Autosomal DNA Tutorial

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Autosomal DNA

Autosomal DNA is a term used in genetic genealogy to describe DNA that is inherited from the autosomal chromosomes. An autosome is any of the numbered chromosomes, as opposed to the sex chromosomes. Humans have 22 pairs of autosomes and one pair of sex chromosomes (the X chromosome and the Y chromosome).

Autosomes are numbered roughly in relation to their sizes. That is, chromosome 1 has approximately 2,800 genes, while chromosome 22 has only about 750 genes. Autosomal DNA tests can be used to confirm with a high level of accuracy most close family relationships: parent-child, sibling, cousin, and second cousin. Even for these relationships, additional contextual and genealogical information is required to confirm the nature of the relationship.

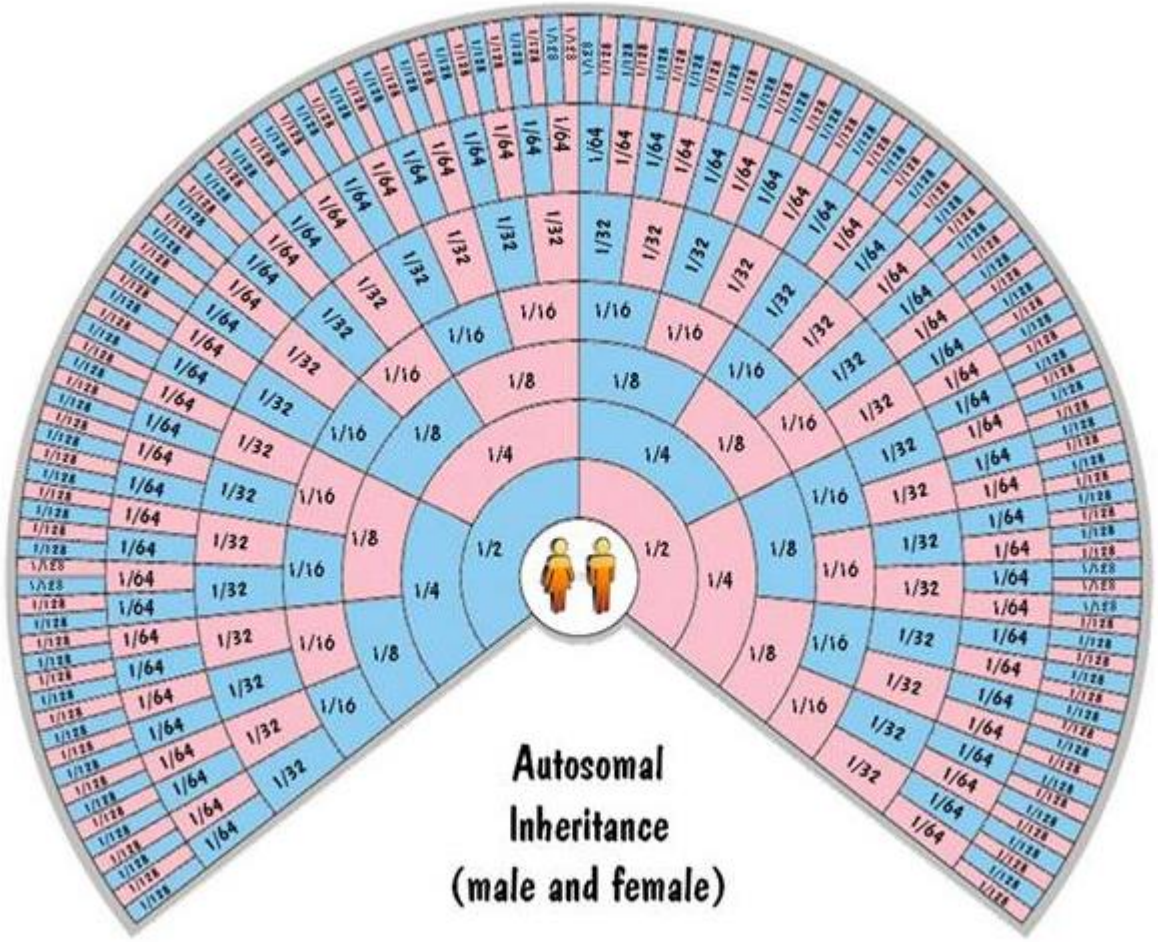
Autosomal DNA

For more distant genealogical relationships, from second cousins once removed to 5th cousins (having common g-g-g-g-grandparents), a more careful approach is necessary, and data needs to be collected from multiple family members. For relationships at the 4th cousin-once-removed to 5th cousin level, you may need to test 10 to 20 or more first and second cousins and see how much autosomal DNA they share with a potential 4th cousin once removed or a potential 5th cousin in order to have sufficient data to generate a statistically significant average amount of autosomal DNA
that is shared among the entire group.

Autosomal DNA Inheritance Chart

This chart shows how your Autosomal DNA dets diluted over time limiting its accuracy to about 5 generations.

