results give comparatively large rates for BigY tests. Note that these extra uncertainties are not fully accounted for in the previous figure.

BigY tests from other families only account for around 10,000 years of lineages. Although the uncertainties on these are larger, they roughly show the same rate as the bulk literature, as illustrated on the next page.

RATES FROM THE LITERATURE

A growing number of studies are performing thorough analyses of the human genome mutation rate. Only a few of these directly provide rates specific to the Y chromosome. At their best, these are studies of large lists of known genealogies, where the years between each father and son are added up, along with the mutations that have accumulated during that time. The ratio of these directly gives the mutation rate. Such studies include Xue et al. (2009) and Helgason et al. (2015). Helgason et al. uniquely provides two estimates, for the palindromic and non-palindromic regions, which show a marginal difference in mutation rate of around 18%. The slower palindromic rate is consistent with the paternally transmitted autosomal rate.

Some studies measure the rate of autosomal mutations in father–mother–child ("triplet") groups and scale this rate to the Y chromosome, based on the ratio of expected mutations inherited from the father and the mother. Examples include Mendez et al. (2013) and Scozzari et al. (2013). These rates are consistent with the slower (palindromic) rate from Helgason et al.

A third method involves taking an genetic-cultural group with a known date of origin, and computing a mutation rate based on that age. Examples include Poznik et al. (2013) for Native Americans, and Francalacci et al. (2013) for Sardinians. Here, we add an extra 10-20% uncertainty to their rates, to reflect the uncertainty in the date at which the tested population formed.

A final method relies on ancient DNA from archaeological remains. Such remains include the Ust'-Ishim burial in western Siberia, Mal'ta Boy and Kostenki 14. Together, these represent over 100,000 years of lineage. Examples include Fu et al. (2014), Karmin et al. (2015) and Trombetta et al. (2015). As these results rely on the archaeological remains themselves, care should be taken when

Consistency among these different methods clearly shows that the mutation rate has not changed significantly over tens of millenia.

RATES FROM ARCHAEOLOGICAL REMAINS

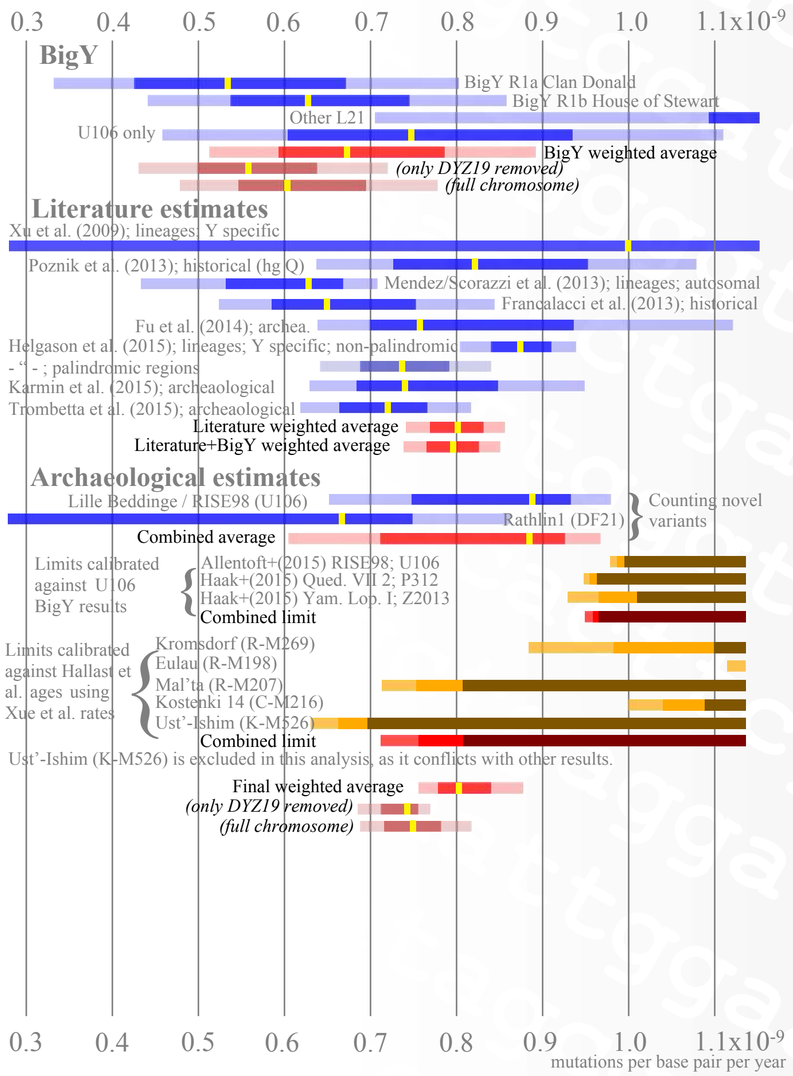
Obtaining rates from archaeological remains depends on having a known date for the archaeological remains, a known haplogroup for those remains (and preferably a good idea of how long it was between the formation of the haplogroup and the individual's lifetime), and the average number of SNPs formed since that haplogroup's formation in present-day lineages.

Typically, ancient DNA results will return an age based on ^{14}C dating, and a haplogroup. Where a study using a known rate has counted the number of SNPs in a modern population of the same haplogroup (e.g. Hallast et al. 2014), a new rate can be estimated based on the ratio of their age to the ^{14}C age, multiplied by the mutation rate they assume. (Note that the Hallast et al. study does not calculate its dates directly from the number of SNPs, but by the rho statistic, hence these limiting rates are indicative only, and should not be rigorously applied.) Normally only limits can be found from archaeological remains, except in cases where novel variants can be accurately found and counted in the ancient DNA.

COMBINING RATES FROM DIFFERENT ESTIMATES

We can use weighted averages to combine the estimates provided by different studies, while taking care to avoid circular reasoning. The chart below shows the mutation rate in SNPs per year per base pair. It can be read as follows:

- Blue lines show the results from individual studies. The yellow point marks the best-estimate value, and the shaded blue regions show the 68.3% and 95% confidence intervals.
- Red lines show the weighted average of several results. Here, the square of the confidence range is used as a weight. Red-grey lines show averages applied to different constraints on the BigY test results.
- Orange lines show the limits obtained from archaeological data. The different shadings (darker→lighter) show the regions ruled out at 99.75%, 95%, 68.3% and 50% confidence.



FINAL MUTATION RATE

The mutation rate that is finally used in this document and elsewhere in the U106 group's output is a weighted combination of the BigY and literature results, limited by the archaeological remains.

This results in a rate which (as of 21 Jan 2016) is 198 years/SNP, with a 95% confidence interval of 181–209 years/SNP, for the euchromatic, non-palindromic region of BigY tests. The full test minus DYZ19 is 129 years/SNP (125–139 years/SNP) and for all bases reported in the BigY test is 125 years/SNP (116–137 years/SNP).

COMPARISON TO YFULL

YFull.com also operate their own age-dating system, which works on a similar basis to ours. The major differences are that we apply more rigorous causality checks, particularly to our uncertainty estimates and, conversely, YFull has the luxury of making its own variant identification from the BAM file. YFull assumes a coverage of 8467165 base pairs compared to our 8753254. Our ages compare to theirs as follows (ages obtained 21 Jan 2016):

	YFull (years)	Our data (years)
M269	6400 (7300–5500)	
.L23	6200 (6900–5600)	
..L51	5800 (6400–5200)	
…L151	4900 (5400–4500)	
….U106	4900 (5400–4500)	4904 (5230–4449)
…..Z18	3500 (4000–3100)	4425 (5155–3497)
……Z372	3000 (3800–2200)	4136 (4756–3423)
…….L257	2800 (4300–1700)	3846 (4377–3249)
…..Z381	4900 (5400–4500)	4720 (5200–3962)
……Z156	4900 (5700–4200)	4535 (5170–3684)
……Z304	4200 (4700–3600)	3615 (3918–3224)
……Z301	4800 (5400–4200)	
…….L48	4800 (5400–4200)	4543 (5170–3705)
……..L47	4500 (5300–3800)	4128 (4644–3533)
……..Z9	4800 (5400–4200)	3878 (4191–3878)
……Z2	4200 (4800–3700)	3665 (3963–3665)
…….Z8	2900 (3500–2300)	2454 (2673–2177)

Notably almost all our ages agree within the uncertainties. The only exception, Z9, is very close and statistically acceptable in this ensemble.

Which is the more accurate test is a complex question to answer: we have more tests, but YFull can perform better and more homogeneous analysis from the BAM file directly. For well-populated clades (>20 tests in YFull), where the full known branching structure is present in the YFull tree, the YFull ages should be more accurate. For small clades, or where branches not present in the YFull tree, our ages should be more accurate. The absolute calibration of both systems matches very well at the U106 level.

CALIBRATING STR TO SNP MUTATION RATES

STR markers also seem to behave like a randomly ticking "clock", so in principle these can be used for age measurements as well. The advantage of using STR markers is that, typically, more people within a clade will have tested for these.

STR dates also provide us with some difficulty. They mutate up and down at a much faster rate than SNPs, so 111 STR markers provides about the same mutation rate as the SNPs in a 10-million-base-pair BigY test. This means that mutations back to the ancestral state are a problem. They can also mutate by more than one step at a time, and the decision has to be made as to whether to count this as one mutation or several. Finally, they also seem to prefer specific values, so will preferentially mutate to these lengths.

This makes the concept of STR dating much more mathematically complicated than SNP dating. Over timescales of a few hundred years, the above problems are negigible, but on longer timescales they become very significant. Usually an exponential multiplier is used to correct STR dates to SNP dates. In the following, we tie the STR age to the SNP ages within U106 using such a scaling relation.

To begin, we discuss methods of age calculation. Each relies on setting an mutation rate for each STR, the source of which we will discuss later.

Infinite allele model: The infinite allele model assumes any variance in an STR is a single mutation, e.g. it treats 15→17 as one "multi-step" mutation, whereas it is possible it was really two mutations: 15→16→17. Generally speaking, the infinite allele model will be more accurate for young clades. For old clades where more than one mutation is likely on some STRs, the step-wise allele model is better.

Step-wise allele model: The step-wise allele model assumes each repeat of an STR is a unique mutation. It counts mutations like 15→17 as two mutations: 15→16→17. Often a hybrid is used which assumes step-wise for all STRs except the multi-copy markers, which are infinite. We do not consider the step-wise model further here.

Variance-based model: Both the step-wise and infinite allele models do not correctly account for back mutations, e.g. 15→16→15. The variance-based method accounts for them in part by taking the mathematical variance of a group of DNA tests, rather than simply counting the mutations.

The infinite allele model we use derives from Dean McGee's tool (http://www.mymcgee.com/tools/yutility.html), which calculates the time to most recent common ancestor (TMRCA) for a grid of individuals. This data can then be combined using the method described below.

The variance-based model is based on a method and tool developed by Ken Nordtvelt, which has undergone substantial modification to include error estimates and include a number of easily changeable options.

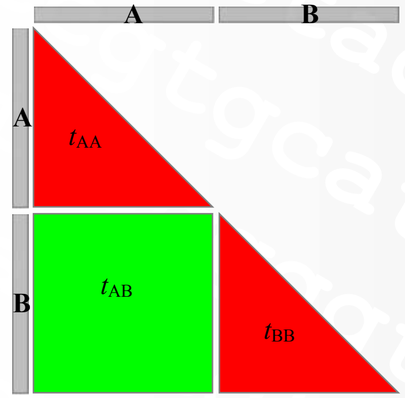
INTRA-CLADE AND INTER-CLADE AGES

McGee's tool calculates TMRCA for two individuals. What we require is the TMRCA for an entire group. In combining age estimates, it is important to consider whether you want the age within a group (the *intra*-clade age) or the age between two groups (the *inter*-clade age): e.g., do you wish to know the relationship between people who are U106+, or the age when Z18 and Z381 last shared a common ancestor?

Intra-clade ages are generally problematic, as they ignore the fact that many people within a clade are closely related: e.g., many calculations of the intra-clade age of U106 will be biased by the fact that half of people are L48+. The calculated age will be pulled down towards the L48 age. For this reason, inter-clade ages are generally used for STR calculations, which compare two clades to each other.

INFINITE AGE COMBINATIONS

For this method, the McGee tool outputs a tabulated matrix of TMRCAs. Assuming that clade "A" is listed at the top and clade "B" is listed at the bottom, the intra-clade TMRCAs of A and B (t_{AA},t_{BB}), and the inter-clade TMRCA of A and B (t_{AB}) will be given from the intersection of these two sets, which will fall in this region of the table:



Either the average or median value can be taken here as an estimation of the TMRCA of the A–B relationship, and the sample standard deviation can be taken as the standard error on this value. On top of this, there will be a systematic error to account for the uncertainties in the mutation rates, and the dataset must be calibrated against the SNP rates to account for non-random elements in the mutations.

