

MANUAL

GENETIC AFFAIRS



Welcome to Genetic Affairs

Unlock the power of your DNA with our advanced genetic analysis and family discovery tools.



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Introduction

This document describes the use of the website <u>https://www.geneticaffairs.com</u> and more specifically, the use of the member section which can be reached under <u>https://members.geneticaffairs.com</u>

The original purpose of Genetic Affairs was the automation of the retrieval of genetic matches from two major DNA testing companies: 23andme and FamilyTreeDNA (FTDNA). This feature, also known as **AutoScan**, allows users to register their DNA accounts and provides an interface to select which profiles updates should be provided, how often and which criteria should be applied.

Nowadays, the most important feature of Genetic Affairs is the automated clustering of shared matches, named **AutoCluster**. AutoCluster organizes your matches into shared match clusters that likely represent branches of your family.

The AutoTree feature identifies common ancestors and reconstructed trees for FTDNA profiles.

In addition to the shared match clustering offered by **AutoCluster**, a cluster approach is available that is based on overlapping segments. This clustering is available under the name **AutoSegment**. Another version of this tool, **hybrid AutoSegment** is available and allows for the clustering of MyHeritage, 23andme, FTDNA and GEDmatch data based on overlapping segments.

Due to recent changes in 23andMe's security protocols, Genetic Affairs is currently unable to support analyses involving 23andMe data. The enhanced security measures prevent us from accessing and processing data as previously done. We apologize for the inconvenience and are monitoring any updates in 23andMe's policies that might allow us to reinstate these features in the future.

Features of Genetic Affairs

The next sections will describe in more detail the various features of Genetic Affairs.

Genetic Affairs **AutoCluster**tm provides the following features:

- Automatic clustering of shared matches using adjustable cM ranges
- Enriched surnames/locations in the clusters of AutoCluster for 23andme profiles
- Adjustable cM limits for the largest segment for FTDNA analyses
- Adjustable cM limits for shared cM between shared matches for 23andme analyses
- Ability to perform AutoClustering using common matches from 23andme that share a segment (and which will most likely triangulate).
- Extended clustering where the shared matches from the initial analysis (given a cM range or starred matches or groups) are used as matches.
- Segment data for FTDNA and 23andme analyses as well as a chromosome browser overview.

Genetic Affairs **AutoFastClust** provides the following features:

- Instant clustering of shared matches using adjustable cM ranges
- User entered match and shared match data, saved in local storage or CSV files

Genetic Affairs **AutoTreetm** provides the following features for **FTDNA** profiles:

- Identification of common ancestors from trees of users by employing a three-step clustering
 - First, a surname clustering is performed
 - o Second, a first name clustering is employed on the surname clusters
 - Finally, using birth and death year information, the final common ancestor clusters are determined
- Automatic reconstruction and visualization of a genealogical tree using the identified common ancestors and DNA matches
- Works for adoptees and profiles with an attached tree
- Identification of common locations using a distance clustering of birth locations of tree persons

Genetic Affairs **AutoPedigree**tm provides the following features for **FTDNA** profiles:

- automates the generation and testing of hypotheses using reconstructed trees from AutoTree.
- developed to identify how a person, for instance an adoptee, fits into a reconstructed AutoTree

Genetic Affairs **AutoSegment**tm provides the following features:

- perform a DNA segment-based clustering method that is available for segment data from MyHeritage, 23andme, FamilyTreeDNA and GEDmatch
- Requires segment data downloaded from testing companies
- Does not require credential information
- Allows for filtering of segments located in known pile-up regions.
- Allows for easy integration into the DNA Painter website

Genetic Affairs **Hybrid AutoSegment**tm provides the following features:

- perform a DNA segment-based clustering method combining segment data from MyHeritage, 23andme, FamilyTreeDNA and GEDmatch into a single analysis
- Requires segment data downloaded from testing companies
- Does not require credential information
- Allows for filtering of segments located in known pile-up regions.
- Allows for easy integration into the DNA Painter website
- Allows for liftover procedure to improve segment locations for FTDNA data

AutoKinship

- Relationship estimation: Provides kinship estimates by analyzing shared DNA among matches, helping users understand relationships based on genetic closeness.
- Visual Kinship Chart: Generates an interactive kinship chart to visually illustrate connections between matches, showing how individuals are related within a family tree context.

AutoLineage

• AutoLineage is designed to help your genetic genealogy research by helping you organize and analyze your DNA matches and trees more effectively.

Last, it is possible to recluster old AutoCluster or MyHeritage AutoCluster analyses, or convert them into Excel files.

Registration

The registration page (<u>https://members.geneticaffairs.com/register</u>) shows information that is required for registration at the site (see Figure 1).

Register		
Name		
E-Mail Address		
Password	۹	
Confirm Password	(P)	
	I Accept The Terms Of Service, our Privacy Policy	
	\square I would like to subscribe to the newsletter	
	♥ REGISTER	

Figure 1. Registration page of the website <u>https://members.geneticaffairs.com/register</u>

After registration, the user is redirected to the landing page (see Figure 2). The landing page offers several options. The "show websites" button shows all registered 23andme and FTDNA websites. By clicking on the "Register a new website", users can register new FTDNA or 23andme websites, using your login credentials. This step is required for the regular AutoCluster analyses.

The "Run AutoCluster" will show all registered 23andme and FTDNA websites. The "Run AutoTree" button will display all registered FTDNA websites since the AutoTree feature is only available for FTDNA profiles.

The "Run AutoSegment" option will show another page with four options, an AutoSegment analysis for MyHeritage, 23andme, FTDNA or GEDmatch. The "Run hybrid AutoSegment" link will allow users to run a combined analysis using data from the aforementioned companies.

The "Run AutoCluster using CSV files" feature allows users to run a clustering analysis using two CSV files, one containing match data and one containing shared matches data. A similar feature is provided by the "Run online AutoFastCluster" where these datasets can be entered manually. The "Recluster MyHeritage AutoClusters" option allows you to recluster old AutoCluster or MyHeritage cluster results.

The "User settings" option will show the different options available for an user. Archived newsletters are available using the "Newsletter archive" button.

The "Show credits" button displays the amount of available credits. Credits can be acquired using the "Subscription" option.



Figure 2. Landing page after registration.

Adding a website

Upon clicking on the link: "Register a new website", the user is redirected to a page that allows adding of website credentials (see Figure 3).



Figure 3 - Landing page for adding a new website

When the "Add FamilyTreeDNA" button is selected, a login page is displayed that will allow the entry of the website credentials (see Figure 4 for an example of FTDNA)

Add new website								
Company	FamilyTreeDNA							
Login:		E3						
Password:	******	٩						
Retype password:	*****	P						
ADD NEW WEBSITE								

Figure 4. Website to add FTDNA credentials.

Upon entering the website credentials, a confirmation message will appear after pressing the "Add new website" (see Figure 5). Note that for FTDNA profiles the KIT id is required for the login field.

	Add new website	
Company	FamilyTreeDNA	
Login:	johndoe@gmail.com	
Password:		Ð
Retype password:		Ð
ADD NEW WEBSITE	i Add new FamilyTreeDNA account? After pressing yes, we will check the credentials which will take some time. Yes, add website No, cancel	

Figure 5. Information pane that will appear after pressing the "Add new website" button.

After the "Yes add website" is selected, Genetic Affairs will use the supplied user credentials to check if these are valid. During this verification, a message is displayed (see Figure 6). See our online FAQ section (<u>https://www.geneticaffairs.com/faq.html</u>) for more information concerning the privacy of your login credentials.

	Add nou un brita									
	Add new website									
Company	23andme									
Login:	@gmail.com									
Password:										
Retype password:										
ADD NEW WEBSITE	Login credentials are currently being checked Please set the update interval after the website has been added									

Figure 6. The message is displayed while your login credentials are verified.

Note that browsers sometimes try to fill in credential data in the login fields. If entering credential information is not successful on multiple occasions, please try using another browser.

The websites page will be displayed if the credentials are successfully tested or if the websites button is selected (see Figure 7).



Figure 7. Website button on the main page

Websites							
Company: 23andme	e	Login: I.com	Profiles:	Edit	Delete		
	e	l.com		Ũ	â		
23andme	e.	මgmail.com	i		â		
FamilyTreeDNA		SHOW 4 FA	MILYTREEDNA I	PROFILES			
		ADD NEW WEBSIT	E				

The websites page shows the registered FTDNA and 23andme websites (see Figure 8).

Figure 8. The main view of all the websites. FTDNA websites are grouped on a different page since they are linked to a single profile for each login.

The red button behind the 23andme websites will delete the website credentials as well as the underlying profiles and updates. The edit button can be used to change the login credentials of the website, for instance after you have changed its password.

The Profiles button will bring up the profiles for a 23andme website (for FTDNA, the website view will display a profile since each FTDNA website is linked to a single profile). The Profile view and their settings will be discussed in the next section.

Note that by default for FTDNA profiles the **KIT** identifier is shown. If multiple kits are managed, tracking the different can become difficult. It is now possible to rename the profile name to something that is easier to remember.

Profiles – starting an analysis

After clicking on the "Profiles" button for a certain website (see Figure 8) all profiles are displayed that are linked to that website login (see Figure 9). Note that there can be several profiles linked to a single 23andme website but only one FTDNA profile linked to a registered FTDNA website.

Fam	nilyTreeD	NA logi	in: Ev	/ert-Jan			
Α	В	CBI	D ^D	E	F	G	н
Profile:	Mails	Hide	Default AutoCluster	AutoTree AutoCluster	Rule based AutoCluster	AutoScan	Delete
Evert-Jan Blom	\leq		0		١₹	0	ā

Several analysis options are available which will be discussed below.

Figure 9. Profiles view for a FTDNA website that allows the selection of a specific analysis.

- A) Shows the name that is linked to the profile. This name can be edited, for instance, to replace the FTDNA kit id number to the actual profile name.
- B) When updates are available this icon turns green, when selected the latest updates are shown.
- C) Hide profile (only for 23andme profiles). When selected another unhide button will appear.
- D) AutoCluster analysis. Opens a new page to perform an AutoCluster analysis.
- E) AutoTree analysis. Opens an AutoCluster analysis page with some options already enabled to perform an AutoTree analysis (see AutoTree).
- F) Delete a profile. This will also delete all linked messages

AutoCluster analysis

AutoCluster (option D in Figure 9) organizes your DNA matches for registered 23andme or FTDNA profiles into shared match clusters that likely represent branches of your family. In the visualization of this analysis, each of the colored cells represents an intersection between two of your matches, meaning, they both match you and each other (see Figure 10). These cells, in turn, are grouped together both physically and by color to create a powerful visual chart of your shared matches clusters



Figure 10. Clustering analysis of shared matches analysis using AutoCluster using a cM range of 600 cM - 50 cM. Various clusters have been identified which have been annotated using genealogical data (M = Maternal, P = Paternal).