Basically you get 50% of your DNA from your parents, 25 % from your grandparents, 12.5% from your great-grandparents, 6.25% from your great-great-grandparents and so on. Fairly soon you will have approximately zero DNA.



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Genealogical relationships beyond the 5th cousin level are more difficult to prove with autosomal DNA testing and, as a rule, these can only be approached using triangulation. In some cases Y-DNA and mtDNA data may also be of help.

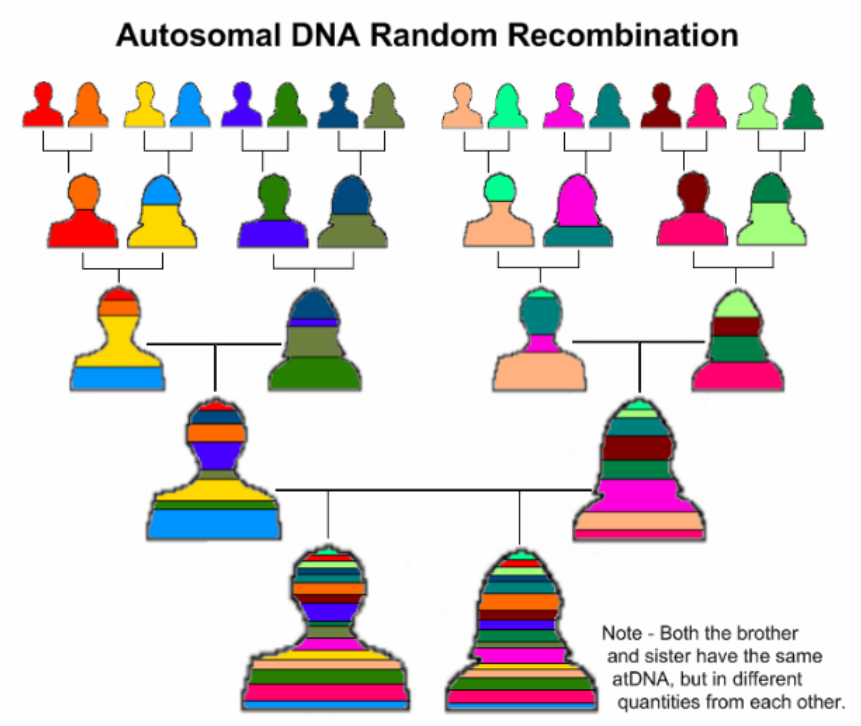
For autosomal DNA testing one should always test the oldest generations first wherever possible - your parents, grandparents (if you are lucky), aunts and uncles. By testing yourself as well as your parents you will be able to determine which segments have been inherited from which parent, and you will also be able to rule out coincidental matches.

If you only have one parent available for testing then you should test you and your parent. You should also test your siblings because they will inherit part of your parents' DNA that you don't carry. Your uncles and aunts will inherit part of your grandparents' DNA that your parents don't have.

Autosomal DNA

The next priority should be to test other close relatives from first to third cousins to get the best representation across your ancestry. Experience suggests that the maximum return is obtained by testing second cousins. They share one set of great grandparents so when someone matches you and a second cousin you get to narrow down your matches to that specific line.