The final age is therefore given by:

$$t_{AB} = \frac{t_{A1–B1} + t_{A1–B2} + \ldots + t_{A1–Bn} + t_{A2–B1} + \ldots + t_{Am–Bn}}{mn}$$

where there are m tests from clade A (A1 through Am) and n tests from clade B (B1 through Bn). The uncertainty is given by:

$$\delta t^2_{AB} = \frac{\sigma(t_{AB})^2}{mn} + \frac{\sigma(\Sigma_{i=1}^{111} \mu_i w_i)^2}{(\Sigma_{i=1}^{111} \mu_i w_i)^2}$$

where $\sigma(t_{AB})$ is the standard deviation among all TMRCAs in the A–B set, μ_i is the mutation rate on marker i and w_i represents a weighting factor which is the fraction of test pairs which are compared on marker i. The left-hand ratio therefore represents the square of the standard error in the mean, and the right-hand ratio represents the square of the fractional uncertainty in the mutation rate. The square root of this gives δt_{AB}, the uncertainty in t_{AB}.

VARIANCE-BASED AGE CALCULATION

Each marker i in test j returns an allele value $x_{i,j}$. The variance among m and n tests in clades A and B, respectively, can be calculated as:

$$\mathrm{Var(AB)}_i = s_{2,A}/m + s_{2,B}/m - 2\, s_{1,A}\, s_{1,B} / mn \,,$$

where $s_{1,A} = \Sigma_{j=1}^{m} x_{i,j}$, and $s_{2,A} = \Sigma_{j=1}^{m} x^2_{i,j}$, and similarly for $s_{1,B}$ and $s_{2,B}$ for $j = 1$ to n. The square of the fractional uncertainty in that variance (at the 68% confidence interval) will be:

$$\sigma^2(\mathrm{Var(AB)})_i \,/\, \mathrm{Var(AB)}^2_i = 2\,(m–1)m^{-2} + 2\,(n–1)n^{-2}\,.$$

Variances on individual markers can be summed, such that:

$$\mathrm{Var(AB)} = \Sigma_{i=1}^{111} \mathrm{Var(AB)}_i$$

with a 68% c.i. fractional uncertainty of:

$$\sigma(\mathrm{Var(AB)})/\mathrm{Var(AB)} = \sqrt{[\Sigma_{i=1}^{111} \sigma^2(\mathrm{Var(AB)})_i \,/\, \mathrm{Var(AB)}^2_i]} \,/\, 111$$

Using a mutation rate for each marker, μ_i, the age of the clade can be deduced by:

$$t(\mathrm{AB}) = \mathrm{Var(AB)}\, \Sigma_{i=1}^{111} \mu_i \,/\, 2$$

and:

$$\sigma(t(\mathrm{AB})) = t(\mathrm{AB})\, \sqrt{\{[\sigma(\mathrm{Var(AB)})/\mathrm{Var(AB)}]^2 + [\sqrt{\Sigma_{i=1}^{111} \sigma^2(\mu_i)}]^2\}}$$

where $\sigma(\mu_i)$ is the (68%) uncertainty in μ_i.

YEARS PER GENERATION

Conventionally, STR mutation rates are given in mutations per generation, whereas we need mutations per year. The conversion of years per generation has adopted many values between 20 and 40 years/gen in the literature. The value varies over time and over societies. Historical studies of populations (particularly in Iceland) indicate it is likely to have been around 35 years/generation over the 16th to 19th Centuries. Since then, a series of scientific and social revolutions have decreased the years/generation (19th Century sanitation improvements, 20th Century medical improvements, birth control) and subsequently increased it again (women's lib. and two-career families).

For pre-modern agrarian communities between 1000 AD and the present, we adopt 35 +/– 3 years/generation (at 95% confidence). For earlier times, we adopt a scaling that drops to 33 +/– 3 years/gen for 1-1000 AD, 32 +/– 3 years/gen for 1000-1 BC, and 31.5 +/– 3 years/SNP before 1000 BC. Throughout, we adopt a zero point of 1950 AD, +/- 15.5 years at 95% confidence.

CHOICE OF MARKERS

There are various reasons why certain markers may be avoided. These include multi-copy markers like DYS464, where we cannot always tell which value belongs to each copy (e.g. 15-16-17-18 could be a=15, b=16, c=17, d=18 or a=18, b=16, c=17, d=15). We might also select only slowly-mutating markers to select against non-random elements in the mutation process. One final possibility is to use q values (a measure of closeness to random mutation; Bird et al. 2012) to select only STRs that mutate in a close-to-random fashion.

CHOICE OF MUTATION RATES

A variety of mutation rates exist in the literature, with a substantial range in mutation rates. We consider a number of rates here. In the table, the mutation rate source is listed, along with the number of markers contained, the number of those markers used in the following analysis, and the relative mutation rate compared to the average of the ensemble for the markers sampled, where larger numbers indicate faster mutations.

Chandler (2006)	67	50	78%
Doug McDonald (unpub.)	80	not used	247%
Charles Kerchner (unpub.)	67	not used	304%
SMGF	30	not used	109%
FTDNA	37	not used	268%
SMGF/Y-Search	21	not used	117%
Y-HRD	16	not used	83%
Vermeulen et al. (2009)	8	not used	143%
Marko Heinila (unpub.)	111	94	78%
Ballantyne et al. (2010)	91	82	118%
Burgarella et al. (2011)	84	83	118%

We have chosen the indicated four datasets on the basis of number of markers covered and consistency with the average STR mutation rate.

The standard deviation of these four rates over the square root of the average number of rates per marker (3.18) approximates the uncertainty in the rate itself, which we take as our standard (68% c.i.) uncertainty. Overall, this yields a 8.8% systematic uncertainty in the total mutation rate and typically a 5.6% statistical uncertainty in the resulting age at a 68% confidence interval. For comparison, all sources of systematic and statistical error typically yield at least a 11% (21%) uncertainty in the age in generations or a 14% (28%) uncertainty in the age in years at the 68% (95%) confidence intervals.

CONSTRAINTS APPLIED IN THE FOLLOWING

The constraints listed below were applied to the analysis that follows.

- No multi-copy markers were used in any calculation. This leaves 94/111 markers.
- No selection with mutation rate was made unless noted. Where noted, markers with μ>0.004 per generation were discounted unless stated otherwise (leaving 78 markers).
- No selection with Bird's q were made unless noted. Where noted, markers with Bird's q>0.05 were discounted unless stated otherwise (leaving 40 markers).

The 94 markers used give a mutation rate of μ=0.315+/-0.028 per generation, or once per 111+/-14 years.

DATA USED IN THE ANALYSIS

The U106 group's STR database was sampled on 3rd June 2015, and includes 2058 STR entries. Of these, 1722 have a relatively secure placement in a clade under U106, with 141728 markers in total, or an average of 82.3 markers each.

CALIBRATION OF STR TO SNP AGES: VARIANCES

STR ages are usually boot-strapped to SNP ages using some variation on the following expression:

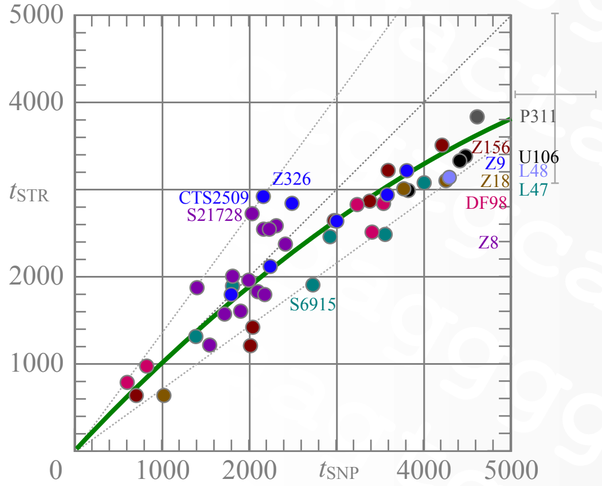
$$t_{STR,corr} = t_{STR,unc} \exp(-t_{STR,unc}/f)$$

where $t_{STR,corr}$ and $t_{STR,unc}$ are the uncorrected and corrected ages derived from STRs, respectively, and f is a fitted scaling factor based on calibration to the SNP-derived ages. We fit the following formula:

$$t_{STR,corr} = t_{STR,unc} f_2 \exp(-t_{STR,unc}/f_1)$$

with two fitting factors, to allow for uncertainties in the systematic calibration of both ages.

The graph below shows the scaling factors derived for the variance method. The details of this data and the fit can be found in the supplementary spreadsheet (str-ages.xls).



The points are colour-coded following the same clade-based scheme used later in the document. The diagonal gray line represents a 1:1 correlation, and the adjacent lines show the tolerable range of fits based on the typical systematic uncertainty (illustrated on the right). The solid green line shows our best fit. The following fitting parameters are derived:

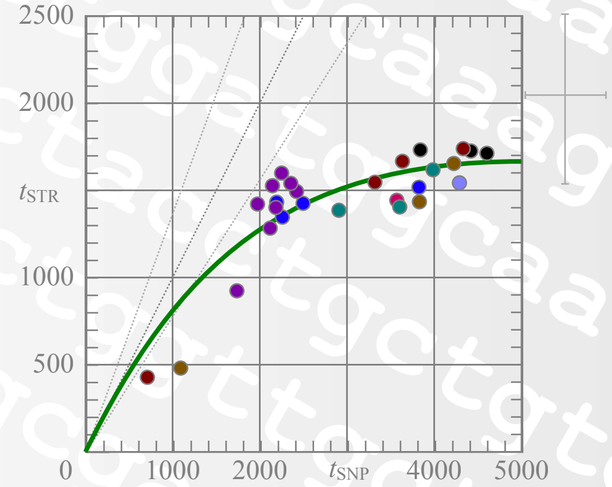
f = 18410 +/– 2096, f_1 = 15000 +/– 4114, f_2 = 1.04 +/– 0.07.

Corrections become important for this method if the predicted age exceeds about 2500 years old. No statistical difference is determined between the one- and two-parameter fits.

Scatter between the two age estimates is around +/- 300 years (standard deviation). This could be reduced by adopting the same "top-down" constraint to the STR-based ages as is currently applied to the SNP-based ages.

CALIBRATION OF STR TO SNP AGES: INFINITE ALLELES

The figure below is similar to the one in the previous panel, except for the infinite alleles method. Note the expanded range on the vertical axis.



The following fitting parameters are derived:

f = 4436 +/– 117, f_1 = 4519 +/– 451, f_2 = 0.99 +/– 0.07.

Corrections may become important at any age. Again, no significant statistical difference is found between the one- and two-parameter fits. The ages asymptote to a much younger age (around 1700 years). Corrections become important in less than 1000 years, and ages more than about 2000 years cannot be meaningfully corrected.