Each color represents one shared match cluster. Members of a cluster match you and most or all of the other cluster members. Everyone in a cluster will likely be on the same ancestral line, although the MRCA between any of the matches and between you and any match may vary. The generational level of the clusters may vary as well. One may be your paternal grandmother's branch; another maybe your paternal grandfather's father's branch (see some genealogical annotations in the AutoCluster example in Figure 10).

You may see several gray cells that do not belong to any color-grouped cluster. They usually represent a shared match where one of the two cousins is too closely related to you to belong to just one cluster. Each of these cousins belongs to a color-grouped cluster, the gray cell indicates that one of them belongs in both clusters. In addition, the clusters are sorted based on the gray cells, these sorted clusters sometimes fall into larger cluster structures that are easy to identify in the visualization (see Figure 11).



Figure 11. Example of the sorting of clusters based on the grey cells between clusters (picture provided by Robert Randolph).

Underneath the graphical representation of the clusters, some information concerning the AutoCluster is placed with respect to the employed settings as well as a searchable and sortable table (see Figure 12).

	Α	B	С	D	E	F	G
	Name Search	▲ cM ▲ M	# ▲ Max	Cluster Search fc	Tree 🔺	Predicted rel Search	Notes preview Search
		122.5	4	1		third cousin	
		114.8	4	1		third cousin	
Ch	uster 4 (4 items)						
		183.3	4	4	7	third cousin	
		165.4	5	4		third cousin	
		94.8	4	4		third cousin	
		92	5	4		third cousin	
Ch	uster 3 (4 items)						
		217.8	3	3	1003	second cousin	
		215.4	3	3		second cousin	
		114.1	3	3		third cousin	
		113.6	3	3		third cousin	
Ch	uster 2 (7 items)						

Figure 12 - Table view of the matches for each cluster based on an AutoCluster analysis for a profile.

- A. Name field, clickable for matches
- B. Shared cM
- C. Number of shared matches
- D. Cluster member
- E. Tree link (the number of people in the tree)
- F. Predicted relationship
- G. Preview of the notes
- H. Click this button to display the complete note

The results of the AutoCluster analysis are compressed and attached as a ZIP file. A unzipped AutoCluster report will contain:

- HTML file containing a visual representation of the AutoCluster analysis
- Excel file containing the chart visualization in a spreadsheet format, especially useful when there are a large number of matches in the HTML chart. In addition, the Excel fike contains all downloaded matches (all matches for FTDNA and 23andme) and the matches per cluster.

Start an AutoCluster analysis

From the profiles view page, click on the AutoCluster button which will display a page that allows for an AutoCluster analysis . For all three DNA testing companies, a maximum and minimum cM threshold can be selected. You can specify a range using these two cM thresholds to define criteria which DNA matches should be examined. In addition, the minimum cluster can be specified. For analyses with a low value for the minimum cM threshold a more powerful server is employed. For FTDNA analyses, this server is the default option.

Some additional options have been implemented that are only implemented for a specific DNA testing company. We will therefore discuss the AutoCluster analysis for each of the different companies.

Start an FTDNA AutoCluster analysis

In addition to the minimum and maximum cM setting it is possible to specify the minimum size of the largest DNA segment (see Figure 13). In addition, the AutoTree functionality is also available for FTDNA analyses (see section below). Another new feature for FTDNA (and 23andme) profiles is the ability to integrate DNA segments into the AutoCluster analysis.



Figure 13. The AutoCluster analysis page for an FTDNA profile.

DNA segment browser for 23andme and FTDNA AutoCluster analyses

Using the segment data and a chromosome browser we can color the segments of matches from a cluster. This allows users to see how much DNA is in common with them (see Figure 14). Before we visualize the shared DNA segments we perform a clustering to group segments that are overlapping (min 5 cM). Next, these segment clusters are visualized using a certain color. In addition to the graphical representation a table is available that contains the detailed information for the segment clusters. Segments for the DNA matches for each AutoCluster cluster are available and can be accessed using the table underneath the chromosome browser. This table contains a link to the detailed chromosome

browser, the number of multiple segment clusters, number of single segment clusters and number of clusters that are on the X chromosome.

It is now possible to generate a chromosome map from your DNA matches from FTDNA or 23andme clusters and import the segment data into DNA painter using the cluster auto painter tool (<u>https://dnapainter.com/tools/cap</u> also see the blog post: <u>https://dnapainter.com/blog/cluster-auto-painter-unravel-your-dna-test-results/</u>). Importing the chromosome map from your clusters of DNA matches into DNA painter allows you to:

- Make notes and identify clusters as maternal or paternal
- Look at the segments behind the clusters and identify potential pile-up regions.



Figure 14. Chromosome segments from DNA matches in clusters.

To supplement the segment analysis on DNA painter, a chromosome browser is available for the matches per cluster as well the combined matches overview (all segments from all matches from all clusters, see Figure 16).

An example for a single cluster can be seen in Figure 15. Cluster 31 from a 23andme clustering has a number of matches that probably have a triangulated segment (visualized using the helix symbol). Information concerning segment clusters, (multiple and single), x match clusters as well as paternal or maternal classification (as obtained from 23andme or FTDNA) is available in the table underneath (Figure 15B). Upon clicking on the segment link in the first column, a chromosome visualization is displayed (see Figure 15C). One segment is shared with the matches of this cluster which is also illustrated in the detailed (see Figure 15D) table that contains the start, stop positions as well as the total length of the segment.





D		Chromosome	segment statist	cs per Auto	Cluster clus	ster
uste	*	single_segments	multiple_segments filter column	x_segments A	Paternal filter column.	Moternal A
- (33 items)						
Segments for cluster 1		2	1	0	6	0
Segments for cluster 2		1	0	0	2	0
Segments for cluster 3		2	2	0	9	0
Segments for cluster 4		2	0	0	2	D
Segments for cluster 5		2	2	2	3	D
Segments for cluster 31		0	1	0	12	0
Segmenta for cluster 32		1	0	0	1	0
Segments for cluster combiner	d	34	32	6	75	0

Chromosome segments

D	Segment Cluster Information									
Claster A Segment du	Chr A	Start A	Step	SNP count	N	eM	Total cM	Paternal A	Maternal A	AutoCluster A
31	1	117750719	162351586	3244	Tod	27.1	25	Р		31
31	1	117750799	161493580	2998	мі	25.9	25	Р		31
31	1	117750799	161234623	2953	<u>Do</u>	25.6	24	P		31
31	1	117827683	161068836	28.89	Jeff	25.3	24	P		н
31	1	117750799	160825313	2862	<u>liff</u>	25.3	24	P		31
31	1	117827683	160800480	2837	P28	25.1	24	P		31
31	1	117827683	159599414	2491	<u>Sa</u>	22.4	21	P		31
31	1	117827683	159278251	2422	Bec	22.3	21	P		31
31	1	117827683	159278251	2422	Ale	22.3	21	P		31
31	1	117750799	158655080	2283	Nic	21.4	20	P		31
31	1	117827683	158725194	2293	Jan	21.3	20	P		31
31	1	119074219	158896122	2141	<u>Kxl</u>	20.7	20	P		31

Figure 15. 23andme segment example. For cluster 31 quite a few triangulating segments seem to be present.

Figure 16. All segment clusters from all clusters visualized in the chromosome browser

AutoCluster analysis using the extend cluster feature

One of the more complicated features is the "extend clusters" feature. This feature allows users to focus on the ancestral lines shared with a particular match. The best way to illustrate this feature is by using the following example. Let's imagine you have a high cM match, like a first or second cousin. You are interested in all ancestral lines shared with this match and see which shared matches form clusters that underly these ancestral lines.



Figure 17. AutoCluster analysis performed using the shared matches shared with a single high cM match.

FTDNA extend cluster AutoCluster analysis

To run the extended AutoCluster for FTDNA first run a regular AutoCluster/AutoTree for the profile of interest and extract the results. In the extracted Gephi folder, a CSV file with the matches will be available. Open the file in Excel and find the match of interest. Next, locate the ResultID2 column and copy the ID of the match and provide it to the AutoCluster panel (see Figure 18).

Enable the "Extend cluster" feature and set the parameters. Now the shared matches of the match of interest are retrieved after which shared matches are downloaded for these matches (see Fig 22 for an example that employed a high cM match).



Figure 18. FTDNA AutoCluster interface with a text box that allows for the group-like clustering

You can also remove matches by placing a single exclamation mark in front of FTDNA id. And you guessed it, by using two exclamation marks, you can remove complete branches. Using the double !! feature, **matches** and their **shared matches** will be removed from the analysis.

AutoTree

AutoCluster first organizes your DNA matches into shared match clusters that likely represent branches of your family. Everyone in a cluster will likely be on the same ancestral line, although the MRCA between any of the matches and between you and any match may vary. The generational level of the clusters may vary as well. One may be your paternal grandmother's branch, another may be your paternal grandfather's father's branch.

By comparing the linked and unlinked trees from the members of a certain cluster, we can identify ancestors that are common amongst those trees. First, we collect the surnames that are present in the trees and create a network using the similarity between surnames. Next, we perform clustering on this network to identify clusters of similar surnames. A similar clustering is performed based on a network using the first names of members of each surname cluster. Our last clustering uses the birth and death years of members of a cluster to find similar persons. As a consequence, initially, large clusters (based on the surnames) are divided up into smaller clusters using the first name and birth/death year clustering. See also the blog post of Roberta Estes from DNAeExplained that covers the AutoTree feature: https://dna-explained.com/2019/12/02/genetic-affairs-reconstructs-trees-from-genetic-clusters-even-without-your-tree-or-common-ancestors/

The common ancestor and location analysis is calculated using members of AutoCluster clusters as well as all using all matches from these clusters. This last step ensures that common ancestors that are present in different clusters (for instance clusters that are part of a supercluster, for instance, the first clusters from the chart of Figure 11) are identified as well.

The overview of these analyses is displayed in the main AutoCluster HTML file in a table (see Figure 19). For each AutoCluster cluster the number of common ancestors, common locations (for two distances) and common surnames are shown. The fields of the tree, common ancestor and common location are clickable and will show more detailed information.

Tre	e	🔺 # ancestors 🛛 🔺 # Radius 100m 🔺 # Radius 5000m			# Radius 5000m		surnames 🔺		
						Search for surnames			
•	(7 items)								
	Tree(s) of cluster 1		334 common ancesto	rs	96 common locations		93 common locations	5	Balentine (37) Sport (18) Baxter (9) Branscum (23) Crisel (11) Bumgarner (9) Harrison (4) Owings (7) Proctor (6)

Figure 19. AutoTree overview which shows the number of common ancestors, locations and surnames for each of the AutoCluster clusters as well as the combined analysis.

If there are not too many matches, an AutoTree analysis is performed on all matches. This particular analysis combines all matches from the clustering in a single cluster and perform the AutoTree. It sometimes finds common ancestors between members of separate clusters (e.g., in a supercluster).

Next, in addition to identifying the common ancestors, we combine the common ancestors and try to reconstruct the underlying genealogical tree. In most cases, only parts of the trees can be reconstructed. But, with some manual efforts, most automatically generated trees can be combined into one or several larger trees. To improve the analysis of the trees, we use a color gradient to differentiate between different DNA matches. In addition, persons in the tree are highlighted when you hover over the edges if they appear in different trees (see Figure 20).