Recombination



Recombination is the exchange of DNA segments between the two copies of a chromosome (maternally inherited and paternally inherited). This occurs during the creation of an egg or sperm for the next generation. Each chromosome is cut at the same location, usually at one or two random spots (crossover points), and DNA segments are swapped between the two copies.

The newly formed chromosome in the egg will be a patchwork of contributions from the maternal grandparents, and the newly formed chromosome in the sperm will be a patchwork of contributions from the paternal grandparents, but there is still one copy of every gene. Some eggs and sperm will retain a complete copy of one grandparent's chromosome without recombination. The creation of each egg and sperm is an independent event, so siblings will inherit different portions of their grandparents' DNA.

Males and females have different rates of recombination. Males average about 27 crossovers per child and females average about 41 crossovers per child.

Triangulation

In autosomal DNA testing the term triangulation is most commonly used to describe the process of reviewing the pedigree charts of clusters of shared matches/in common with matches in order to identify a common ancestor or ancestral couple. This process is sometimes also known as tree triangulation.

Accurate triangulation requires multiple test-takers who all have deep and accurate family trees. Even when these conditions are met, not all real cousins will triangulate due to random recombination and inheritance factors. We can never forget the random factors affecting DNA inheritance. That randomness may result in some techniques working for some people and not others. Where it works, triangulation is a valid technique.

Triangulation

Autosomal DNA triangulation works on the same principles as triangulation for YDNA and mtDNA. We start with the known and work back to the unknown, and we combine DNA evidence with sound genealogical evidence to draw a conclusion. For autosomal DNA we are looking at specific segments of DNA and trying to determine the ancestor or ancestral couple from whom we inherited that DNA. For this process to work we need relatives who are closely related to us with known genealogies.

The technique can also be used with third, fourth and fifth cousins but it is important that both parties have sound genealogies and can trace their ancestors on all their family lines for the appropriate number of generations in order to rule out the possibility of a relationship on a different pathway. The assignment of segments to fourth and fifth cousins is more secure if the match can also be triangulated with other close family members (e.g., a parent, an aunt or uncle, a first or a second cousin).

Chances of a Match

Three testing companies have provided percentages showing the chances of matching a known cousin at the differing degrees of relationship. I've compiled the statistics into the table below.

Relationship	23andMe (unphased)	Family Finder (unphased)	AncestryDNA (phased)
2nd cousin	> 99%	> 99%	100%
3rd cousin	~ 90%	> 90%	98%
4th cousin	~ 45%	> 50%	71%
5th cousin	~ 15%	> 10%	32%
6th cousin or more distant	< 5%	Remote (typically less than 2%)	11%

AncestryDNA phases the genotypes before doing the matching process. (Phasing is the process of assigning alleles to the maternal and paternal chromosomes.

Phasing

We have seen how autosomal DNA triangulation can be a very useful tool when DNA evidence is combined with sound genealogical research to draw conclusions about close relationships up to about the fourth or fifth cousin level.

It would be similarly extremely unlikely for three or more fifth, sixth or more distant cousins all to match on the same segment through IBD descent from a specific ancestor. This of course assumes that they have inherited enough DNA from their mutual ancestor to show up as a match at all.

Phasing is the process of sorting out the DNA letters we receive from our parents and assigning them to the maternal and paternal chromosomes. Our autosomal chromosomes come in pairs. We receive one set of 22 autosomes from our mom and another set from our dad.

Phasing

Phasing matters most with the smaller segments under 15 cMs where there is a law of diminishing returns. As the segments get smaller the chances that the segments will be false positive pseudosegments (mishmashes of As, Cs, Ts and Gs from both the maternal and paternal chromosome) will tend to increase. Independent research from genetic genealogists suggests that 15 cM is the threshold where segments can be assumed to be IBD with reasonable confidence, whereas only 42% of 7 cM segments are likely to be IBD. Even when phasing is done there is still the possibility of false matches with the smaller segments. A study by Durand et al (2014) found that over 67% of phased 2-4 cM segments were false positives (matches found in the child but not in the parents).

Autosomal DNA testing for genetic genealogy is still very much in its infancy, and we clearly have a lot to learn about the interpretation of results.