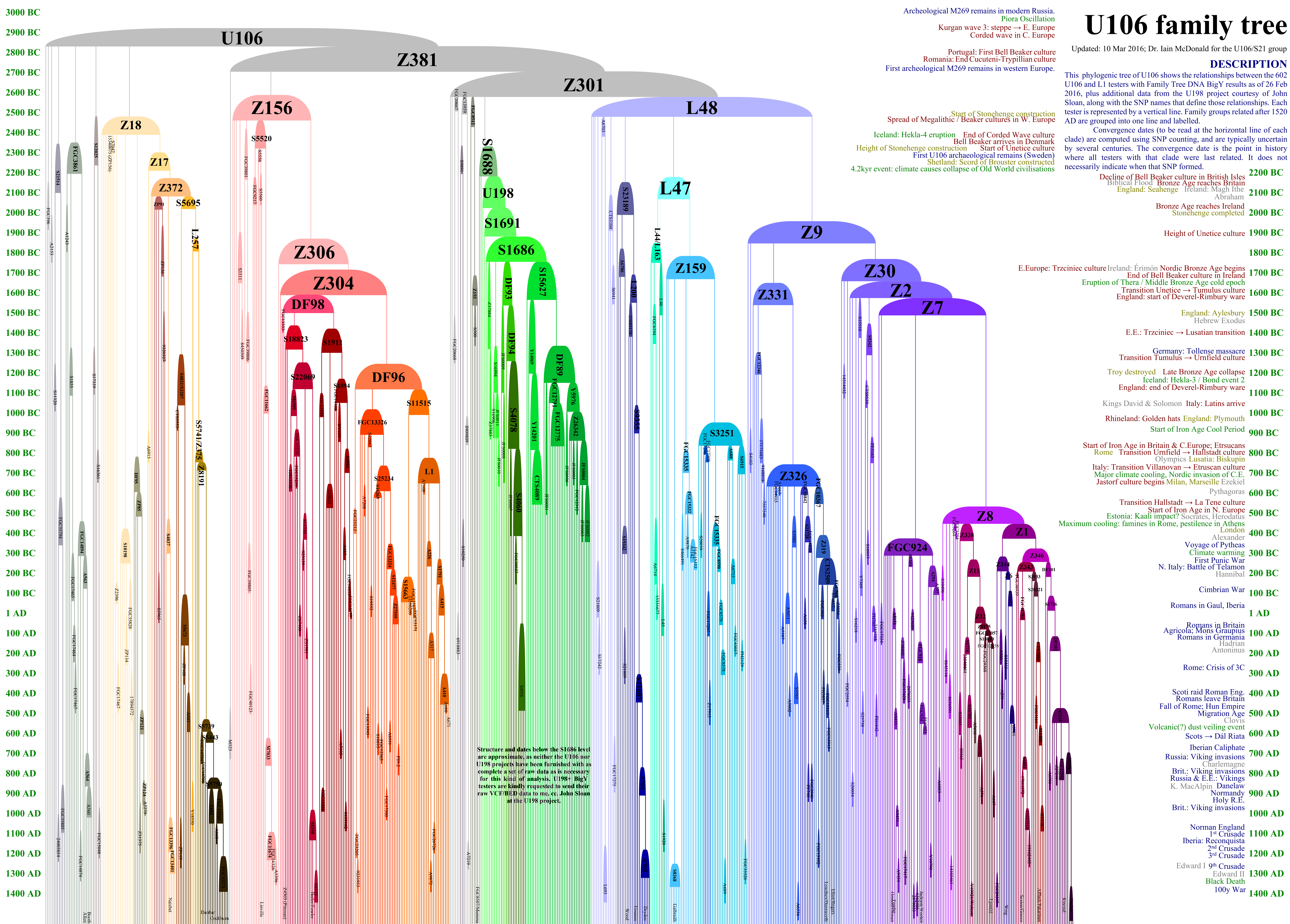
U106 family tree

Updated: 10 Mar 2016; Dr. Iain McDonald for the U106/S21 group

DESCRIPTION

This phylogenic tree of U106 shows the relationships between the 602 U106 and L1 testers with Family Tree DNA BigY results as of 26 Feb 2016, plus additional data from the U198 project courtesy of John Sloan, along with the SNP names that define those relationships. Each tester is represented by a vertical line. Family groups related after 1520 AD are grouped into one line and labelled.

Convergence dates (to be read at the horizontal line of each clade) are computed using SNP counting, and are typically uncertain by several centuries. The convergence date is the point in history where all testers with that clade were last related. It does not necessarily indicate when that SNP formed.



3000 BC
2900 BC
2800 BC
2700 BC
2600 BC
2500 BC
2400 BC
2300 BC
2200 BC
2100 BC
2000 BC
1900 BC
1800 BC
1700 BC
1600 BC
1500 BC
1400 BC
1300 BC
1200 BC
1100 BC
1000 BC
900 BC
800 BC
700 BC
600 BC
500 BC
400 BC
300 BC
200 BC
100 BC
1 AD
100 AD
200 AD
300 AD
400 AD
500 AD
600 AD
700 AD
800 AD
900 AD
1000 AD
1100 AD
1200 AD
1300 AD
1400 AD
U106
Z381
Z301
Z156
L48
Z18
Z17
Z372
S5695
L257
S5520
Z306
Z304
DF98
S18823
S1911
S22069
DF96
S11515
FGC13326
L1
S1688
U198
S1691
S1686
DF93
S15627
DF94
DF89
S4078
L47
S23189
Z159
Z9
Z30
Z2
Z7
Z331
S3251
Z326
Z8
Z1
FGC924
Archeological M269 remains in modern Russia.
Piora Oscillation
Kurgan wave 3: steppe → E. Europe
Corded wave in C. Europe
Portugal: First Bell Beaker culture
Romania: End Cucuteni-Trypillian culture
First archeological M269 remains in western Europe.
Start of Stonehenge construction
Spread of Megalithic / Beaker cultures in W. Europe
Iceland: Hekla-4 eruption
End of Corded Wave culture
Bell Beaker arrives in Denmark
Height of Stonehenge construction
Start of Unetice culture
First U106 archaeological remains (Sweden)
Shetland: Scord of Brouster constructed
4.2kyr event: climate causes collapse of Old World civilisations
Decline of Bell Beaker culture in British Isles
Biblical Flood
Bronze Age reaches Britain
England: Seahenge
Ireland: Magh Ithe
Abraham
Bronze Age reaches Ireland
Stonehenge completed
Height of Unetice culture
E.Europe: Trzciniec culture
Ireland: Érimón
Nordic Bronze Age begins
End of Bell Beaker culture in Ireland
Eruption of Thera / Middle Bronze Age cold epoch
Transition Unetice → Tumulus culture
England: start of Deverel-Rimbury ware
England: Aylesbury
Hebrew Exodus
E.E.: Trzciniec → Lusatian transition
Germany: Tollense massacre
Transition Tumulus → Urnfield culture
Troy destroyed
Late Bronze Age collapse
Iceland: Hekla-3 / Bond event 2
England: end of Deverel-Rimbury ware
Kings David & Solomon
Italy: Latins arrive
Rhineland: Golden hats
England: Plymouth
Start of Iron Age Cool Period
Start of Iron Age in Britain & C.Europe; Etruscans
Rome
Transition Urnfield → Hallstadt culture
Olympics
Lusatia: Biskupin
Italy: Transition Villanovan → Etruscan culture
Major climate cooling, Nordic invasion of C.E.
Jastorf culture begins
Milan, Marseille
Ezekiel
Pythagoras
Transition Hallstadt → La Tene culture
Start of Iron Age in N. Europe
Estonia: Kaali impact?
Socrates, Herodatus
Maximum cooling: famines in Rome, pestilence in Athens
London
Alexander
Voyage of Pytheas
Climate warming
First Punic War
N. Italy: Battle of Telamon
Hannibal
Cimbrian War
Romans in Gaul, Iberia
Romans in Britain
Agricola; Mons Graupius
Romans in Germania
Hadrian
Antoninus
Rome: Crisis of 3C
Scoti raid Roman Eng.
Romans leave Britain
Fall of Rome; Hun Empire
Migration Age
Clovis
Volcanic(?) dust veiling event
Scots → Dál Riata
Iberian Caliphate
Russia: Viking invasions
Charlemagne
Brit.: Viking invasions
Russia & E.E.: Vikings
K. MacAlpin
Danelaw
Normandy
Holy R.E.
Brit.: Viking invasions
Norman England
1st Crusade
Iberia: Reconquista
2nd Crusade
3rd Crusade
Edward I
9th Crusade
Edward II
Black Death
100y War
Structure and dates below the S1686 level are approximate, as neither the U106 nor U198 projects have been furnished with as complete a set of raw data as is necessary for this kind of analysis. U198+ BigY testers are kindly requested to send their raw VCF/BED data to me, cc. John Sloan at the U198 project.

Geographical distribution of U106

Updated: 11 February 2016
Dr. Iain McDonald
on behalf of the U106/S21 group

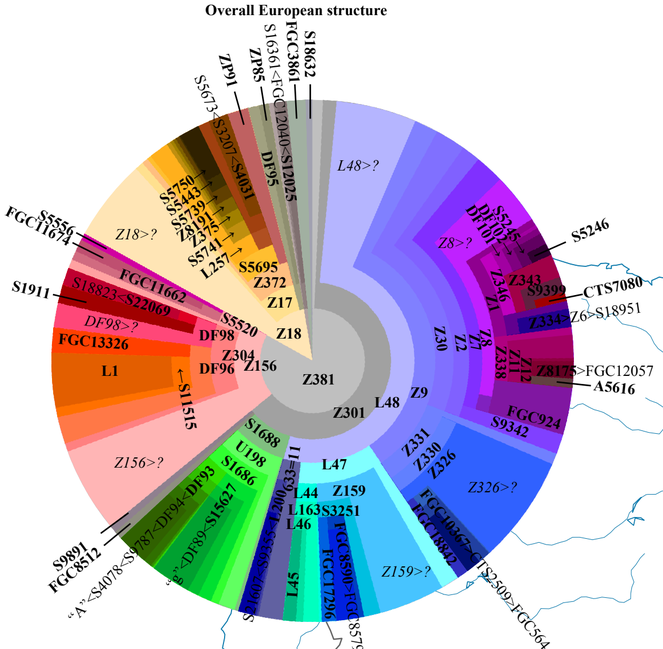
Distribution of all U106 by region and sub-clade

This represents the geographical and phylogenic distribution of 1613 U106+ tests from Family Tree DNA. These include members from the U106, U198 and L1 haplogroup projects, and several geographical projects. Information was collected during February 2016. Geography is self-reported by the testers. Phylogenic position is based on the last SNP tested, thus some testers may branch into lower sub-clades for which they have not been tested.

● = 1 tested person

The proportions of each clade are given for each region. The size of the pie chart is scaled to the size of the tested population. Note that multiple tests from the same family will skew the distributions, and that this has not been accounted for here.

Overall European structure



Migrations

METHODS TO IDENTIFY MIGRATION PATHS

There are three primary ways to work out how and when a clade spread.
(1) Look at the current distribution of people. This tells you something about who is related to who, but not when and why.
(2) Look at archaeological DNA results. As already discussed, this is very good at determining upper limits to when clades formed. It can also tell you something about the earliest phases of a population's presence in an area. However, these are only glimpses: snapshots into a forgotten world, and very few and far between. There's only so much information they can give on particular time periods and particular migrations, unless they happen to be very large. Nevertheless, this is the most effective method for older populations.
(3) Look at the ages of MRCAs from different countries. This is perhaps the most powerful tool for more recent populations, but perhaps also the most difficult to obtain good data from. We will discuss this later.

CURRENT DISTRIBUTION

The current relative distribution of U106 and its subclades can be found on the following pages. These come from a compilation of projects at Family Tree DNA, not least the U106 project itself. They are therefore biased by the content of those projects.

Some projects are more active than others at recruiting members,.Some are more active in getting members to test to more-recent SNPs. Some families have DNA tested many members, some only one, despite being of the same size in the present-day population. These fractions should not be taken as absolute proportions, but as guides or indicators for further work.

ARCHAEOLOGICAL Y-DNA

Following the maps of current distribution are several maps showing the archaeological DNA results up to 2000 BC, shortly after U106 formed. These are broken up by period to highlight the differences between them.

From these, it can clearly be seen that haplogroup R was essentially absent from Europe until some time shortly before 2600 BC. It was, however, present in modern Russia, and this has been used to indicate a rapid spread of both R1a and R1b into Europe, during the period circa 3300 BC to 2500 BC (see Haak et al. 2015). This has been associated with the archaeological Kurgan and Yamnaya cultures, and can possibly be credited with bringing Indo-European language and culture to Europe.

THE ROUTE INTO EUROPE

The route our ancestors took into Europe is not precisely known. From the ancient DNA record, R1a and R1b both seem to appear "overnight" sometime during the early third millenium BC.

The exact origin of these people is not known. They may have come from as far south as the southern Caucasus, or as far north as the tundra-covered northern Ural mountains. What we do know is that they somehow spawned the Yamnaya and other cultures who lived north of the Black Sea during the last fourth millenium BC.

It is thought that R1a, at least, helped create the Corded Ware cultural horizon in north-eastern Europe. It is not clear whether or not R1b came with them, due to the relatively smaller number of R1b DNA results during this crucial period.

There are two likely routes of R1b into Europe. The first is to the north of the Carpathian Mountains, through relatively flat plains of modern-day Poland. This follows the Corded Ware culture, and there are some slight preferences for this route from ancient DNA.

The second is down the Black Sea coast and up the River Danube. This is the preferred route from analysing the geography of M269+ U106- P312- Y-DNA results using a "minimal spanning tree"-like method.

A third route, arriving from modern-day Turkey with the advent of farming at the beginning of the Neolithic, appears ruled out by the dates obtained from our results, and the dates and places obtained from ancient DNA.

Distinguishing between the two remaining routes cannot yet be done with confidence. It will need better constraints – either in time or place – from archaeological DNA.

LOCATION OF THE FIRST U106

It is clear that the first major place of U106 settlement was in modern-day Germany, whether it was on the border with Poland, or with Austria, or as far west as the Rhine valley. The exact location depends on the migration pathway into Germany, and the exact time that U106 formed relative to the migration westwards.

Either way, our earliest U106 ancestors were very probably German. U106 has often thought to be the `Germanic' cousin of the `Celtic' P312. In fact, these are misnomers, as both U106 and P312 predate these cultures by over 1000 years. More properly, early U106 probably formed much of the Bell Beaker cultures of central Europe, and later the western half of the Unetice culture.