Figure 20. Reconstructed genealogical tree based on the descendants of common ancestor J Freeman and A harding. Quite some of the descendants are a common ancestor as well. At the far right, the linked DNA matches are displayed (in green the profile of the tested person). In yellow/brown are tree persons that are retrieved from an unlinked tree.

The visualized tree persons are clickable which will show more detailed information. In addition, upon clicking on the cM value the relationship probabilities are displayed (adapted from the <u>Shared cM</u> <u>Project 3.0 tool v4 project</u>, see Figure 21).

Elke Hegna with tree 8417734 shares 176.0 cM. Relationship probabilities:

51.03% GGGG Aunt / Uncle Great-Great-Great-Great-Great-Great-Great-Great-Great-Great-Aunt / Uncle 1C3R 2C1R Half 1C2R Half 2C

34.51% Great-Great-Great-Great-Great-Great-Great-Great-Great-Great-Aunt / Uncle Half GG-Niece / Nephew Half GG-Aunt / Uncle Half Great-Great-Great-Niece / Nephew Half Great-Great-Aunt / Uncle Half Great-Great-Niece / Nephew 1C2R 2C Half 1C1R

10.59% Half GGGG-Aunt / Uncle 1C4R 2C2R 3C Half 1C3R Half 2C1R

2.04% Great-Great-Grandparent Great-Great-Aunt / Uncle Great-Great-Niece / Nephew Half Great-Aunt / Uncle Half Great-Niece / Nephew 1C1R Half 1C

1.82% 1C5R 2C3R 3C1R Half 1C4R Half 2C2R Half 3C

×

Figure 21. Relationship probabilities for a DNA match (relationship probabilities adapted from shared cM project 3.0 v4).

The complete list of common ancestor clusters with information regarding the underlying tree persons, their spouses, descendant, and linked DNA matches are available in the common ancestor's table (see Figure 22). This table is also available in de the Excel file.

Common Ancestor clusters

Common ancestors are identified using three analyses. First, a surname clustering is performed followed by a first name clustering. Last a clustering applied based on the birth and death years. In some cases the common ancestor clusters are not visualized in the recobstructed tree. In this scenario, descendants of that common ancestor are covered by another branch of tree or via the husband. Information for each of the persons in each common ancestor cluster and their linked DNA matches is displayed in the table underneath.

Tree Name Birth Death Descendant Name сМ Notes preview Search cl Search first name location location Search tree name ▼ William Conrad Easterly 1865 - 1937 m Emily Jane Chism 1859 - 1920 (3 items) 0 8417734 (27) William Conrad Easterly 1865 Big Flat, ... Maggie Ellen East... Elke Hegna 176. 0 157289583 (29) Big Flat.... William Conrad Easterly 1865 -William Hopkins E... Vania Pamplin 134. O Fulton Co... Jesse Pinkney Ea... den Braber/Easterly (155) William Conrad Easterly 1865 - 1937 tested person -1 ▼ Emily Jane Chism 1859 - 1920 m William Conrad Easterly 1865 - (3 items) 0 8417734 (27) Emily Jane Chism 1859 - 1920 Fowler, M... Maggie Ellen East... Elke Hegna 176. 0 157289583 (29) Emily Jane Chism 1859 - 1920 Fowler, M... William Hopkins E... Vania Pamplin 134. 0 den Braber/Easterly (155) Emily Jane Chism 1859 - 1920 Izard Cou.... Jesse Pinkney Ea... tested person -1

Figure 22. Common ancestor table that contains all identified common ancestors, husbands, descendants and linked DNA matches.

Using the birth location information from tree persons, we calculate location clusters using a minimum location distance. These clusters are available using the common location table which contains the location, linked tree, linked DNA match and the tree persons linked to this location (see Figure 23).

Common locations table using 5000m

We collect locations of recent ancestors that are present in the trees from different DNA matches. Locations that are characteristic of the identified clusters can yield information concerning the historical or demographic significance of a cluster. To identify these location clusters, we perform a clustering based on the distances between the entered birth locations of the persons from each of trees of the DNA matches. Birth locations that are identical or in close proximity (i.e. within a certain radius of meters) are placed into location clusters. These common locations are displayed in the table underneath.

Cluster			
Search for cluster			
🕶 0 (1 item)			
Location cluster with 3	trees and 6 people using a	distance	
Common Io	Tree	Match 🔺	Persons
Search	Search	Search	Search
Tennessee	MARGARET METZ EA sharondroberts (194.505 cM)		Mary Polly Gibbons 1814 - 1919 m Zachariah Kitchens 1820 - Wiley F Hale 1817 - 1865 m Malinda King 1810 - Zachariah Kitchens 1820 - m Mary Polly Gibbons 1814 - 1919 Margaret E. Hale 1846 - 1902 m John T. Kitchen 1846 - 1898
Tennessee	Cook Family Tree	Fredrick Kershner (133.951 cM)	Nancy C Rose 1855 - m Berry A Rose 1847 - 1939
Tennessee	Charles Mckean Famil	Mayra Mendiola (92.193 cM)	Louisa Mitchell 1827 - 1872 m James Wesley Easterly 1826 - 1875

Figure 23. Common locations table that contains the location clusters and matches of this cluster.

AutoPedigree

AutoPedigree is a feature that employs the AutoTree predictions. It is developed to identify how a person, for instance an adopee, fits into a reconstructed AutoTree. In short, the AutoPedigree feature automates the generation and testing of hypotheses using reconstructed trees from AutoTree.

Our approach has been inspired by the WATO tool that has been built to help solve DNA puzzles (including unknown parentage cases) by undertaking calculations as described by Leah Larkin in her series Science the heck out of your DNA.developed to identify how a person, for instance an adopee, fits into a reconstructed AutoTree. Our approach has been inspired by the WATO tool that has been built to help solve DNA puzzles (including unknown parentage cases) by undertake cases) by undertaking calculations as described by Leah Larkin in her series Science the heck out of your DNA.



Figure 24. Partial AutoPedigree with different generated hypotheses and their ranks. The actual place in the tree is represented with the green tester rectangle

Filter

Based on the common ancestors from the reconstructed trees, we create siblings for each of the identified ancestors. Next, we generate descendants (also called hypotheses) that could serve as a hypothesis (see example in Figure 24). What that means is the following, each generated descendant could represent the actual test taker (for instance an adoptee). But given the cM values of the DNA matches in the tree, some generated descendants are more probable than others. This probability is a measure of how likely a certain relationship is to occur. For instance, a DNA match sharing 229 cM has a probability of 54% of being a 2C but a 0% probability of being a 4C (see Figure 42 for screenshot of the shared cM project that employs these probabilities as well).

By multiplying each of the probabilities for each of the DNA matches in the tree, a score can be calculated for a certain generated hypothesis. This type of analysis can also manually be performed using the online WATO tool (see Figure 26).

The Shared cM Project 4.0 tool v4

Enter the total number of cM for your match here:



Then any relationships that fit will stand out below Click here for a shareable link to the cM amount above New Click on any relationship to view a histogram

Relationship prol	babilities (based on stats from The DNA Geek)										
53.99%	Half GG-Aunt / Uncle 2C Half 1C1R 1C2R Half GG-Niece / Nephew										
34.51% Half 2C 2C1R Half 1C2R 1C3R											
8.96%	Great-Great-Aunt / Uncle Half Great-Aunt / Uncle Half 1C 1C1R Half Great-Niece / Nephew Great-Great-Niece / Nephew										
2.54%	2.54% Half 1C3R † Half 2C1R † 3C 2C2R										
this relationship probabilities, but fall	has a positive probability for 229cM in thednageek's table of soutside the bounds of the recorded cM range (99th percentile)										





Figure 26. WATO representation of the AutoPedigree output. The different hypotheses are provided with a score.

All generated AutoPedigree trees are available in the WATO format, allowing users to import them into WATO. This allows for further tweaking of the trees, for instance if the AutoTree wrongly identified a common ancestor. In addition, matches from other companies that are known to be descendants as well can then be added.

The table underneath the visualized AutoPedigree tree summarizes the different hypotheses (see Figure 27). Each row represents a hypothesis, the MRCA and the ranked combined odds ratios. This odds ratio score is calculated based on the probability of that hypothesis dividid by the smallest probability of another hypothesis. Next, we compare the score to the next, slightly smaller, score and calculated the ratio between them. For instance, if the best combined odds ratio is 200 and the second best is 50, the compared score would be 4 (200 divided by 50). The last columns show the probability of each DNA match and the generated hypothesis. By clicking on the download button above the table, the complete table becomes available as an Excel file.

			Download spreadsheet wit						
Hypoth	eses	Most Common Re	ecent Ancestor	Calculations	with 0 ignored	Probability of each I	ONA match and hypoth	esis	
Hypothesis A	Child	MCRA A	MCRA A	Combined -	Compared ≜ to	match J.W.	match M.S. A	match A.C – 134.6 cM	match J.H. A
Titler Colui	Inter c	niter column	Inter column	ratio	previous				
hyp_44	child2	James Wesley Easterly 1826 - 1875	Louisa Mitchell 1827 - 1877	634	1	14.16%	38.89%	50.57%	32.15%
hyp_14	child2	Jesse Pinkney Easterly 1894 - 1982	Opal Nellene Reece 1910 - 1986	617	2	28.14%	19.06%	50.57%	32.15%
hyp_8	child3	William Conrad Easterly 1865 - 1937	Emily Jane Chism 1859 - 1920	311	2.2	14.16%	19.06%	50.57%	32.15%
hyp_15	child3	Jesse Pinkney Easterly 1894 - 1982	Opal Nellene Reece 1910 - 1986	139	1.8	52.78%	5.37%	23.28%	29.83%
hyp_7	child2	William Conrad Easterly 1865 - 1937	Emily Jane Chism 1859 - 1920	77	1.2	52.78%	30.3%	16.11%	4.23%
hyp_45	child3	James Wesley Easterly 1826 - 1875	Louisa Mitchell 1827 - 1877	63	3.9	4.24%	30.3%	23.28%	29.83%
hyp_43	child1	James Wesley Easterly 1826 - 1875	Louisa Mitchell 1827 - 1877	16	1.2	52.78%	6.39%	16.11%	4.23%
hyp_24	child2	Beard Hopkins Easterly 1857 - 1925		13	1.2	4.24%	6.39%	23.28%	29.83%
hyp_9	child4	William Conrad Easterly 1865 - 1937	Emily Jane Chism 1859 - 1920	11	6.2	4.24%	5.37%	23.28%	29.83%
hyp_30	child2	Eunice Faith Juanita Easterly 1942		2	1.6	0.68%	5.37%	23.28%	29.83%
hyp_3	child3	William Hopkins Easterly 1897 - 1965	Grace U Mcclung 1899 - 1982	1	1.1	4.24%	5.37%	16.11%	4.23%
hyp_13	child1	Jesse Pinkney Easterly 1894 - 1982	Opal Nellene Reece 1910 - 1986	1	1	0.68%	30.3%	16.11%	4.23%
hyp_1	child1	William Hopkins Easterly 1897 - 1965	Grace U Mcclung 1899 - 1982			52.78%	30.3%	0.0%	0.0%
hyp_2	child2	William Hopkins Easterly 1897 - 1965	Grace U Mcclung 1899 - 1982			14.16%	19.06%	0.0%	0.0%
hyp_4	child4	William Hopkins Easterly 1897 - 1965	Grace U Mcclung 1899 - 1982			0.0%	0.0%	50.57%	32.15%
hyp_5	child5	William Hopkins Easterly 1897 - 1965	Grace U Mcclung 1899 - 1982			0.0%	0.0%	23.28%	29.83%
hyp_6	child1	William Conrad Easterly 1865 - 1937	Emily Jane Chism 1859 - 1920			28.14%	38.89%	0.0%	0.0%

Figure 27. AutoPedigree scores table

Some hypotheses contain probabilities that are 0.0%, indicating that this relationship is not possible when taking into account the cM value of the DNA match and the proposed genealogical link. For instance, a DNA match that shares 200 cM cannot have a relationship of a 4C.