The earliest (and so far only) ancient U106 burial is dated to between 2275 and 2032 BC, and comes from the Nordic Bronze Age culture of southern Sweden (Lilla Beddinge), rather than Germany. Although likely from several centuries after the formation of U106, this indicates that U106 spread quite quickly and effectively to these areas. Sadly, we do not currently know of any descendents of this particular branch of U106, which may have died out.

Later pages in this section show this formation and dispersion of U106 graphically.

FURTHER ANCIENT DNA

This section is left blank for further DNA results as they arrive.

6500-4000 BC: the situation in Europe

Updated: 16 June 2015
Dr. Iain McDonald
on behalf of the U106/S21 group

DESCRIPTION

This page details the archaeological DNA obtained from burials before 4000 BC, which show the context of Europe into which R-M269 arrived. Below are the symbols used in this page:

Haplogroup I:
Haplogroup C:
Haplogroup E:
Haplogroup G:
Haplogroup R:
Other haplogroup:

Opacity scales with age, such that the above represent (from left to right) ages of <6000 BC, 6000-5500 BC, 5500-5000 BC, 5000-4500 BC and 4500-4000 BC.

Yuzhnyy Oleni Ostrov
R1a1-M459, Pages65.2
Haak+ 2015
5500-5000 BC

R1a

R1b

Scandinavia: 100% I

Russia: 100% R

Sok River
R1b1-M343,L278
(xM478,M269)
Haak+ 2015
5650-5555 BC

Motala
I2c2, various I2a1
Haak+ 2015
5898-5331 BC

Serteya
R1a1
Chekunova 2014
4000 BC

Central Europe:
58% G
21% I
8% C
4% T, H, E

Halberstadt
3*G2a2a; 2*G2a2a1
Haak+ 2015
various: 5207-4946 BC
LBK

Karsdorf
T1a
Haak+ 2015
5207-5070 BC
LBK

Derenburg
2*F-M89 (xGHIJK), G2a2b
Brandt 2013
5300 - c. 5000 BC
LBK

Loschbour
I2a1
Haak+ 2015
6220-5990 BC

Szőlőskert-Tangazdaság
G2a2b
Szécsényi-Nagy 2014
est. 5100 BC
LBKT

Berekalja
I2a
Gamba 2014
4490-4360 BC
Lengyel

Berekalja
C1a2
Szécsenyi-Nagy 2014
5300-4950 BC

Kompolt-Kigyoser
C1a2
Gamba 2014
5210-4990 BC
Late ALP

Tiszaszőlős-Domaháza
I2a
Szécsényi-Nagy 2014
5780-5650 BC

Bicske-Galagonyás
E1b1b1a1
Szécsényi-Nagy 2015
5000-4800 BC
Sopot

Balatonszemes-Bagódomb
I1-M253
Szécsényi-Nagy 2014
est. 5000 BC
LBKT

Tolna-Mözs
G2a2b, F-M89 (GHIJK?)
Szécsényi-Nagy 2014
various dates between 5300-5020 BC

Alsónyék-Bátaszék & Lánycsók
2*F (GHIJK?), H2, G2, 3*G2a, G2a2b1
Szécsényi-Nagy 2014
various dates between 5840-5550 BC

Alsónyék-elkerülő
J2
Szécsényi-Nagy 2015
5000-4910
Sopot

Vinkovci & Vukovar
5 G2a, G2a, I2a1
Szécsényi-Nagy 2014
est. 6000-5500 BC

La Branya
C1a2
Olalde 2014
5940-5690 BC

El Portalón
H2, I2a2a
Gunther 2015
4960-4628 BC

Els Trocs
F (xG, xJ), I2a1b1
R1b1-M343(xM269)
Haak 2015
5311-5068 BC

Avellaner
3*G2a, E1b-M35.1+ V13+
Lacan 2011b
5000 BC
Epicardial

Iberia:
30% G
20% I
10% C
10% E
10% H
10% R

R1b-M343
R1b1-P25
R1b1a-P297
11300 BC (12900 BC - 9700 BC)
M73

The dark grey area shows the time period nominally covered by this chart. The light grey area shows the same period, allowing for uncertainties in dates.

R1b1a2-M269
4100 BC (5350 BC - 3200 BC)
R1b1a2a-L23
4000 BC (5000 BC - 3100 BC)
R1b1a2a1-L51
3500 BC (4450 BC - 2800 BC)

PF7558
Z2103
Z2106
L584
L277.1
Z2118
S1161
R1b1a2a1a-P310
2700 BC (3450 BC - 2150 BC)
S1194
P312
R1b1a2a1a1-U106
2600 BC (3050 BC - 2100 BC)

4000-3000 BC: the rise of M269

Updated: 17 June 2015
Dr. Iain McDonald
on behalf of the U106/S21 group

DESCRIPTION

This page details the archaeological DNA obtained from burials between 4000 and 3000 BC, which show Europe during the initial R-M269 expansion, before U106 formed. Below are the symbols used in this page:

Haplogroup I:
Haplogroup C:
Haplogroup E:
Haplogroup G:
Haplogroup R:
Other haplogroup:

Opacity scales with age, such that the above represent (from left to right) ages of 4000-3800 BC, 3800-3600 BC, 3600-3400 BC, 3400-3200 BC and 3200-3000 BC. Many dates are uncertain, but the central (usually most likely) estimate is used to select the symbol colour. The Serteya burial, dating to 4000 BC, is carried over from the previous page. Burials dating to 3000 BC are carried over onto the next page.

R1a

R1b

Lopatino
R1b-M269>L23>Z2015
2*R1b-P297xL51 (M269?)
Haak 2015
3339-2917 BC
Yamnaya

Kutuluk
R1b-M269>L23>Z2015
Haak 2015
3300-2700 BC
Yamnaya

Ishkinovka
R1b-M269>L23(xZ2015)
Haak 2015
3300-2700 BC
Yamnaya

Serteya
R1a1
Chekunova 2014
4000 BC

Russia: 100% R

Central Europe:

50% I
33% G
17% R?

Esperstedt
Haak+2015, Mathieson+2015
Esperstedt: 3887 - 3797 BC, I
Esperstedt: 3360 - 3086 BC, I2a1b1a
Salzmünde/Shishzig, 3400 - 3025 BC, G2a2a
F-P316* (xGHIJKNOP), 3887 - 3797 BC
Baalburg, Salzmünde/Bernburg_TRB

Quedlinburg
R-P224?
Haak+ 2015
3645-3537 BC
NB: possible damage

Peshany
R1b1a2-M269(xL51)
Haak 2015
3334-2635 BC
Yamnaya

Western Europe:

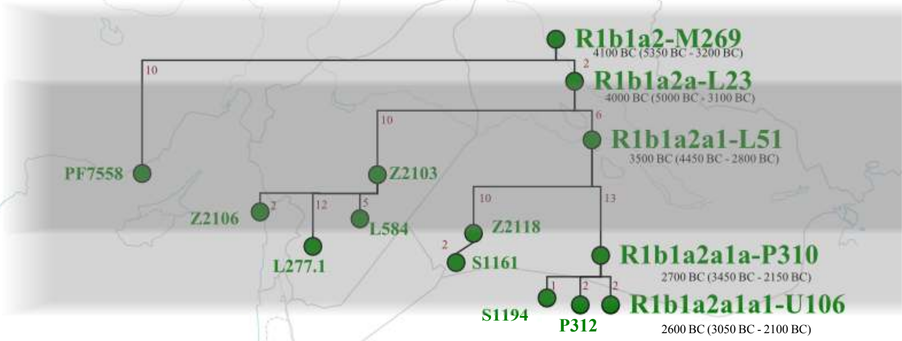
81% G
19% I

Otzi
G2a1b2-L91
Keller 2012
3350-3100 BC

Remedello di Sotto
I2 [I2a1a]
Allentoft+ 2015
3483-3107 BC

Trielles
20*G2a-P15,
2*I2a1-M438,P37.2
Lacan 2011
3000 BC

La Mina
2*I2a1a1 (or 1 H2?)
Haak+ 2015
3900-3600 BC
Megalithic



3000-2500 BC: M269 invades Europe

Updated: 17 June 2015
Dr. Iain McDonald
on behalf of the U106/S21 group

DESCRIPTION

This page details the archaeological DNA obtained from burials between 3000 and 2500 BC, which show Europe during the initial R-M269 expansion, before U106 formed. Below are the symbols used in this page:

Haplogroup I:
Haplogroup C:
Haplogroup E:
Haplogroup G:
Haplogroup R:
Other haplogroup:

Opacity scales with age, such that the above represent (from left to right) ages of 3000-2900 BC, 2900-2800 BC, 2800-2700 BC, 2700-2600 BC and 2600-2500 BC. Many dates are uncertain, but the central (usually most likely) estimate is used to select the symbol colour. Burials dating to 3000 BC are carried over from the previous page. The Ajvide burial on Gotland is included due to its large uncertainty, but good representation of the expected population (as observed in previous and later epochs). Tildes (~) prefix better-known SNPs which are phylogenically equivalent in tested modern populations.

Southern Scandinavia:
67% R
33% I

Ajvide
I2a1
Skoglund 2014
2800-2000 BC
Pitted Ware

Kyndelose
R1a [R1a1a1-Page7]
Allentoft+ 2015
2851-2492 BC
Nordic Middle Neolithic

Viby
R1a [R1a1a1-Page7]
Allentoft+ 2015
2621-2472 BC
Battle Axe

Central Europe:
75% R
8% G?
17% I (or J?)

Naumovo
R1a1
Chekunova 2014
2500 BC
Zhizhitskaya

Sertaya
R1a1 + N1c
Chekunova 2014
2500 BC
Zhizhitskaya

Oblaczkowo
R1b1
Allentoft+ 2015
2865-2578 BC
Corded Ware

Jagodno
G?, I or J?
Gworys 2013
2800 BC
Corded Ware

Kromsdorf
R1b-M269xU106, R1b-M343(M269?)
Lee 2012, Oliveri 2013
2600-2500 BC
Bell Beaker

Eulau
R1a1
Brandt 2013
2600 BC
Corded Ware

Bergheinfeld
R1a
[R1a1a1-M417xZ647]
Allentoft+ 2015
2829-2465 BC
Corded Ware

Tiefbrunn
R1a + R1
[2*R1a1a1]
Allentoft+ 2015
2880-2580 BC
Corded Ware

Dolmen de La Pierre Fritte
2*I2a1
Lacan 2011c
2750-2725 BC
Megalithic

Lánycsók
R1b-M343 + I2a2a
Szécsényi-Nagy 2015
2860-2620 BC

Remedello di Sotto
2*I2 [I2a1a1a-L672/S327]
Allentoft+ 2015
2908-2578 BC

Southern & Western Europe:
77% G
23% I

El Mirador
1*I2a1, 3*I2a2, 1*I, 2*G2a
Mathieson+ 2015
2880 - 2630 BC

Trielles
20*G2a-P15,
2*I2a1-M438,P37.2
Lacan 2011
3000 BC

R1a

Russia:
83% R
8% N
8% I

Ekaterinovka
R1b-M269>L23>Z2015
Haak 2015
2910-2875 BC
Yamnaya

Stalingrad Quarry
R1b [Z2107~Z2105]
Allentoft+ 2015
2857-2497 BC

Kutuluk
R1b-M269>L23>Z2015
Haak 2015
3300-2700 BC
Yamnaya

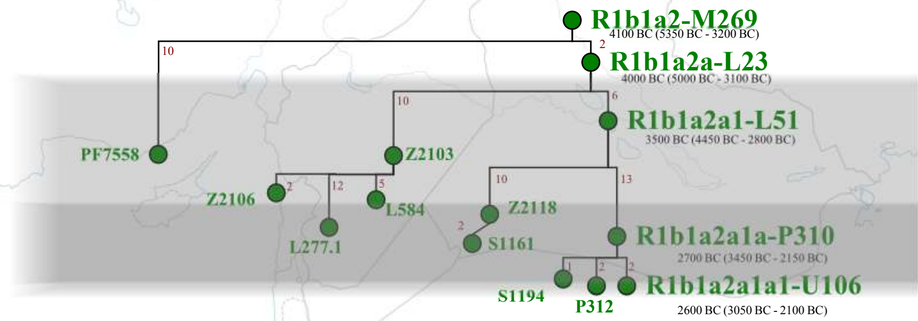
Ishkinovka
R1b-M269>L23(xZ2015)
Haak 2015
3300-2700 BC
Yamnaya

R1b

Ulan
I2a [I2a2a1b1b2-S12195]
Allentoft+ 2015
2849-2146 BC
Yamnaya

Termta
3*R1b [PF6482-M269, CTS9416~Z2105, Z2105]
Allentoft+ 2015
2887-2634 BC [1 of 3]
Yamnaya

Peshany
R1b1a2-M269(xL51)
Haak 2015
3334-2635 BC
Yamnaya



2500-2000 BC: early growth of U106

Updated: 17 June 2015
Dr. Iain McDonald
on behalf of the U106/S21 group

DESCRIPTION

This page details the archaeological DNA obtained from burials between 2500 and 2000 BC, which show Europe during the initial R-M269 expansion, before U106 formed. Below are the symbols used in this page:

Haplogroup I:
Haplogroup C:
Haplogroup E:
Haplogroup G:
Haplogroup R:
Other haplogroup:

Opacity scales with age, such that the above represent (from left to right) ages of 2500-2400 BC, 2400-2300 BC, 2300-2200 BC, 2200-2100 BC and 2100-2000 BC. Many dates are uncertain, but the central (usually most likely) estimate is used to select the symbol colour. Burials dating to around 2500 BC are carried over from the previous page. Tildes (~) prefix better-known SNPs which are phylogenically equivalent in tested modern populations.

Southern Scandinavia:
75% R
25% I

Central Europe:
54% R
46% I

Russia:
67% R
16% N
16% I

R1a

Rathlin
R1b:
P312>L21>DF13>DF21>Z30233, 2026-1885 BC
P312>L21>DF13, 2024-1741 BC
P312>L21, 1736-1534 BC
Cassidy+ 2015
Early Bronze Age

Marjberg
R1a [R1a1a1-Page7]
Allentoft+ 2015
2191-1972 BC
Nordic Late Neolithic

Ajvide
I2a1
Skoglund 2014
2800-2000 BC
Pitted Ware

Viby
R1a [R1a1a1-Page7]
Allentoft+ 2015
2621-2472 BC
Battle Axe

Lilla Beddinge
R1b-L106
Allentoft+ 2015
2275-2032 BC
Battle Axe / Nordic Late Neolithic

Esperstedt
R1a1a1*
Haak+ 2015
2473-2348 BC
Corded Ware

Łęki Małe
R1a [R1a1a1]
Allentoft+ 2015
2286-2048 BC
Corded Ware

Quedlinburg
R1b-P312
Haak+ 2015
2296-2206 BC
Bell Beaker

Eulau
I2
Haak+ 2015
2131-1982 BC [1 of 2]
Unetice

Esperstedt
R1a1a1*
Haak+ 2015
2473-2348 BC
Corded Ware
I2a2+I2c2
2131-1961 BC
Unetice

Knežević
R1b [P310]
Allentoft+ 2015
Undated
Bell Beaker

Ausburg/Untermeitingen
R1b [L150~M269]
Allentoft+ 2015
Undated
Bell Beaker

Österhofen
2*R1b [L51, P312>U152]
Allentoft+ 2015
Undated
Bell Beaker

Erd
I2 [I2a2a1a2a2-SK1247]
Allentoft+ 2015
Vatya

Foldvar
I2 [I2a2a-L596]
Allentoft+ 2015
2128-1909 BC
Vatya

Remedello di Sotto
I2 [I2a1a1a-L672]
Allentoft+ 2015
2134-1773 BC

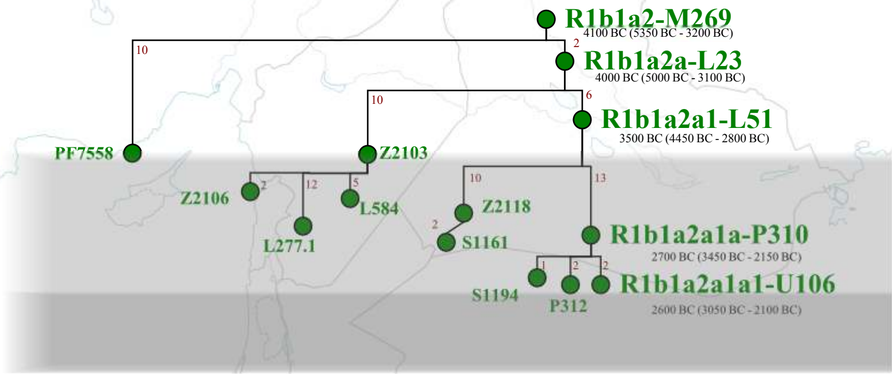
Naumovo
R1a1
Chekunova 2014
2500 BC
Zhizhitskaya

Sertaya
R1a1 + N1c
Chekunova 2014
2500 BC
Zhizhitskaya

Stepnoe
R1a [R1a1a1b-S224]
Allentoft+ 2015
2126-1896 BC
Sintashta

Bulanovo
R1a [R1a1a1b2a2-Z2124]
Allentoft+ 2015
2298 - 2045 BC
Sintashta

Ulan
I2a [I2a2a1b1b2-S12195]
Allentoft+ 2015
2849-2146 BC
Yamnaya



The origins of U106: 4400 BC to 2900 BC

Updated: 04 February 2016
Dr. Iain McDonald
on behalf of the U106/S21 group

WARNING!

THESE MAPS CONTAIN MANY INFERENCES THAT CANNOT CURRENTLY BE PROVED WITH ANY SCIENTIFIC RIGOUR, BUT WHICH ARE NECESSARY TO PRESENT A COHERENT PICTURE. THEY WILL NOT BE A TRUE REPRESENTATION OF HISTORICAL MIGRATIONS. THESE MAPS ONLY REPRESENT A STARTING HYPOTHESIS FOR CONTINUING WORK. DO NOT TAKE THESE RESULTS AS BEING THE ONLY VIABLE OPTION.

(1) Arrival into Europe

The origin of U106 can now be placed around 2900 BC. There is roughly a 2-in-3 chance of it being within 300 years of this date. Ancient DNA shows few haplogroup R men in Europe before about 3000 BC. It is known from branches further up the tree and archaeological results that haplogroup R arose in Asia. We can presume that U106 was founded somewhere late in this migration from Asia to Europe.

(2) Kurgan hypothesis

Key mutations often arise during population expansion events. These will typically shortly precede (or occur during) migration events when one group takes over another. The important Asia–Europe migration taking place around the time of U106's formation was the Kurgan expansion out of the Russian Steppe.

(3) Upstream SNPs

The geographical median of SNPs between M269 and U106 follows an east–west trend in surveys of both modern and ancient DNA. We can use this to infer that the M269→U106 sequence follows a migration from the east to the west. The Kurgan expansion is the only known, major migration that fits both the likely range of dates and the east–west movement. In addition it appears to arise from the trans-Ural area near concentrations of groups further up the haplogroup R1b tree (e.g. V88).

(4) Urheimat

The Gimbutas interpretation of the Kurgan hypothesis credits the Kurgans with the introduction of the Indo-European language family to Europe. The origin of this language is referred to as the *Urheimat*, and is generally considered to have been in the period 4200–3500 BC. Its location is unknown and may be anywhere from the Causcasus to the trans-Ural region shown here, or perhaps even further east.

(5) M269

M269 formed around 4400 BC, but the uncertainty in its age is roughly 600 years, so identifications with a particular culture are highly speculative. Kurgan wave 1, migration from the Volga to the Dneiper, took place around 4500–4000 BC and could be an origin for M269. Wave 2 probably occurred from the Maykop culture (3700–3000 BC, indicated in cyan). The high variance and unusual M269 population found in this area (Hovhannisyan et al. 2014).

(6) M269→L23→L51

Hovhannisyan et al. (2014, fig. 6) notes a dissimilarity between the Ossetian, Azerbaijani, Turko–Armenian and European M269 populations. L51 does not clearly appear in the ancient DNA results itself. However, ancient DNA from the Yamnaya culture (modern day Russia and Ukraine) show that L23 and its subclade Z2013 dominate here. Additionally, FTDNA shows M269+ L23- and L23+ L51- tests are much more focussed on the Black Sea than L51+ tests. We interpret this as indicating L51 formed just after a migration started heading from the Black Sea towards western Europe.

(7) M269→L23→Z2013

L51 and its brother clade, Z2013, seem to have entered eastern Europe along with L51, but Z2013 does not seem to have participated much in the subsequent expansion west. PF7580 and its subclades are clearly concentrated in the Turkish highlands; CTS7822, particularly CTS9219, is closely associated with the Balkan peninsula; L277 is spread around eastern Europe; while CTS7763 may be Balkan or Anatolian. Meanwhile, ancient DNA from Haak et al. (2015) shows a very strong expansion of Z2013 throughout modern-day Russia. These data suggest Z2013 went north, south and east, while L51 went west.

(8) Caucasian populations

There are few cases where specific SNPs have been tested among people from the Caucasus, at least at FTDNA. Two M269 testers are L23→Z2013 and L23→L51→P310. Combined with the Hovhannisyan and ancient DNA (Haak et al.) results, we suggest they are probably mostly L23→Z2013.

(9) M269→L23→Z2013→PF7580 and the Hittites

The variance of PF7580 in Turkey and Syria suggests a coalescence age of 3000–5000 years. The Hittites are thought to have arrived in Anatolia before 2000 BC. On this basis, we ascribe the origin of PF7580 to a Hittite population.

(10) L51→PF7589: migration into Europe

L51 through to P311 represents an ~800-year gap in our knowledge. This may represent a hiatus in the east–west population movement which could be traced by historical cultures. This lack of structure makes it difficult to tell what went on in this ~800-year period.

Some information can be found from related clades. L51→PF7589 shares the European focus of L51→P311, but is not widely found in Germany. The median location of PF7589 in FTDNA tests is close to Salzburg, but the east–west migration means the split between P311 and PF7589 is likely to be further east. So although L51 may represent the population that launched into Europe, they perhaps (initially) did not get very far.

(11) The path into Europe: North or South?

The path our ancestors took into Europe isn't well defined. Travel in prehistoric Europe was predominantly by watercourses, driven by mountain ranges, dense forestation and marshland. Two main options are considered. Firstly, a path up the Danube river, which leads directly from the Black Sea to Germany, where P311 is first found in the archaeological DNA, and where there is the easternmost substantial concentration of P311 in the present population. This better with the distribution of M269+ P311– clades, which in Europe are strongest in countries bordering the Danube. However, it is not clear these were spread by the same migration as P311.

Secondly, through Poland or the Ukraine to the Baltic. This is the area through which the Corded Ware culture spread. The modern population in Corded Ware culture dominated areas is largely R1a, thus more obvious candidates to dominate the Corded Ware culture. A Baltic migration with a U106 origin in or near modern Denmark better fits with a north–south dichotomy in U106 clades (Z18 and Z9 in the north, Z156 and U198 in the south).

There are still very few archaeological samples on which to base a decision. The only Bronze-Age U106 result so far is in southern Sweden, and comes several centuries after the U106 origin. Taking P311 as a whole, several Bell Beaker sites have been found in the Rhine valley, again presumed to be several centuries after the formation of P311.

The path into Europe remains ill defined. On balance, my personal consideration is that the northern route has the better evidence.

(12) Early P311 and arrival in Germany

P311 splits into U106 and P312. This split probably occurred sometime during the march westwards. If our ancestors took a northern route, the lack of P311 in Poland (where R1a dominates) suggests it cannot have been further east than the Germano-Polish border. If our ancestors took a southern route, the easternmost likely place is Austria. The founding of P311 itself may have been slightly earlier, and considerably further east.

Either way, P311 seems to have been present in Germany before 2500 BC, around the time that the Corded Ware culture, and more specifically the Single Grave Culture, were setting up shop there… perhaps quite literally, given the Baltic amber trade. The Single Grave Culture is one point considered for the start of the Bell Beaker culture. Ancient DNA shows P311 played a significant part in the Bell Beaker culture in Germany, and the spread of the Bell Beakers may have been instrumental in spreading P311 throughout western Europe.

(13) P312 and U106

P312 is U106's bigger (though not necessarily older) brother. It's worth considering how P312 fared after the P312–U106 split. Early P312 is found at the Quedlinburg site in central Germany, and remains from its subclade, U152, have been found in south-eastern Germany. Both these individuals were buried in Bell Beaker culture fashion. Slightly younger results from Rathlin Island (N. Ireland) show that it spread westwards quickly.