The generated hypotheses are visualized as descendants in the reconstructed AutoTree visualizations. For instance, hyp_14_child2 represents the 14th hypothesis and the second child. Instead of supplying the scores, we are providing the rank of the score in a badge. Scores that have a probability of 0 are placed in a red badge, the top 5 scores are placed in a green badge and the remainder in an orange badge. Upon clickin on the badge, a popup will appear that holds more information concerning the calculation of the score (see Figure 28).

A lot of hypotheses are tested for AutoPedigree. Therefore, in order to improve the visibility we prune the AutoTree tree by only displaying generated descendants if positive probabilities are available for that branch.



Figure 28. Pop up visualization upon clicking on the rank badge.

In some cases a DNA match has multiple links with the tested person, for instance DNA match M.M. that shares 157.9 cM and has common ancestor J Ozinga and B Ozinga (see Figure 29). The amount of cMs that is shared with the tested person is therefore inflated. To correct for this, we employ an approach that divides the amount of shared cM based on the different genealogical paths. We therefore attribute a larger fraction of cM to the DNA match if the tested hypothesis has a shorter path to the hypothesis as compared to the other path(s). Despite this measure, caution should be taken when encountering these DNA matches. The grey cells in the AutoCluster charts can be indicative of matches that are linked to more clusters and therefore linked via multiple ancestors.





Figure 29. AutoTree reconstruction with a DNA match that has multiple links with the tested person.

A single inaccurate hypothesis in the tree can potentially nullify the overall hypothesis, making its score zero. Unfortunately, these hypotheses are sometimes inevitable, for instance because a match is related via multiple genealogical links whereas only one line is identified. In this case you might end up with a predicted 4C (based on the reconstructed tree) that shares much more DNA with the tester as is expected based on the 4C relationship. If necessary, we therefore also perform the same automated analysis while ignoring one or two of these cases. If the rank badge starts with a digit, this digit will represent the number of ignored probabilities (2_RANK:4 indicates a hypothesis ranked 4th for which 2 probabilities were ignored). See Figure 30 for an example pop up with information concerning an ignored probability.



Figure 30. AutoPedigree with an ignored probability.

Invoking the AutoPedigree can be accomplished by going to the AutoCluster or AutoTree interface (see Figure 31). Select the common ancestor (AutoTree) option as well as the AutoPedigree. Next, select the min cM threshold for the analysis. This threshold indicates the minimum of shared cM that a match should share with the tested person. It is advised to use a 40 cM limit but it is possible to go down to 30 cM.



Figure 31. AutoPedigree interface, the min shared cM and half relationships can be set.

AutoSegment – segment based clustering

AutoSegment automatically organizes your matches using shared segment clusters. It employs locally downloaded segment files, therefore no scraping and website credentials are not needed. It works for MyHeritage, 23andme, FTDNA and GEDmatch segments (see Figure 32 for MyHeritage example). An AutoSegment analysis can be started using this link: <u>https://members.geneticaffairs.com/autosegment</u>



Figure 32. AutoSegment chart for MyHeritage profile with manually added maternal/paternal annotations

Let's first examine some visual examples before explaining how the AutoSegment works. Underneath the main visual chart, a table (see Figure 33) is available that contains general characteristics of the clusters. For instance, for which chromosomes segment clusters have been found, the number of paternal/maternal matches linked to a cluster, the number of segments and number of segment clusters for that cluster.

Chromosome segment statistics per AutoSegment cluster

The following table shows the AutoSegment statistics per cluster. A link to each cluser is provided. In addition, the chromosomes linked to the segment clusters and the number of DNA matches for each cluster is shown. Last, the number of paternal/maternal DNA matches is available as well the number of segments and number of segment clusters.

		Download spreadshee	t AutoSegme	ent statistics	
Cluster 🔺	chr 13	#mat Pat filter colu filter cc	Mat 🔺	Nr of se	Nr of Se filter column
AutoSegment cluster 1	1,2,3,4,5,6,7,8,9,10,12,13,	9	1	24	117
AutoSegment cluster 15	1,8,9,10,13	5		6	26
AutoSegment cluster 42	7,13,18	25		3	35
AutoSegment cluster 43	13	6		1	7
AutoSegment cluster 57	13	5		1	5
AutoSegment cluster 73	13	8		1	8
AutoSegment cluster 76	13,20	5		2	7
AutoSegment cluster 86	11,13	5		2	8

Figure 33. Table that shows main characteristics of each AutoSegment cluster

After clicking on one of the links, a report displayed. This chart will display a chromosome representation (see Figure 34) of the chromosome locations that are underlying these segments.



Figure 34. Chromosome visualization of the segment clusters

In this case, the segment cluster contains segments located on chromosome 13.

Underneath the chromosome browser a table is displayed that contain the segment clusters that are identified (see Figure 35). This is the core of AutoSegment, these are the overlapping segments that are identified from the analysis. This table shows the main cluster where the DNA matches are residing, the segment cluster, chromosome, start and end positions. To quickly assess the segment overlap between the segments, a visual segment representation is added. The number of SNPs is reported as well as the name of the underlying DNA matches, the DTC, the cM of the segment as well of the total cM of the match. If the match has a paternal or maternal annotation, this information will be displayed in the last two columns.

Clus A	Segment 🔺	C 🔺	Start Search	Stop Search	Segment representation	SN A	Name Search for name	DTC	cM A	To Pa A	M
➡ 67 (8 iter	ns)										
73	67	13	29732994	41948151		7424		MyHer	19.1	32.5	
73	67	13	29732994	42438349		7680		MyHer	19.6	27.2	
73	67	13	29732994	42438349		7680		MyHer	19.6	26.2	
73	67	13	29732994	42438349		7680		MyHer	19.6	19.6	
73	67	13	29732994	42438349		7680		MyHer	19.6	19.6	
73	67	13	30079484	42696870		7552		MyHer	18.6	26.2	
73	67	13	31275633	39495776		4992		MyHer	12	33.7	
73	67	13	33593270	41948151		4864		MyHer	11.7	19.3	

Segment Cluster Information

Figure 35. Segment cluster table

However, and this is also a word of caution, AutoSegment identifies overlapping segments that do not necessarily triangulate! The reason that we want to identify overlapping segments is because some of them will actually triangulate, and we are interested in identifying triangulated segments because they can point to a common ancestor. Conveniently, MyHeritage provides a chromosome browser (https://www.myheritage.nl/dna/chromosome-browser) that will show if these segments are

\bigcirc	MyHeritage	Start	Stamboom	Ontdekkingen	DNA	Gezondheid	Onderzoek
<	U +	U +		U +	U +	U +	U + >
11	Getrianguleerd Chromosoom 13	segment					
12	Genomische positie: RSID: Segmentgrootte: Aantal SNP's:	33593270 - 41948151 rs2283368 - rs9525479 11,7 cM 4.864					
13							

triangulating (see

Figure 36). The segments identified were checked and found to be triangulating.

Q	MyHeritage	Start	Stamboom	Ontdekkingen	DNA	Gezondheid	Onderzoek
<	U +	U +		U +	U +	U +	U + >
11	Getrianguleerd Chromosoom 13	segment					
12	Genomische positie: RSID: Segmentgrootte: Aantal SNP's:	33593270 - 41948151 rs2283368 - rs9525479 11,7 cM 4.864					
13							

Figure 36. Chromosome browser representation of MyHeritage, indicating that the visualized segments triangulate.

AutoSegment concepts

We will now discuss the underlying concepts of the AutoSegment clustering. The regular AutoCluster analyses are all based on shared match data, meaning that if a DNA match shares DNA with another match, they are shared matches and they might end up in the same cluster.

Let's examine one of these clusters. In this example, we will look at a cluster (see Figure 37) from a AutoCluster analysis based on a FamilyTreeDNA profile. As you can see, it's a well connected cluster, meaning that all members are sharing DNA with each other.



Figure 37. Example cluster from regular AutoCluster analysis for a FTDNA profile.

Now let's examine these matches using the chromosome browser of FamilyTreeDNA (see Figure 38). As we can immediately see in the chromosome browser, all shared DNA segments are not shared by other DNA matches. The exception seems to be on chromosome 7, but the two matches are closely related (mother/daughter). The only overlap with another match seems to be on chromosome 19.



Figure 38. Chromosome browser of all members of a certain FTDNA cluster

So in a way, by only looking at overlapping segments we are restricting ourselves. The fact that these matches in the cluster do not share a cluster does not make them less useful. They will still probably share a common ancestor. The question now might be asked, why then look at overlapping segments? There are several reasons. First, if the overlapping segments are found to be triangulating, they might be good candidates for DNA painting procedures. Second, the overlapping segment analyses can be performed using locally downloaded files, so there is no need to supply the website credentials and scrape the website. Third, some DNA testing companies, such as MyHeritage and GEDmatch, were not covered by our website. Last, since the overlapping segments are a generic procedure, we can employ the same approach DNA segment data from several DNA testing companies, thereby combining the matches into one single clustering. We will cover this hybrid approach in a later stage.

Another example of the use of shared match data vs overlapping segments is illustrated in Figure 39. This figure shows a regular AutoClusters chart as compared to the AutoSegment output. Since the AutoSegment analysis is restricted to overlapping segments, it shows a lower number of grey cells.



Figure 39. MyHeritage AutoClusters (based on shared matches), vs AutoSegment (based on overlapping segments).

Now for the underlying concepts of an AutoSegment analysis. Remember that DNA matches are linked via their shared matches but in this case, we don't have these available. Instead, we use segments and use these to link the matches.

The analysis starts out like a regular AutoCluster analysis. The user can define a specific max-min cM (for instance matches between 400 cM - 20 cM) but in addition a min segment overlap needs to be provided. For instance, 10 cM.

This segment overlap will be used as a minimum overlap measure, segments that share less than the provided amount, will not be linked. We calculate the segment overlap between 2 segments by looking at the overlap (see Figure 40). To provide the exact amount of overlapping cM information, we employ a human genetic map (build 37). However, before we calculate this overlap, we first check if the maternal/paternal is set, and if so, is the same for the underlying matches.