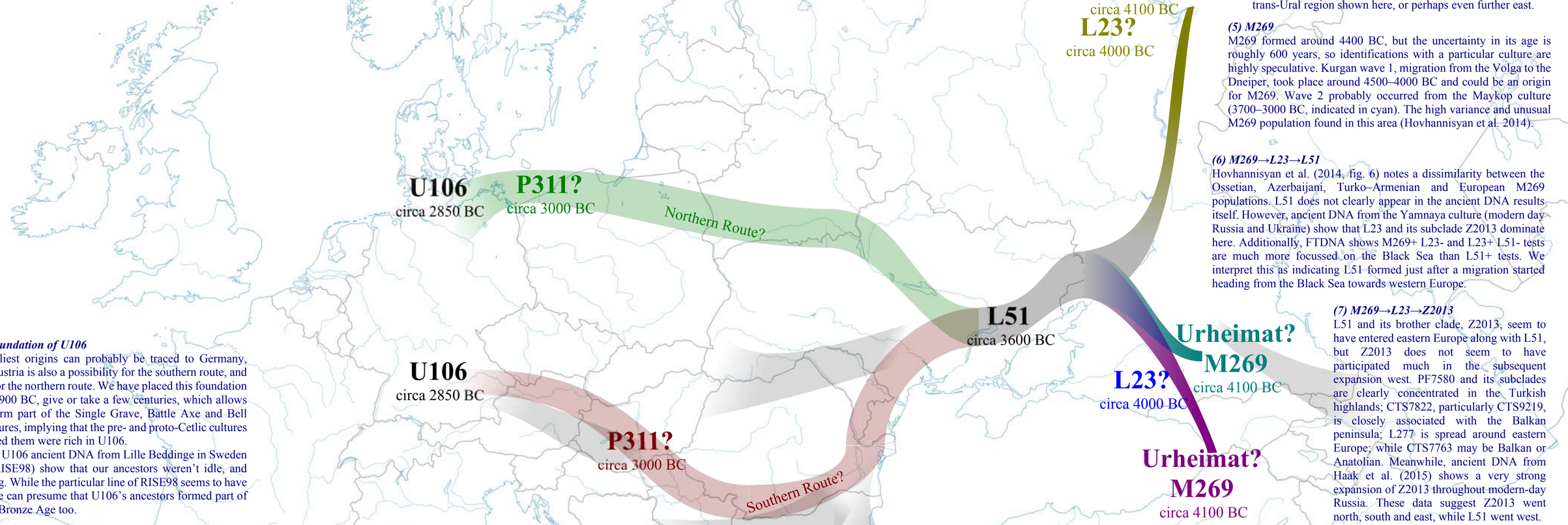
U106 now appears more commonly in Germany and Scandinavia. By contrast, P312 dominates in most of western Europe. U152 is found mostly in Switzerland and Italy, DF27 in Iberia, and L21 among Brythonic peoples. P312 therefore appears to have travelled south and west, while U106 mostly either stayed in Germany or moved north.

In the southern scenario, this may represent a northerly movement of U106 along the Rhine, while P312 went west. In the northern scenario, this would represent U106 initially staying in the Baltic region, while P312 continued around the North Sea coast.

(14) The foundation of U106

U106's earliest origins can probably be traced to Germany, although Austria is also a possibility for the southern route, and Denmark for the northern route. We have placed this foundation at around 2900 BC, give or take a few centuries, which allows U106 to form part of the Single Grave, Battle Axe and Bell Beaker cultures, implying that the pre- and proto-Cetlic cultures that followed them were rich in U106.

The U106 ancient DNA from Lille Beddinge in Sweden (skeleton RISE98) show that our ancestors weren't idle, and kept moving. While the particular line of RISE98 seems to have died out, we can presume that U106's ancestors formed part of the Nordic Bronze Age too.

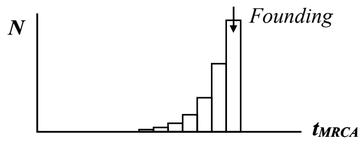


Migrations from STRs

MIGRATION PATHS FROM STRs

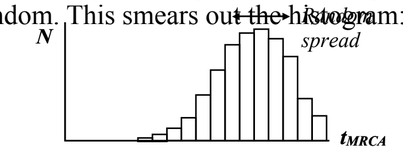
Migrations in recent times can be traced through histograms of times since the most-recent common ancestor (TMRCA) from STRs. This is an extension of the previous STR-dating method that can be used to disentangle geographies and migrations.

The principle works by measuring the TMRCA for every pair of men within a clade and comparing the distribution of people. In an iideal but growing population, where a man sires two sons, who each have two sons, who each have two sons, etc., the histogram will look like this:

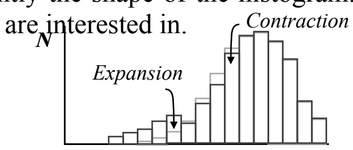


All men are related to the founding father. Half of men are related through one son, half through the other, so half of the table of TMRCAs will be a generation younger. Of each of those halves, half will be related another generation down, etc. So we end up with a histogram that halves with each generation, like the one above.

Generations aren't exactly the same length and the mutation process is random. This smears out the histogram:



In the real world, the expansion and contraction of populations occurs in response to external and internal events. This means that clumps form in the histogram during periods of population expansion, and gaps appear during population contraction. This modifies slightly the shape of the histogram. It is these bumps and voids that we are interested in.



These effects are subtle, and best illustrated through a real-world example. Here, we consider two examples:

`U106>Z381>Z156>DF98` ,

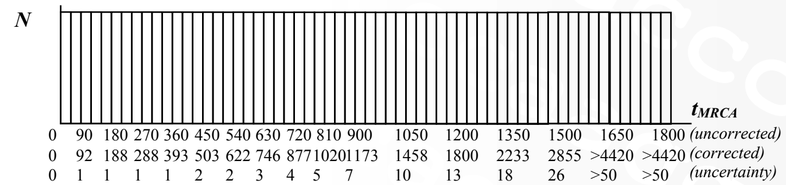
and

`U106>Z381>Z301>L48>Z9>Z30>Z2>Z7>Z8` .

These have very different backgrounds. DF98 concentrates in the Rhine valley and is known to have at least two Norman or Norman-era migrations. Z8 concentrates in the Low Countries, Germany and England and looks much more Germanic. We expect to see differences in their structure. First, however, we have to perform a calibration.

HISTOGRAM TMRCA CALIBRATION

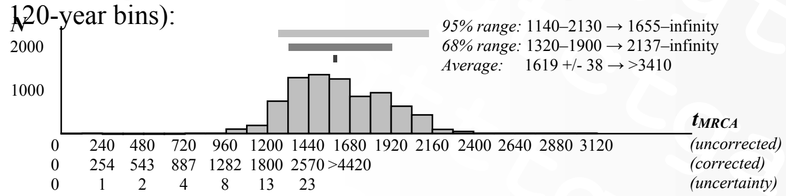
Earlier, we discussed how the STR data were calibrated against the SNP data for the infinite alleles method. Corrections to the infinite alleles method become significant after a few centuries, and the method becomes largely useless much after 2000 years ago. This means we expect the following translation of STR-based ages to reality:



Note how the correspondence is lost after ~3000 years as the ages tend to infinity as our correction function fails to fit. The uncertainty in this case is in the accuracy of our fitting function, and doesn't take random spread into account.

CHARACTERISING RANDOM SPREAD THROUGH INTER-CLADE TMRCA DISTRIBUTIONS

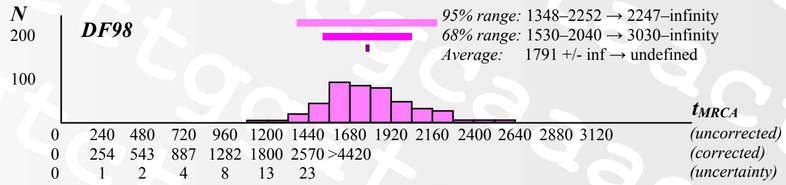
We can now take our example clades, DF98 and Z8, and measure their characteristic intrinsic spread. This spread is a function of testing depth (number of mutations tested) and age of population (number of mutations accumulated). DF98 and Z8 are last related by Z381, around 4400 years ago. Comparing the STR TMRCAs for DF98–Z8 pairs*, we arrive at the following histogram (binned into 120-year bins):



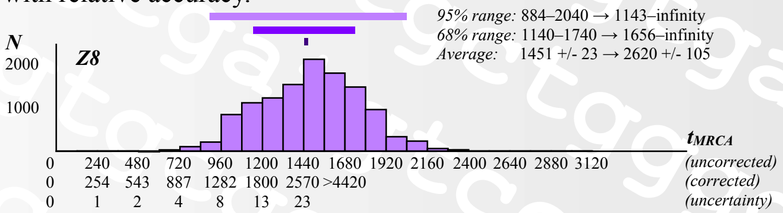
This histogram exemplifies the limitations of this approach. The true relationship age is around 4400 years ago for all pairs in this histogram. Three randomly sampled pairs from this histogram are most likely to have relations predicted to be close to 1320, 1619 and 1900 years ago (a typical uncertainty of 290 years). These ages would be corrected to circa 2137, >3410 and >4420 years. DF98 is predicted to be ~3600 years old, and Z8 to be ~2400 years old. Any migrations between the ~4400-year-old Z381 foundation and the ~3600-year-old DF98 foundation will be lost in this random spread. Migrations around the Z8 foundation might be recoverable, but only if they are very significant. The limitations of this method probably lie around 1000-2000 years ago, depending on the number of testers and scale of the migration.

(NB: Only Z8>Z334 tests were used in this analysis due to the large number of Z8 testers and the computational power required, which scales as $N_{testers}^2$).

The same analysis can be performed on DF98 and Z8 themselves, using their subclades, S1911 and S18823, and Z1 and Z11, respectively.



Despite the correction, the age of DF98 is not predicted: it is older than age the infinite alleles method is stable over. The spread of the histogram, however, has reduced from 290 years (uncorrected) for Z381 to 255 years (uncorrected) for DF98. This suggests that, at best, we can expect an accuracy of ~200 years in the dating of migrations within DF98. This is roughly what we would expect, as it is similar to the STR mutation rate (~1 per 140 years at 67 markers). Structure in the histogram younger than ~2000 years ago can probably be dated with relative accuracy.



In the case of Z8, the age is slightly over-predicted at ~2620 years instead of ~2400 years, but agrees well once the uncertainties are considered. Despite having a younger age, the spread of TMRCAs remains at ~300 years. Due to this spread, we can't use this method to understand any structure in Z8 before about 1300 years ago.

In these *inter*-clade histographs of DF98 and Z8, there is a nearly Gaussian ("normal" or "bell-curve") distribution of values with a characteristic spread. However, the distribution isn't quite Gaussian: e.g. Z8 has a 'bulge' around 1500 (corrected) years ago. These imperfections can reflect parallel or opposing mutations, typically from early in the history of that clade. In this case, it is due to a comparative lack of mutations in the Z1>Z344>14436052 and Z338>Z11>Z8175>…>FGC12059 clades. This will lead to artefacts in the *intra*-clade spreads that we will use to work out relationships.

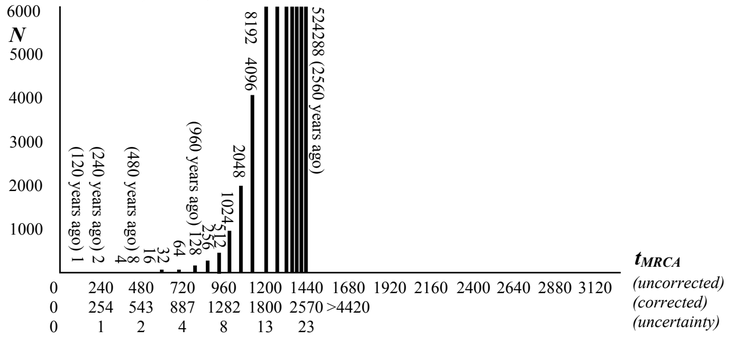
Experiments on several clades has shown the random spread can be characterised fairly consistently as ~200 years for young clades, rising to ~300 years for older clades. but varying between the two values, depending on the clade in question. We can therefore adopt a 200~300 year time period as the typical random spread in an uncalibrated TMRCA distribution. Other features are therefore likely to be due to internal structure.

The examples above show that we are limited to tracing migrations younger than 1000~2000 years, depending on the clade involved. Migrations older than this will be lost in the noise of the clade, but may still be recoverable using other methods like geographical distribution.

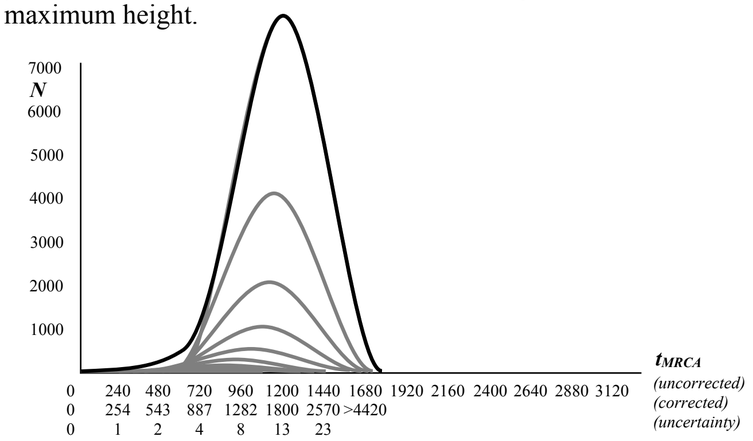
INTRA-CLADE HISTOGRAMS: EXPECTATIONS

Turning our attention to the *intra*-clade TMRCA histograms, we can form an expectation of what one should look like.