Figure 40. Segment overlap calculation

If the identified segment overlap exceeds the minimum overlap, the segments are linked. After linking all overlapping segments, we can examine these segment networks and identify segment clusters in them (see Figure 41 where we visualize 9 linked segments and are able to identify 2 segment clusters).



Figure 41. Linked segments. Numbers represent segments whereas an link between segments indicates an overlap.

After identifying many of these segment clusters, it's time to go back to our DNA matches. Ultimately, we want to cluster the DNA matches based on the segment clusters (see Figure 42).



Figure 42. Convert segment clusters to DNA match clusters

By using these segment clusters we can link DNA matches. Here is how it works, we examine all of the segment clusters and check if a certain DNA matchA and DNA matchB have a segment linked to this segment cluster. If so, a link between these two matches is created. Now the table described in Figure 33 makes more sense. Some high cM matches might be linked via several segment clusters and others only via one segment cluster.

Underneath the segment cluster chart, as described before, a table is displayed (see Figure 43) that contains the general segment statistics, cluster and segment cluster id, chromosome, start, end and visual representation.

Cluster Segment clu	Segment 🔺	Chr 🔺 Sear	Start A	Stop 🔺	Segment representation	SNP c	Name Segment clu	cM A	To 🔺
▼ 22 (3 items)									
1	22	1	14576441	20755790		2076	the lines	13.4	0
1	22	1	14128833	20755790	_88.000.000.000.000.000.000.000.000.000.	2276	-	14.3	0
1	22	1	14576441	20755790		2076	-	13.4	0
▼ 25 (2 items)									
1	25	1	30872516	47629198		4065	terms in the	18.2	0
	25	1	30660319	47629198		4165		18.7	0
1	31	11	76690268	108633181		8400	free of the	28.5	0
	31	11	87132158	108633181		2231	-	20.3	0
1	39	1	56987614	99291699		11597	terms in the	43.5	0
1	39	1	66512322	99626055		8797	-	30.3	0

Segment Cluster Information

Figure 43. Segment cluster information with general segment statistics.

In addition to the segment cluster tables displayed on each cluster page, there is a large table on the main page that describes all identified segment clusters. This overview is useful because in some cases DNA matches are not clustered but do appear in certain segment clusters. This complete overview allows the analysis of the DNA matches that did not make it into the main clustering (see Figure 44).

Download spreadsheet with segment clusters													
Cluster Search f	Segmen	C 🔺	Start Search	Stop Search	Segment representation	S A	Name Search		DTC A	CM Max	To 🔺	Pa	M
👻 65 (4 item	ns)												
<u>59</u>	65	10	100076444	116653136		8704			MyHerit	15.9	30.8		
<u>59</u>	65	10	97345469	116653136		10112			MyHerit	18.8	27.3		
<u>59</u>	65	10	98118257	117713221		10112			MyHerit	18.7	26.2		
<u>59</u>	65	10	99329418	112305321		6400			MyHerit	10.2	23.5		
👻 40 (2 item	ns)												
<u>75</u>	40	2	101598312	119751867		8320			MyHerit	17	50.3		
<u>75</u>	40	2	106811149	117686719		4480			MyHerit	10.1	18.4		
👻 87 (2 item	ns)												
<u>40</u>	87	7	101630597	121217473		8576			MyHerit	16.3	24.5		
<u>40</u>	87	7	105088693	121217473		7040	_		MyHerit	13.8	20.2		
🔻 74 (2 item	ns)												
<u>51</u>	74	8	1022799	4446859		4352			MyHerit	10.6	23.1		
<u>51</u>	74	8	164984	5323185		6144			MyHerit	13.6	28.8		
🔻 76 (2 item	ns)												
<u>50</u>	76	9	103675970	124109921		12544			MyHerit	28	28		
<u>50</u>	76	9	107370498	118885534		7552			MyHerit	17.9	24.6		

Individual segment Cluster Information

The following table shows the 376 DNA segments for each of the 96 identified segment clusters.

Figure 44. . Individual segment cluster overview for all identified segment clusters

In some areas in the genome, there are regions where a considerable amount of people are sharing certain DNA segments. These regions are known as so-called pile-up regions. For example, the segments from the segment cluster represented in

Cluster 🔺	Segment 🔺	C 🔺	Start 🔺	Stop 🔺	Segment representation	SNP c 🔺	Name 🔺	cM 🔺	To 🔺
Segment clu	Segment clu	Sea	Search fc	Search fc		Search fc	Segment clu	Max (Total
6 (103 items)									
• • (105 mms)	4	17	2000/066	22065728		0.01		12.2	120.6
1	0	15	20004900	33903738		- 0144		43.5	138.0
1	6	15	20004966	33439811		5632	-	41.3	125.3
1	б	15	20004966	34079300		6272		43.7	122.7
1	6	15	20004966	33439811		5632	-	41.3	121.1
1	6	15	20004966	33871785		6016		42.7	104.4
1	6	15	20004966	29581108	_00000000000000000000000000000000000000	3840	-	33.9	97.9
1	б	15	20004966	33439811		5632	-	41.3	95.4
1	6	15	20004966	33965738		6144	-	43.3	94.4
1	6	15	20004966	33719535		5888	-	42.5	93.9
1	б	15	20004966	33871785		6016	-	42.7	93.3
1	6	15	20004966	27770160		3072	-	30.8	92.2
1	6	15	20004966	28328485		3456	-	31.9	90.7
1	6	15	20004966	33719535		5888	-	42.5	90.4
1	6	15	20004966	33439811		5632	Inclusion.	41.3	89.9
1	6	15	20004966	33871785		6016	-	42.7	88.7
1	6	15	20004966	33582463		5760		41.9	88.2
1	6	15	20004966	27770160		3072		30.8	87.7
1	6	15	20004966	27770160		3072	-	30.8	87.6
1	6	15	20004966	33965738		6144	-	43.3	87.2

Segment Cluster Information

Figure 45 all are found on chromosome 15. This region overlaps with pile-up regions that are identified in a study of Li *et al* 2014 (see Figure 46).

Cluster 🔺	Segment 🔺	C 🔺	Start A	Stop 🔺	Segment representation	SNP c	Name 🔺	cM 🔺	To
▼ 6 (103 items)									
1	6	15	20004966	33965738		6144	-	43.3	138.6
1	6	15	20004966	33439811		5632	-	41.3	125.3
1	6	15	20004966	34079300		6272		43.7	122.7
1	6	15	20004966	33439811		5632	-	41.3	121.1
1	6	15	20004966	33871785		6016		42.7	104.4
1	6	15	20004966	29581108		3840	-	33.9	97.9
1	6	15	20004966	33439811		5632	-	41.3	95.4
1	6	15	20004966	33965738		6144	-	43.3	94.4
1	6	15	20004966	33719535		5888	-	42.5	93.9
1	б	15	20004966	33871785		6016	-	42.7	93.3
1	6	15	20004966	27770160		3072	-	30.8	92.2
1	6	15	20004966	28328485		3456	-	31.9	90.7
1	6	15	20004966	33719535		5888	-	42.5	90.4
1	6	15	20004966	33439811		5632	Inclusion.	41.3	89.9
1	6	15	20004966	33871785		6016	-	42.7	88.7
1	6	15	20004966	33582463		5760		41.9	88.2
1	6	15	20004966	27770160	_10000000000000000000000000000000000000	3072		30.8	87.7
1	6	15	20004966	27770160		3072	-	30.8	87.6
1	6	15	20004966	33965738		6144	-	43.3	87.2

Segment Cluster Information

Figure 45. Example segment cluster that overlaps with a known pile up region

Chromosome	Starting position	Ending position	Genetic length (in cM)
chr9	38,293,483	72,605,261	8.15
chr8	10,428,647	13,469,693	7.96
chr21	16,344,186	19,375,168	6.91
chr10	44,555,093	53,240,188	7.58
chr22	16,051,881	25,095,451	20.82
chr2	85,304,243	99,558,013	6.53
chr1	118,434,520	153,401,108	9.95
chr15	20,060,673	25,145,260	10.46
chr17	77,186,666	78,417,478	5.66
chr15	27,115,823	30,295,750	9.29
chr17	59,518,083	64,970,531	6.23
chr2	132,695,025	141,442,636	9.16
chr16	19,393,068	24,031,556	6.18
chr2	192,352,906	198,110,229	5.04
Total	14 regions		119.92

doi:10.1371/journal.pgen.1004144.t003

Figure 46. Identified pile up regions in a study of Li et al 2014

We have added an option to the AutoSegment interface that allows users to filter these known pile-up regions. Segments that are located in these regions are removed from the analysis.

However, there can still be some regions in the genome that are enriched with respect to segments. To provide insights into these regions, we visualize the number of segments for each chromosome (see Figure 47). The pileup report link is found in the main HTML, the file called pileup_report.html and created in the chromosomes folder.

Information

Pile up regions

AutoSegment identifies DNA segment clusters by clustering overlapping segment data from various DNA testing companies (MyHeritage, 23andme, FamilyTreeDNA and GEDmatch) using a minimum segment overlap of 10 cM. In some cases, certain areas on your DNA might be overrepresented with respect to the number of DNA matches that match on that area. These areas are also known as pile-up regions. By plotting the segment occurrences for each chromosome, we can obtain insights in these regions. For this particular analysis, previously identified pile up regions from the paper of Li et al (2014) were used to remove segments that are in these pile-up regions. As a consequence, a total of 349

segments were filtered that were present in known pile-up regions.



Figure 47. Personal pileup region report

GEDmatch triangulation data

The word of caution that was mentioned in the beginning needs additional explanations with respect to the GEDmatch analysis. For MyHeritage and FTDNA analyses we employ two files, one containing the DNA match data and one containing the segments. For 23andme analyses, the segments and DNA matches combined into one file. For GEDmatch we also employ 2 files, one segment file but the other file contain triangulation data (see next section how to obtain this data set). Since we have this triangulation data, we can now verify overlapping segments and only keep the overlapping segments that have a triangulation segment linked to them. This greatly improves the accuracy of the predictions.

Excel cluster representation

In some cases, the numbers of cluster in the HTML file is too large to open using a browser. Luckily, an Excel is created that will show a spreadsheet representation of the generated clusters. Usually, this file can be opened if the HTML is unresponsive.



Figure 48. Excel representation of the AutoSegment clusters

In addition, to still view the cluster table and segment cluster table information is contained in the HTML, open the file that ends with "_chart.html" which is the same HTML file without the chart.

Paternal & Maternal annotations

Profiles from FTDNA and 23andme that have tested some close relatives can sometimes have the maternal and paternal labels (see Figure 49). This information is used to assess if certain overlapping segments are valid. It is also possible to add these labels manually. To enable this, open the DNA match file CSV using a spreadsheet edit and add the "maternal" or "paternal" annotation in the last column of this spreadsheet. Save the modified spreadsheet as a CSV file and use it for the AutoSegment clustering.