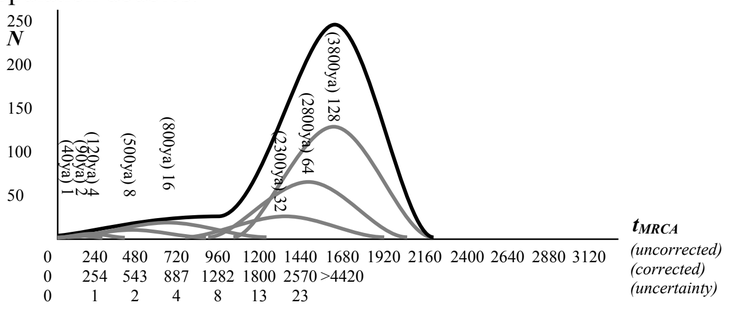
In the simplified population presented previously, we have a slowly growing population, with a new branch forming every 90-140 years or so, depending on where 67 or 111 markers are being tested (let's say 120 years). Half of the population goes into each branch, so half of the MRCAs are 140 years closer to the present than the other half. Of that closer half, half are another 140 years closer to the present, etc., so the histogram of TMRCAs halves every 140 years of real (corrected) time, following a $1/t^2$ law.



These will each be smeared out with a Gaussian of 200-300 years in width in *un*corrected time (width being the width at half the maximum height.



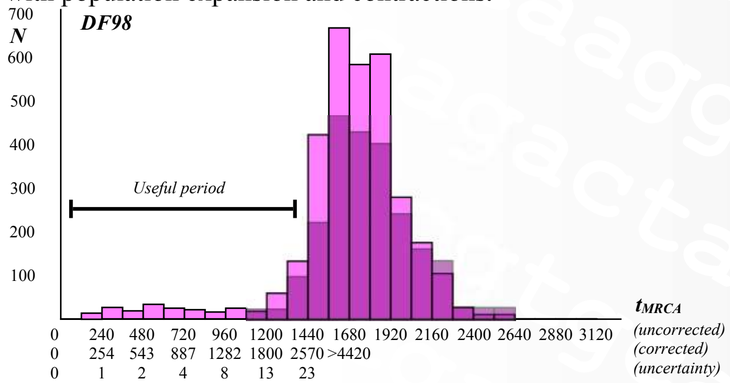
But in the real world, populations grow and shrink, so we can instead expect to find a new branch forming every time that population doubles.

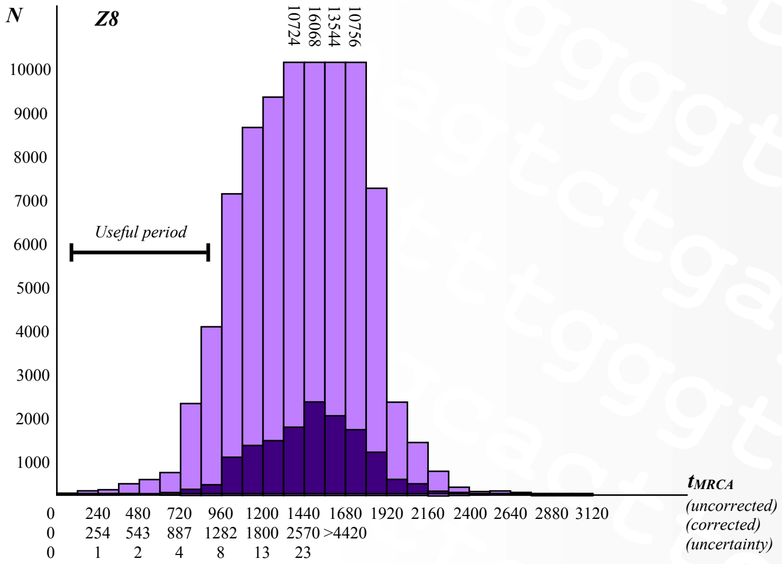


INTRA-CLADE EXAMPLES

In reality, things will be rather more complicated, as large branches are resiliant to population shrinkage, while small branches aren't, and branches occur in a random process, not a strictly timed fashion. But the basic structure is of a large peak when the population first formed, followed by a tail containing useful information about when that population grew in size.

We can now look at the real-world examples for Z8 and DF98 to see what they reveal. This should show us roughly what happened with population expansion and contractions.



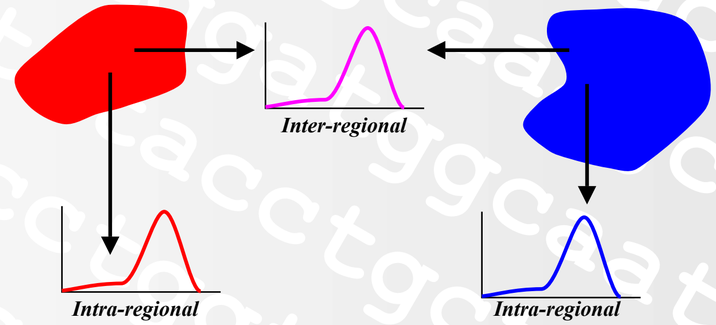


Both graphs show a strong peak, then a weak tail to modern times. DF98 has a long and markedly constant tail, showing continued and even accelerating population expansion for the last 1800 years. By contrast, the rapidly falling tail of Z8 shows that the modern growth of Z8 is at most at the background rate, and significant growth of Z8 above the general population probably ceased at least 900 years ago.

Separating this useful "tail" from the bulk of the TMRCAs is not easy, so some care must be taken about assuming hard evidence from such distributions.

COMPARING GEOGRAPHICAL REGIONS

The major advantage of this test comes when we start to split up these intra-clade TMRCAs by region to provide *inter-regional* and *intra-regional* TMRCA histograms for different regions or countries.



Peaks in an *intra-regional* TMRCA histogram indicate a growth of that clade in that country: e.g. a growth in England around 900 years ago could indicate the rapid growth from a Norman population. By contrast, peaks in an *inter-regional* TMRCA histogram indicate a migration: e.g. if there is a peak in the Anglo-French inter-regional histogram around 900 years ago, we can attest this to a migration between England and France (or vice versa) around that time.

Used together, these techniques can be used to trace migrations. For example, if no peak (or peak much older than 900 years) is seen in the French histogram, we can infer the direction of migration was from France to England. If a younger peak exists in the French histogram, we can infer migration from England to France.

The imprint of peaks in the population of origin should show up in the destination region too. For example, we could expect a peak in the inter-regional TMRCAs of Scotland and of Ireland around 900 years ago, as although the Norman influences from England didn't arrive there straight away, they still brought their Norman signatures with them when they came.

As seen in the previous example, these peaks are very hard to detect and can be very ambiguous. Treated with caution, and with careful examination of the underlying and associated evidence, they can prove useful in tracing past migrations.

VARIANCE AS AN ORIGIN INDICATOR

A final piece of evidence we can use is the position of the peak. The oldest population should have the oldest average TMRCA. Often this effect is hard to identify, but can be very useful where the "founder effect" exists: a slow-moving migration where one or a small number of people from a clade move into an area long after that clade has been founded. In these cases, the average TMRCA for that region may be considerable younger than the original population. The use of genetic variance as an indicator of origin is a well-known tool. However, stastistical spread can cause substantial differences on its own, so care must again be taken when using this method.

Ancestral STRs:

```
P311    13 24 14 11 11-14 12 12 12 13 13 29 17 9-10 11 11 25 15 19 29 15-15-17-17 11 11 19-23 16 15 17 17 37-38 12 12   11 9 15-16 8 10 10 8 10 10 12 23-23 16 10 12 12 15 8 12 22 20 13 12 11 13 11 11 12 12   35 15 9 16 12 26 26 19 12 11 13 12 10 9 12 12 10 11 11 30 12 13 24 13 10 10 20 15 19 13 24 17 12 15 24 12 23 18 10 14 17 9 12 11
U106    13 24 14 11 11-14 12 12 12 13 13 29 17 9-10 11 11 25 15 19 29 15-15-17-17 11 11 19-23 16 15 17 17 37-38 12 12   11 9 15-16 8 10 10 8 10 10 12 23-23 16 10 12 12 15 8 12 22 20 13 12 11 13 11 11 13 12   35 15 9 16 12 26 26 19 12 11 13 12 10 9 12 12 10 11 11 30 12 13 24 13 10 10 20 15 19 13 24 17 12 15 24 12 23 18 10 14 17 9 12 11
```

DYS492=13 is derived at the U106 level. **PPV:** An M269+ test with this result is 98.6% likely to be U106. **Accuracy:** Splits M269+ U106+ from M269+ U106- with 97.2% balanced accuracy.
DYS390=24 is ancestral, but 23 is modal for U106. **PPV:** An M269+ test with 23 is 84% likely to be U106. **Accuracy:** Splits M269+ U106+ from M269+ U106- with 73% balanced accuracy.
DYS522=10 is ancestral, but 11 is common in P312. **PPV:** An M269+ test with 10 is 65% likely to be U106. **Accuracy:** Splits M269+ U106+ from M269+ U106- with 76% balanced accuracy.
M269+, DYS492=13 and DYS390=23 together give 98.8% likelihood of being U106 [PPV].
M269+, DYS492=13 and DYS522=10 together give 99.9% likelihood of being U106 [PPV].
M269+, DYS492=13, DYS390=23 and DYS522=10 together give 99.4% likelihood of being U106 [PPV].
NB: DYS492=14 is more commonly found in P311>P312>U152 (PPV for U106: 43%). However, M269+, DYS492=14 and DYS390=23 together give 84% likelihood of being U106.

Factsheet: U106

Dr. Iain McDonald on behalf of the U106/S21 group
Compiled: 17 Feb 2016 based on data from 09 Feb 2016

ISOGG: **R1b1a1a2a1a1 [2015]**

Parent: **P311 (S128) / L11 (S127)**

Alternative (equivalent) SNPs: **U106 (S21, M405)**

Major subclades: **Z381** **Z18** FGC3861 S12025 S18632 A2147 FGC396

Likely origin: Germany, 2900 BC
Possible earliest association(s):
Corded Ware Culture, Single Grave Culture
Protruding-Foot Beaker Culture, Bell Beaker Culture
Primary regions:
Germany, Low Countries, north/east France, south/east England
Compared to parent, less common in:
Iberia, Italy, Scotland, Ireland, Wales

Number of testers at 67/111 markers (N) = 2612/1321
Mean TMRCA from infinite alleles (μ) = 1970/1955 years (uncorrected)
Standard deviation among TMRCAs (s) = 321/235 years (uncorrected)

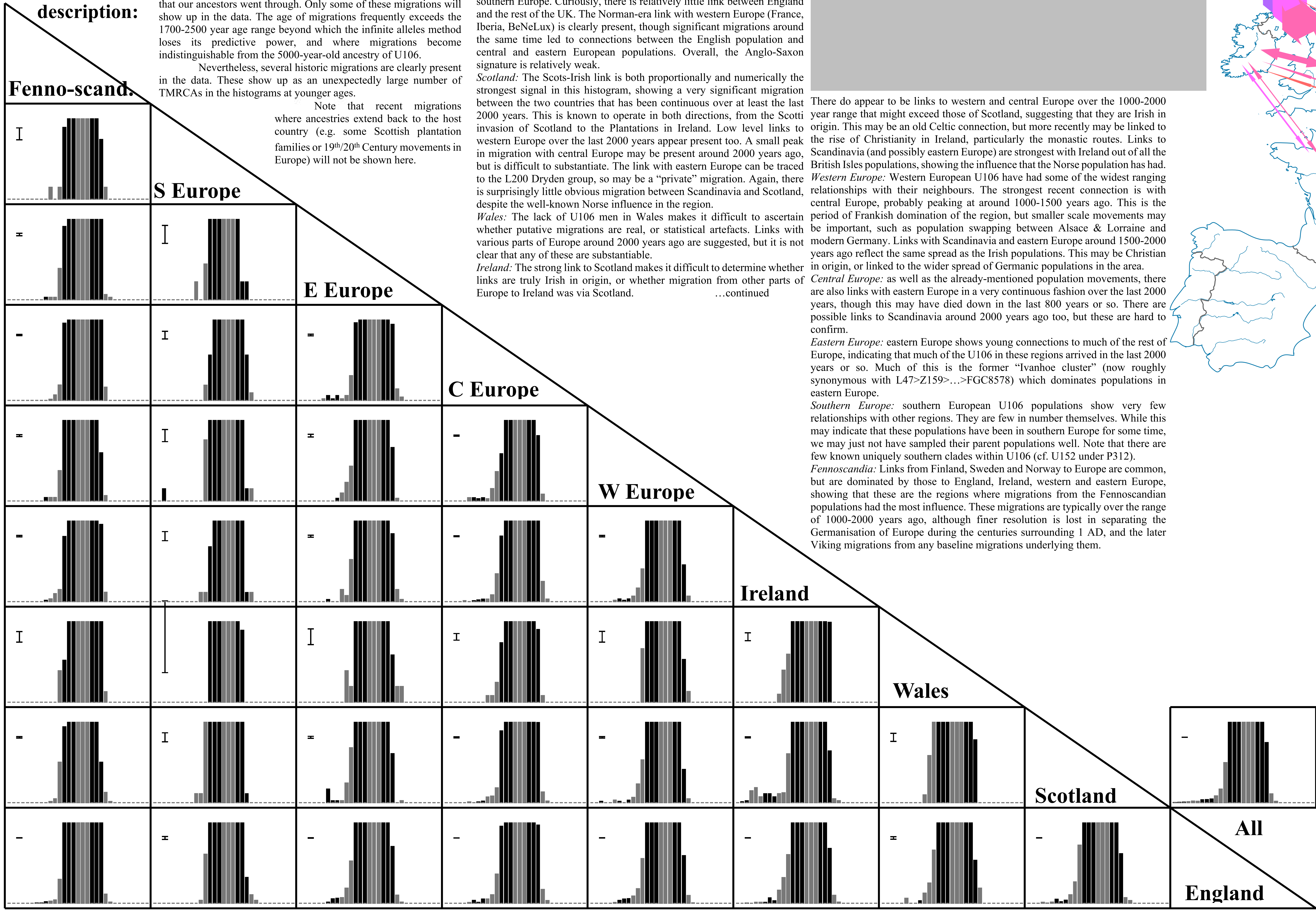
Infinite alleles TMRCAs

Histogram description:

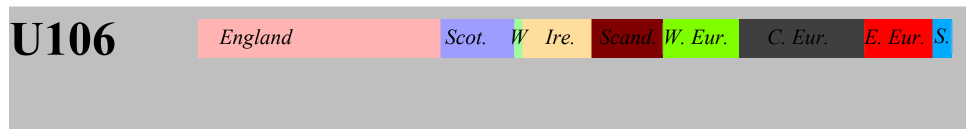
At this highest level, the TMRCA histograms show all the migrations that our ancestors went through. Only some of these migrations will show up in the data. The age of migrations frequently exceeds the 1700-2500 year age range beyond which the infinite alleles method loses its predictive power, and where migrations become indistinguishable from the 5000-year-old ancestry of U106.

Nevertheless, several historic migrations are clearly present in the data. These show up as an unexpectedly large number of TMRCAs in the histograms at younger ages.

Note that recent migrations where ancestries extend back to the host country (e.g. some Scottish plantation families or 19th/20th Century movements in Europe) will not be shown here.

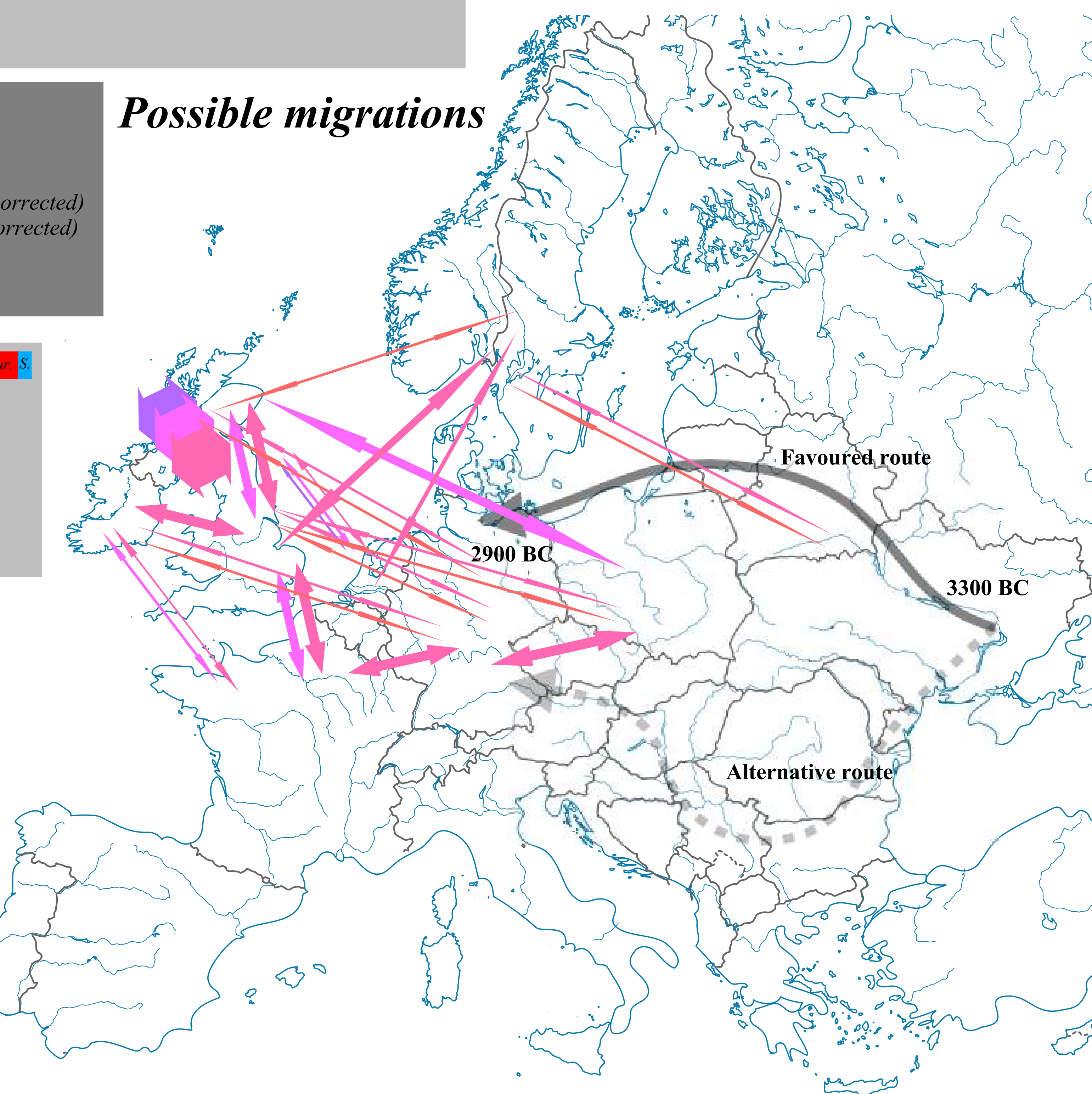


England: There are significant relationships between England and all other parts of Europe except southern Europe and perhaps Fenno-Scandia (Norway, Sweden, Finland). Norse Vikings therefore left little mark in England, while English Crusaders do not appear responsible for U106 in southern Europe. Curiously, there is relatively little link between England and the rest of the UK. The Norman-era link with western Europe (France, Iberia, BeNeLux) is clearly present, though significant migrations around the same time led to connections between the English population and central and eastern European populations. Overall, the Anglo-Saxon signature is relatively weak.
Scotland: The Scots-Irish link is both proportionally and numerically the strongest signal in this histogram, showing a very significant migration between the two countries that has been continuous over at least the last 2000 years. This is known to operate in both directions, from the Scotti invasion of Scotland to the Plantations in Ireland. Low level links to western Europe over the last 2000 years appear present too. A small peak in migration with central Europe may be present around 2000 years ago, but is difficult to substantiate. The link with eastern Europe can be traced to the L200 Dryden group, so may be a "private" migration. Again, there is surprisingly little obvious migration between Scandinavia and Scotland, despite the well-known Norse influence in the region.
Wales: The lack of U106 men in Wales makes it difficult to ascertain whether putative migrations are real, or statistical artefacts. Links with various parts of Europe around 2000 years ago are suggested, but it is not clear that any of these are substantiable.
Ireland: The strong link to Scotland makes it difficult to determine whether links are truly Irish in origin, or whether migration from other parts of Europe to Ireland was via Scotland. …continued



There do appear to be links to western and central Europe over the 1000-2000 year range that might exceed those of Scotland, suggesting that they are Irish in origin. This may be an old Celtic connection, but more recently may be linked to the rise of Christianity in Ireland, particularly the monastic routes. Links to Scandinavia (and possibly eastern Europe) are strongest with Ireland out of all the British Isles populations, showing the influence that the Norse population has had.
Western Europe: Western European U106 have had some of the widest ranging relationships with their neighbours. The strongest recent connection is with central Europe, probably peaking at around 1000-1500 years ago. This is the period of Frankish domination of the region, but smaller scale movements may be important, such as population swapping between Alsace & Lorraine and modern Germany. Links with Scandinavia and eastern Europe around 1500-2000 years ago reflect the same spread as the Irish populations. This may be Christian in origin, or linked to the wider spread of Germanic populations in the area.
Central Europe: as well as the already-mentioned population movements, there are also links with eastern Europe in a very continuous fashion over the last 2000 years, though this may have died down in the last 800 years or so. There are possible links to Scandinavia around 2000 years ago too, but these are hard to confirm.
Eastern Europe: eastern Europe shows young connections to much of the rest of Europe, indicating that much of the U106 in these regions arrived in the last 2000 years or so. Much of this is the former "Ivanhoe cluster" (now roughly synonymous with L47>Z159>…>FGC8578) which dominates populations in eastern Europe.
Southern Europe: southern European U106 populations show very few relationships with other regions. They are few in number themselves. While this may indicate that these populations have been in southern Europe for some time, we may just not have sampled their parent populations well. Note that there are few known uniquely southern clades within U106 (cf. U152 under P312).
Fennoscandia: Links from Finland, Sweden and Norway to Europe are common, but are dominated by those to England, Ireland, western and central Europe, showing that these are the regions where migrations from the Fennoscandian populations had the most influence. These migrations are typically over the range of 1000-2000 years ago, although finer resolution is lost in separating the Germanisation of Europe during the centuries surrounding 1 AD, and the later Viking migrations from any baseline migrations underlying them.

Possible migrations



About this map

This map represents the possible place of origin of the common ancestor of men of this clade, and the route that their ancestors took to get there. It also shows some of the migrations that their descendents seem to have been involved in. ***Do not take this map as proven fact. Do not even take this map as a probable scenario.*** It is only a guess at which is going on and what is important. It is one interpretation of a limited set of data. There are many other possibilities.

Migrations are coded into these diagrams as a series of arrows. Grey arrows denote mutations that have happened prior to the SNP. Coloured arrows denote migrations in specific time periods, ranging from blue (last 500 years) to red (2000 years ago or older). Note that, in general, migrations older than about 1700 years quickly become difficult to identify, even when only 111-marker results are taken into account.

A note on histograms:

The series of histograms shows the time to most-recent common ancestor, as calculated by the infinite alleles method using McGee's tool with its standard setup parameters.

Each vertical bar on the histogram shows people related during a 120-year period, according to the tool. The first bar is therefore 0-120 years ago, the second 120-240 years, etc. Series of three bars are grouped into alternate light and dark bands of 360 years to aid counting: the first is 0-360 years, the second 360-720 years, etc.

Corrections need to be applied to these ages in order to adjust them for back mutations and for asymmetric/non-linear effects in the mutation direction. These corrections are tabulated in the accompanying table.

Mutations are also random, which will cause the ages to spread out. The typical spread is calculated by fitting a Gaussian profile to the resulting histogram. The resulting spread (σ) will give the typical uncertainty in each age, specifically the positive and negative spread within which about 2/3 of points of that age will fall.

The top of each histogram is clipped to show the relevant detail at low levels.

TMRCA correction factors

Age: Raw	Age: Corr.	Date	Uncertainty
0	0	1950 AD	+/- 0 years
120	123	1827 AD	+/- 0 years
240	254	1696 AD	+/- 1 years
360	393	1557 AD	+/- 2 years
480	543	1407 AD	+/- 2 years
600	703	1247 AD	+/- 5 years
720	877	1073 AD	+/- 7 years
840	1069	881 AD	+/- 10 years
960	1282	668 AD	+/- 14 years
1080	1522	428 AD	+/- 19 years
1200	1800	150 AD	+/- 25 years
1320	2136	188 BC	+/- 33 years
1440	2570	622 BC	+/- 43 years
1560	3231	1283 BC	+/- 59 years
1680	>4420	>2372 BC	(undefined)

Corresponding ranges
Black: 0-360 years → 1557 AD onwards
Grey: 360-720 years → 1073 – 1557 AD
Black: 720-1080 years → 428 – 1073 AD
Grey: 1080-1440 years → 622 BC – 428 AD
Black: 1440-1800 years → before 622 BC