All (2004)	🛉 Paternal (209)	🛉 Maternal (297)	👬 Both (2)		0	Calculating Family Mat	ching
Name	Match Date	Relationship Range 🔻	Shared cM	Longest Block X-Match	Linked Relationship	Ancestral Surnames	٥
	07/06/2015	Father/ Son	3,384	267	Father	(van der Veen) / Abeles / Aukes / Aetzes / Adema / Alberts / Alles /	e
	02/11/2016	Mother/ Daughter	3,384	267 X-Match	Mother	(Boetje) / (Deddes) / (Hoekstra) / (Jager) / (Luimstra) / (van der Harst) / Aabes / Abels	0

Figure 49. Paternal and maternal annotations for FTDNA

AutoSegment ICW for FTDNA

Regular AutoSegment analyses for FTDNA involve the use of the segment data that is contained in the CSV file. As mentioned before, AutoSegment is only able to identify overlapping segments when supplied with segment data from an external file. Therefore, an important part of a regular AutoSegment analysis is the manual analysis of the overlapping segment clusters, to verify if the segments triangulate. For FTDNA analyses, this would for instance involve the use of the matrix tool, as explained in this blog post of Roberta: https://dna-explained.com/2019/11/06/triangulation-in-action-at-family-tree-dna/. And, although the matrix tool does not tell you if DNA matches match each other on the same segment where they match you, there's a good probability that they do (especially if only one matching segment is involved).

The AutoSegment ICW feature uses shared match data to verify the overlapping segments. If segments overlap, we check if the underlying matches are a shared match. If they are, we keep the overlapping segment, if they aren't, the segments are not seen as overlapping segments and won't make it together in a triangulated group.

Further downstream analysis of the segment-based clusters is similar to the analysis of shared match clusters. In addition, because of the shared segment nature of these clusters, the identified clusters are also excellent candidates for the cluster auto painter feature from DNA Painter (https://dnapainter.com/tools/cap).

Since the AutoSegment ICW approach employs shared match data, the tool needs to be started using the AutoCluster interface.

Perfo	orm Aut	oCluste	r analys	is with B	Auto Blom	Tree	e featu	re for p	rofile Evert-Jan
Selecting a	a min threshol	d of 30 cM (d ensure tha	or lower) will at there is end	automaticall ough memor	ly result ry to cre	in the ate all	usage of a necessary f	more powerfu files.	Il server. This is to
Start AutoCluster analysis with matches which share a max of	Stop AutoCluster analysis with matches which share less than	Minimum size of largest DNA segment shared with the match	Sorting of Family Finder Matches	AutoTree identify common ancestors from trees	Y DNA	mtDNA DNA	ADownload segments for DNA Painter	AutoPedigree generate hypotheses automatically	AutoSegment analysis
250 c 💙	50 ct 🗸	10 ct 🗸	perc 🗸						
Paste here	the FTDNA ider	ntifiers in case o stering	f an ID based		Ex	tend sters	Min clu	ster size	
							2	~	Perform
									analysis

As a consequence, the AutoTree functionality and extend cluster features are also possible with AutoSegment ICW. It is therefore possible to identify common ancestors based on trees of matches and/or perform a targeted clustering using only a selection of your matches.

AutoSegment – retrieve offline data

This section describes how to obtain the DNA match and/or segment matches for MyHeritage, 23andme, FamilyTreeDNA and segment/triangulation data for GEDmatch.

Retrieving segment data for MyHeritage

Login to MyHeritage and visit the DNA matches page: <u>https://www.myheritage.com/dna/matches</u>

Click on the three vertical dots to expand the menu. Select the first option to retrieve the DNA match list, the second option will allow the retrieval of the segment data. Both files will be mailed.

DNA results This is you Select another person *	Test add The more re Get sugges	ditional family members slatives test their DNA, the more you'll discover. titions
Overview Ethnicity Estimate	DNA Matches Tools	\sim
Showing 1–10 of 4,273 DNA Matches		Y Filters F Sort by Q
		i≣ Export entire DNA Matches list
		Export shared DNA segment info for all DNA Matches
		(i) What are DNA Matches?
		e
		Review DNA Match View tree

Save the attachments from the e-mails and go to the Genetic Affairs AutoSegment page for MyHeritage: <u>https://members.geneticaffairs.com/autosegment/addWebsite/MyHeritage</u>

		Hi ejbl	om,		
Run an Au	toSegment analysis fo	or MyHeritage (using the segme	nt data and DNA match	file.
The MyHeritage D icon at the top rig and the file v	NA match file is avain the of the list of match will be emailed to you	lable on the Di es to expand ti within a few m	NA Matches page he menu. Click o inutes. Please ur	e by clicking the 'three v n 'Export complete DN/ nzip the file before uplo	rertical dots' A match list' bading.
The segment file i all DNA Matche	s on the same page a s' and the file will be e	s the DNA mat mailed to you upload	ch file. Click on 'E within a few min ding	Export shared DNA seg utes. Please unzip the f	ment info for île before
For more info conc p	erning the segment d ost from Patsy Colem	ownload, checl an that describ	k the faq section bes her findings v	of DNA Painter. Click h with AutoSegment.	ere for a blog
Start Stop AutoSegment AutoSegment analysis analysis with with matches matches which share which share a max of less than	Min Min cluster overlapping size segment size	Remove known pileups	AutoSegment name	Select match file	Select segment file
250 c 💙 25 ch 👻	15 ck 💙 2 👻			Bestand klezen Geen besta	and Bestand Kiezen Geen bestand gekozen

Adjust the search parameters, fill in the name field and select the segment file and match file. After you start the analysis, results will appear within 15 min in your mailbox. If no results appear, it is possible that too low cM values were employed. Try raising the minimum cM settings and retry the analysis.

Retrieving segment data for FamilyTreeDNA

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Login into FamilyTreeDNA and visit your Family Finder page:

<u>https://www.familytreedna.com/my/familyfinder</u>. Go to the bottom of the page and click on "Download Matches: CSV"

	02/25/2014	2nd Cousin - 4th Cousin	53	21	B ⁺	0
c i i i	12/20/2018	2nd Cousin - 4th Cousin	53	17	<u>n</u> +	D
c 🔮 🖓 🛶	05/06/2019	2nd Cousin - 4th Cousin	44	17	A Waterin (Hardenbe	k erg) O
C III	01/16/2017	2nd Cousin - 4th Cousin	42	17	<u>n</u> +	٥
Download Matches: CSV Download Filtered Matches: CSV					1-30 of 1970 《 < > 》 Page 1	/ 66 Go

Save the matches file to your local drive. Next, visit the chromosome browser page for the segment data: <u>https://www.familytreedna.com/my/family-finder/chromosome-browser</u>

Compare	DNA	A Matches			Q Search Fi	Download All Segments
With		Name	Relationship Range 🛧	Shared Segments	Shared cM	Longest Block
To compare, start by selecting up to 7 from your DNA matches		C ₀				:
		P ₀				:
		C 0				
		0				:

Click on "Download all segments" to download the segment data.

Next, go to the Genetic Affairs AutoSegment page for FamilyTreeDNA: https://members.geneticaffairs.com/autosegment/addWebsite/FamilyTreeDNA

			Hi ejbl	om,					
Run a	an AutoSegmer	it analysis for	FamilyTreeD	NA using the set	gment data and match	file.			
The FTDNA	The FTDNA DNA match file is available on the bottom of the FamilyFinder page. Click on 'Download Matches: CSV' link and the file will be made available to you.								
The segment The button is ftDNA, For more info o	Matches: CSV' link and the file will be made available to you. The segment file is available via the button 'Download All Segments' in the ftDNA chromosome browser. The button is at the top right of the page above the list of matches. Please note that if you transferred to ftDNA, you'll need to unlock their chromosome browser before you can download your data. For more info concerning the segment download, check the faq section of DNA Painter. Click here for a blog post from Patsy Coleman that describes her findings with AutoSegment.								
Start Stop AutoSegment AutoSegme analysis analysis with with matches matches which share which shar a max of less than	Min overlapping segment size e	Min cluster size	Remove known pileups	AutoSegment name	Select match file	Select segment file			
250 c 💙 45 cM	▼ 15 cħ ▼	2 ~			Bestand klezen Geen best	and Bestand Kiezen Geen bestand gekozen			

Adjust the search parameters, fill in the name field and select the segment file and match file. After you start the analysis, results will appear within 15 min in your mailbox. If no results appear, it is possible that too low cM values were employed. Try raising the minimum cM settings and retry the analysis.

If you still possess a segment/match CSV file for 23andme, you can run AutoSegment for 23andme. First, go to the Genetic Affairs AutoSegment page for 23andme:

https://members.geneticaffairs.com/autosegment/addWebsite/23andme

				Hi ejbl	om,	
F Down St d	Run an Autos nload aggreg croll to the be ownload the	egment ana ate data' on ottom of the file directly t	lysis for 23an the 23andme page to see t to your comp [name	dme using t e DNA Relativ his link). If yo uter: Downk e]_relatives_	he segment data. T ves page under 'Far ou are logged in, th oad Segment Data download.csv)	he file is available via the link mily and Friends' (you will need to e following direct link will also from 23andme (usually called
For m	ore info con	cerning the s post from Pa	segment dow atsy Coleman	nload, checł that describ	the faq section of es her findings with	DNA Painter. Click <mark>here</mark> for a blog n AutoSegment.
Start AutoSegment analysis with matches which share a max of	Stop AutoSegment analysis with matches which share less than	Min overlapping segment size	Min cluster size	Remove known pileups	AutoSegment name	Select segment file

Adjust the search parameters, fill in the name field and select the segment/match file. After you start the analysis, results will appear within 15 min in your mailbox. If no results appear, it is possible that too low cM values were employed. Try raising the minimum cM settings and retry the analysis.

Retrieving segment data for GEDmatch

GEDmatch is the only company that provides triangulated segments in addition to regular DNA segment data. AutoSegment employs both files since the regular DNA segment file contains information concerning the DNA matches that is not available in the triangulated segments file. The triangulated segment file greatly improves the quality of the AutoSegment predictions.

Log into GEDmatch and select the Tier 1 – Segment Search (https://www.gedmatch.com/segment_search.php):



Fill in the concerned Kit Number, select the max number of closest matches to consider (for instance 5000) and enable the "Prevent Hard Breaks" option:

$\mathbf{T}_{match}^{[GED]}$ Tools for Genealogy Research	Home Log out							
GEDmatch [®] DNA Segment Search								
This utility allows you to find other kits with matching chromosome segment than 2100 cm's are skipped to save resources.)	s. (Note that matches closer							
Kit Number:								
Max number of closest matches to consider:	[1000 V] Max Kits							
Build to Display (Choose just one):	○ B36 ● B37 ○ B38							
SNP count minimum threshold to be considered a matching segment (Leave blank for dynamically-calculated value (200 - 400))								
Minimum segment cM size to be included in total: (Leave blank for default value = 7)								
Prevent Hard Breaks (default is to create hard breaks when distance between SNP's exceeds 500,000 base positions):								
Chromosome to scan (or all)								
Optional segment start to match: (Only use if a specific chromosome is specified)								
Optional segment end to match: (Only use if a specific chromosome is specified)								
Show graphic bar for Chromosome?	YesNo							
	Submit							

Click on submit and let the tool analyze the data. After it is finished, click on the "here" button in the top of the screen and save the **csv** file to your local drive.

Next, we will download the triangulated data from GEDmatch. From the main page, select the triangulation option:

Tier 1
 One-To-Many DNA Comparison Beta One-To-Many DNA Comparison Q-Matching One-To-One Segment Search Phasing Triangulation Multiple Kit Analysis (MKA) Lazarus My Evil Twin Combine multiple kits into 1 superkit NEW! Clusters, Single Kit input, Basic Version NEW! Find common ancestors (MRCA) from DNA
matches NEW

Fill in the concerned Kit Number, select the max number of closest matches to consider (for instance 5000) and start the analysis by clicking on the submit button:

Tools for Genealogy Research	Home Log out
GEDmatch Segment Triang	ulation
his utility finds people who match you with your top close matches as shown pper threshold limit that you specify. It then compares those matches against e rromosome and position, or by kit number, chromosome and position, and the re ach matching segment larger than 7 cM. Close relatives can be excluded fi reshold limit. All kits must have completed batch processing to be included in	in the one-to-many results and below the each other. Results can be sorted by n displayed in tabular and graphical forma room results by specifying an upper segment n results.
Kit Number:	
Max number of closest matches to consider:	500 V Max Kits
Upper Segment Threshold Limit:	3000 ∨ cM
Minimum Segment length:	7 cm ✔ Minimum cM
Chromosome to triangulate (or all)	
Build to Display (Choose just one):	○ B36
Display Options:	 Show results sorted by chromosome segment start position Show results sorted by kit_number, chromosome, segment start position Show results sorted both ways
Cross-match triangulated segments with others within chromosome. Cross-match limit per chromosome: Note: For maxKits > 500, limiting the cross-match limit to 200 is advised, as rross-matching is CPU-intensive and when a set of triangulated segments for a chromosome is large (>200), the time to cross-match segments can grow very large. Only triangulated segments > 7 cM considered for cross-matching.	Cross-Match Limit
	Cubanit

Browse to the end of the page and locate the download link:

GEDmatch Segment Triangulation -- (V0.3)

Triangulation w	ith Kit					
All kits shown in co with a total matching Matches above 3000 3-Way (Triangulate Segments shown are Triangulated Segme	lumns Kit1 and Kit2 are taken from the closest 3000 matches t g segment count less than 3000 cM. cM (total) are not shown. d) segment matches are shown in green. This is an indication o larger than 7 cM and between 200 and 400 SNPs. nts : 501 of 501	to M020545 of common ancestry.				
Click HERE to do Triangulated results	wnload triangulated segment data to a comma-separated CSV i sorted by Chromosome, Start Position:	file.				
Click HERE to do Triangulated results Chr	wnload triangulated segment data to a comma-separated CSV i sorted by Chromosome, Start Position: Kit 1	file. Kit 2	Start	End	сM	
Click HERE to do Triangulated results Chr 1	wnload triangulated segment data to a comma-separated CSV t sorted by Chromosome, Start Position: Kit 1	file. Kit 2	Start	End	сM	
Click HERE to do Triangulated results Chr 1 1	wnload triangulated segment data to a comma-separated CSV t sorted by Chromosome, Start Position: Kit 1	file. Kit 2	Start 776546	End 4488979	cM	
Click HERE to do Triangulated results Chr 1 1 1 1	wnload triangulated segment data to a comma-separated CSV t sorted by Chromosome, Start Position: Kit 1	file. Kit 2	Start 776546 798959	End 4488979 3000924	cM 11.7 7.3	

Download the triangulated data by clicking on the 'here' link in the bottom of the screen. This will allow you to save a **CSV** file to your local drive.

Next, go to the Genetic Affairs AutoSegment page for GEDmatch: https://members.geneticaffairs.com/autosegment/addWebsite/GEDmatch

			Hi ejbl	om,					
Run an AutoSe	gment analysis i	for GEDmatch s available to	n using the s Gedmatch T	egment data an Tier 1 subscriber	d triangulated data (ple 's only).	ease note: this			
Gedmatch pro to include	Gedmatch provides a downloadable file of all segments via their 'Segment Search' report. Please make sure to include enough matches, for instance 5000. In addition, enable the option "Prevent Hard Breaks"								
GEDmatch also	o provides a tria	angulated seg	ments which	n are used to ve	rify identified overlappir	ng segments.			
Click here for a	blog post from	Patsy Colema	an that desc	ribes her finding	gs with AutoSegment ar	nd GEDmatch.			
Start Stop AutoSegment AutoSegme analysis analysis with with matches matches which share which shar a max of less than	Min overlapping segment size e	Min cluster size	Remove known pileups	AutoSegment name	Select triangulated file	Select segment file			
250 c 🗸 🚺	• 9 cM •	2 🗸			Bestand kiezen Geen best.	and Bestand Kiezen Geen bestand gekozen			

Adjust the search parameters, fill in the name field and select the triangulated and segment file. After you start the analysis, results will appear within 15 min in your mailbox. If no results appear, it is possible that too low cM values were employed. Try raising the minimum cM settings and retry the analysis.

Hybrid AutoSegment – combine MyHeritage, FTDNA, 23andme and GEDmatch

The logical successor of AutoSegment would be a version that would create a clustering output using the segments of all four different companies. This hybrid AutoSegment is now available using the link https://members.geneticaffairs.com/hybridautosegment (see Figure 50).

Hi ejblom,										
Run a hybrid AutoSegment analysis that combines segment data for MyHeritage, FamilyTreeDNA, 23andme and GEDmatch segment data.										
	Click here for a tutorial how to obtain the DNA match and segment data.									
Name o	of analysis	Min overlap segment s	ping Remove kr ize pileup: ()	own Perform liftove s for FTDNA	er Min cluster size 🖲					
hybrid_	clustering 💧	10 cM	~		2 🗸					
Company 🚯	Max shared 🚯		Min shared 🚯	Match fi	ile 🔁	Segment file 0				
MyHeritage	250 cM	✓ 25 0	·M ~	Bestand ki	ezen Geen bestand gekozen	Bestand kiezen Geen bestand gekozen				
	250-14			Match fi Bestand ki	ile 🕄 ezen Geen bestand gekozen	Segment file () Bestand kiezen				
FamilyTreeDNA	250 cM	• 35 (·M •							
GEDmatch	250 cM	✓ 15 0	:M ~	Triangul Bestand ki	ated file () ezen Geen bestand gekozen	Segment file () Bestand Kiezen Geen bestand gekozen				
				Match/seg	ments file 0					
23andme	250 cM	✓ 25 0	:M ~	Bestand ki	ezen Geen bestand gekozen					
				PERFORM H	YBRID AUTOSEGMENT ANALYSIS					
						-				

Figure 50. Hybrid AutoSegment interface

The interface options are quite similar to the default AutoSegment. However, the min overlapping segment size is now set for all four companies. The overlay messages will show which matches and from which company they are (see Figure 51).



Figure 51. Hybrid AutoSegment chart with overlay

The table that contains some general statistics and links to the individual cluster reports is now also reporting the number of matches per DNA testing company (see Figure 52).

The following table shows the AutoSegment statistics per cluster. A link to each cluser is provided. In addition, the chromosomes linked to the segment clusters and the number of DNA matches for each cluster is shown. Last, the number of paternal/maternal DNA matches is available as well the number of segments and number of segment clusters.									
Cluster A	chr 🔺	#mat filter colu	#F	#23 🔺	#MH	#G A Pat A filter cc	Mat 🔺	Nr of se	Nr of Se
AutoSegment cluster 1	1,3,7,10,11	7	2	1	2	2	2	7	25
AutoSegment cluster 3	3,12	4			4			3	7
AutoSegment cluster 4	8,11,18	4	1	1	2		1	3	9
AutoSegment cluster 5	11,18	3		1	2			2	6
AutoSegment cluster 6	1,2,3,4,8,X	8	1	3	3	1	1	7	22

Chromosome segment statistics per AutoSegment cluster

Figure 52. General cluster statistics

The same holds true for the table in the lower section of the main HTML page. This now contains the individual segment cluster information with information per DNA testing company.

Individual segment Cluster Information

The following table shows the 204 DNA segments for each of the 71 identified segment clusters

Download spreadsheet with segment clusters						t clusters						
Cluster A Search f	Segment cli	C A	Start A Search	Search	Segment representation	Sear	Name Search	DTC A	Max)	To 🔺	Pa 🔺	M A
👻 11 (3 item	is)											
<u>38</u>	11	5	1	5050830		1329		23andme	12.5	40		
<u>37</u>	11	5	14782	4841505		3840		MyHeritage	12.2	91.5		
<u>39</u>	11	5	81437	5176672		493		GEDmatch	15.9	26.6		
🔻 50 (2 item	IS)											
-	50	10	100076444	116653136		8704		MyHeritage	15.9	30.8		
1	50	10	92994805	117029683		6600		FTDNA	24	351		М
🗢 60 (2 item	IS)											
1	60	7	100159726	123897167		4700		FTDNA	18.3	351		М
-	60	7	97601052	129663496		1561		GEDmatch	24.3	23.3		
💌 24 (5 item	is)											
<u>23</u>	24	20	10752610	52308169		21504		MyHeritage	51	51		
<u>23</u>	24	20	17599087	44076324		11648		MyHeritage	24.5	32.1		
23	24	20	17770834	43374640		5400		FTDNA	23.6	53		М
23	24	20	17771604	44076324		11520		MyHeritage	24.2	30.2		
23	24	20	35335891	52308169		10112		MyHeritage	26.7	33.6		

Figure 53. Hybrid AutoSegment segment cluster chart

Information concerning the DNA match and segment files is provided in the main HTML file (see Figure 54). Also information is provided if there are any warnings concerning issues while importing these files. The results of the pile-up removal procedure (if selected) are also provided. The same for the FTDNA liftover analysis, with information concerning the largest change.

For GEDmatch some additional information is provided for the triangulation file. In some cases these triangulation reports generate data that are not provided in the segment file. We still would like to keep these segments since they represent high quality triangulated data.

To predict the cM values of these unknown matches, we first cluster (flatten) the triangulated segments such that we obtain a single segment cluster per segment location. The reason this is important because the triangulation file can contain many segments that are more or less on the same location. After clustering these matches we take the largest segment from the segment cluster and combine the cM values for each them. The combined cM values for these flattened cM values are then the total shared cM with this particular DNA match. It's most likely a deflated score but at least we use the available data to come to a certain value. These reconstructed matches can easily be detected by looking at the notes field of GEDmach matches. That will contain information concerning the reconstruction of this DNA match.

Settings used for this AutoSegment analysis

A **MyHeritage** analysis was performed for "aaa". From the supplied CSV file(null), a total number of 6508 matches were retrieved. After applying the cM settings (min 30.0 cM, max 600.0 cM), we removed a total of 6391 matches and continued the clustering analysis using 117 DNA matches. A total of 14 rules (14 rules that exclude segments, excluding thereby 31 segments) were employed.

A FTDNA analysis was performed for "aaa". From the supplied CSV file(s), a total number of 1943 matches were retrieved. After applying the cM settings (min 45.0 cM, max 600.0 cM), we removed a total of 1536 matches and continued the clustering analysis using 407 DNA matches. After importing the segment file, 4 warnings were obtained: Warning, for match Robert George White we already imported some segments, the match might be duplicated within the segment file? Warning, for match Claus Behrendt Møller we already imported some segments, the match John Wright Ballard we already imported some segments, the match might be duplicated within the segment file? Warning, for match John Wright Ballard we already imported some segments, the match might be duplicated within the segment file? A total of 14 rules (14 rules that exclude segments, excluding thereby 7 segments) were employed.

FamilyTreeDNA liftover procedure

The provided segments by the DNA testing companies are all based on a certain <u>human reference</u> <u>genome</u>. Since the hybrid AutoSegment tool identifies putative triangulating segments based on DNA segment positions, it is important to have the correct coordinates. FamilyTreeDNA employs the human genome build 36 wheras the other companies all support build 37. Most FamilyTreeDNA segments will have similar coordinates for both builds but in some cases, there can be a large difference (especially for chromosome 19). There are <u>liftover</u> tools that can convert the coordinates between different reference genomes. For 124 segments, we changed the start/end positions for total number of 208211196 basepairs. The largest segment change was found for: Daryl G. May Chr19:46394266-54350202 (15.6 cM) after liftover: 41702426:49658390 (12.8 cM)

A 23andme analysis was performed for "aaa". From the supplied CSV file(s), a total number of 1334 matches were retrieved. After applying the cM settings (min 30.0 cM, max 600.0 cM), we removed a total of 1309 matches and continued the clustering analysis using 25 DNA matches. During importing the segments from the CSV file, we couldn't find segment data for 301 DNA matches (probably due to their DNA sharing settings). A total of 14 rules (14 rules that exclude segments, excluding thereby 5 segments) were employed.

A **GEDmatch** analysis was performed for "aaa". From the supplied flat segment CSV file(s), a total number of 3889 matches were retrieved. After applying the cM settings (min 20.0 cM, max 600.0 cM), we removed a total of 3852 matches and continued using 37 DNA matches.

A total number of 7868 triangulated segments were obtained from the GEDmatch triangulation report. Of these segments, a total number of 48 segments were linked to an existing segment that was retrieved from the previously imported segment file. A total number of 6944 triangulated segments were not linked because the minimum segment size criterium (10cM) was not met. In addition, a total of 871 triangulated segments could not be linked to a DNA match from the existing DNA match list. However, based on the kit numbers, we were able to identify 81 previously unknown DNA matches using 871 segments. The unlinked triangulated DNA segments were converted to DNA matches after which the triangulating segments will be used to assess their total shared cM.

A total of 871 unlinked segments that triangulate with another segment were retrieved for 81 unknown DNA matches. These triangulating DNA segments often overlap with other trianuglation segments and should therefore be condensed. After performing this step, a total of 47 flattened segments were obtained. These flattened segments are better representatives of the actual segments shared by the kit owner and can also be used to assess the total amount of shared cM. Due to the cM settings (600.0-20.0 cM), a total of 80 reconstructed DNA matches based on triangulated matches were discarded, as were 0 triangulating segments because they did not meet the minimum segment size of 10 cM. A total of 1 DNA matches were reconstructed based on the triangulated segments and added to the existing set of 37 DNA matches. A total of 14 rules (14 rules that exclude segments, excluding thereby 4 segments) were employed.

Figure 54. Hybrid AutoSegment settings

In addition to the general statistics we also supply information concerning the DNA segments. This will include the number of segments imported, after filtering and the number of overlapping segments (see Figure 55).

DNA Segment statistics

Based on the 587 DNA matches we were able to retrieve 317 DNA segments (after filtering using a minimum segment size of 10cM, a total of 317 segments remained). Interestingly, we weren't able to retrieve segment data from the provided segment data file for the following 4 matches: A total number of 7 DNA segments were imported that originated from the X chromosome (chromosome 23). A total of 646 segment overlapping segment pairs were found (using the min cM overlap of 10 cM) whereas 99192 segment combination did not overlap and were therefore not used for the clustering analysis. Based on the 646 overlapping segment combinations a total of 71 segment clusters (containing 204 segments) resulted from the segment clustering and are linked to 152 DNA matches (59 FTDNA matches, 65 MyHeritage matches, 16 23andme matches, 12 GEDmatch matches,).

DNA match cluster analysis using overlapping segments.

As was mentioned in the previous sections, a clustering of overlapping segments was performed that resulted in segment clusters. This clustering links individual segments, so certain high cM DNA matches that share multiple DNA segments might be linked to multiple DNA segment clusters. To link the DNA matches, we do another clustering using their segment cluster membership. So 2 DNA matches can be linked if they are a member of the segment cluster. This clustering tries to mimic a regular clustering analyses that is performed using shared matches. Using this methodology, we were able to cluster 145 (56 FTDNA matches, 63 MyHeritage matches, 16 23andme matches, 10 GEDmatch matches,) DNA matches. Note that you can still obtain valueable information for the missing matches by examining the segment clusters (a summary is available in a table on this page). A summary concering the discarded cluster members is provided underneath:

Note: The following 7 matches met the inclusion parameters but were placed in an cluster that contains an amount of matches that is lower than the min cluster size of 2 matches and are therefore not included in the chart:

Figure 55. Hybrid AutoSegment segment statistics.

AutoLineage

What is AutoLineage

AutoLineage is designed to boost your genetic genealogy research by helping you organize and analyze your DNA matches and trees more effectively. **Clusters:**

AutoLineage assists in exploring your DNA Matches more efficiently in groups rather than as numerous individuals, and gain insights about branches in your family tree. **Common ancestors:** In addition, using trees that are associated to DNA matches and your profile, AutoLineage also aids in identifying common ancestors. Moreover, it works across DNA testing companies. **Locations:** Visualize locations linked to events of individuals from trees on a map. Track ancestral migration patterns, identify regional clusters, and correlate family movements with historical events.

Required steps

The following flow chart shows which steps are provided by AutoLineage:



Import shared matches

After importing your matches, import or set the shared matches. Shared matches are necessary for the next step, the clustering analysis.



....

Cluster matches

After importing the matches and shared matches, organize your DNA Matches into clusters of matches that likely descended from common ancestors.



Gather trees

Gather trees from various sites and use the tree management page to import these into AutoLineage application. To find back trees more easily, save the tree using a filename that contains the name of the linked DNA match as well as the shared cM.



Link trees to DNA matches

The next step is to link trees to DNA matches. Linking trees to DNA matches allows for the identification of common ancestors in these trees. Please visit the tree management page to link each tree to a DNA match individually, or use the wizard to perform this task more quickly.

Link profile to tree

Another recommended step is to link your profile to a tree. Linking a tree to a profile allows the common ancestor identification to identify common ancestors that are linked to your own tree and trees of other DNA matches. Please first import the tree linked to your profile using the tree management page, next visit the tree page linked to this profile and start the linking process.



More help sections on AutoLineage can be found online:

https://members.geneticaffairs.com/autolineage/

AutoKinship

At this moment, no information is available in this manual regarding AutoKinship. Please use the following blog post to get started on the AutoKinship tool for MyHeritage matches:

https://patriciacolemangenealogy.com/2021/10/07/autokinship/

Additional settings

More additional settings and links are available from the top-menu (see Figure 56). The different settings and options will be discussed below.



Figure 56. General settings from top-menu

- A) Members & Frontpage
 - 1) Members Home
 - 2) Genetic Affairs frontpage links
- B) Websites & Profiles
 - 1) Shows all websites
 - 2) Allows adding a new website
 - 3) Manage all profiles. This view shows all profiles for all websites. It can also be used to modify the settings for all profiles.
 - 4) This section will display all registered websites
- C) Settings
 - 1) User settings allows the modification of user settings with respect to user credentials, subscriptions and payment methods. The next section will go into more detail
 - 2) Subscribing or unsubscribing to the newsletter can be performed using this link
 - 3) This page shows all available credits
 - 4) When a valid credit card has been registered, it is possible to use this page to obtain credits based on a single charge
 - 5) Allows disabling all regular updates with one action
 - 6) Delete user account
- D) Contact us option (see the Contact us section below)
- E) Notifications pane and general announcements. The notifications panel will display account info updates, DNA match updates, information about single or monthly subscriptions that will be available as notifications. The general announcements will be used to show more general messages like new functionalities or promotional information.
- F) User settings allow changing information concerning your name, e-mail address, password, subscriptions, payment information, and invoices. Our next section will discuss this in more detail.

User settings and Payments

The user settings are available from the top-menu after selecting the user icon (most right option) and clicking on the "Your Settings" link (see Figure 56C). The different settings and options will be discussed below.

Settings		Contact Information
🕼 Profile	А	Name
■ Security	В	E-Mail Address
Billing		UPDATE
🚔 Subscription	С	
🚍 Payment Method	D	
Invoices	Е	

Figure 57. User settings

- A) Profile settings allows changing the name and e-mail address
- B) Allows changing the password
- C) Shows the different monthly subscription options (see Figure 76). The monthly subscriptions allow for a monthly addition of credits to your account. In addition, monthly subscription yields 10% bonus credits on top of the acquired credits.
- D) Store your credit card settings in this section. We use the Stripe (<u>www.stripe.com</u>) payment processing platform to store credit card information. In addition, the Stripe platform also performs monthly or single payments. Note that the VAT field only is required for companies, most users can ignore this option.
- E) Invoices from the monthly subscriptions can be downloaded from this invoices page.

O Free Trial										
You are currently within your free trial period. Your trial will expire on October 15th, 2018.										
Subscribe	Subscribe									
You are no	t subscribed to a plan. Ch	oose from the plans below to get started.								
All subscrip	tion plan prices are exclu	ding applicable VAT.								
Pro	습 PLAN FEATURES	\$5.00 / Monthly	SELECT							
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Pro	☆ PLAN FEATURES	\$7.00 / Monthly	SELECT							
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Pro	☆ PLAN FEATURES	\$14.00 / Monthly	SELECT							
Pro	습 PLAN FEATURES	\$15.00 / Monthly	SELECT							

Figure 58 - Subscription options

Blog posts and Facebook groups

After the launch of AutoCluster a new user group has been founded, please visit us on Facebook.

In addition, several (often AutoCluster related) blog posts have been created in the last year. These describe the use of AutoCluster and sometimes illustrate this tool with genealogical evidence.

Comparison of ICW AutoCluster and AutoSegment AutoCluster by Patricia Coleman Genetic Affairs Hybrid AutoSegment Cluster by Patricia Coleman **GEDmatch AutoSegment by Patricia Coleman** Genetic Affairs AutoFastClusters by Patricia Coleman Manual AutoClusters for LivingDNA by Patricia Coleman DNAeXplained - Genetic Affairs: AutoPedigree Combines AutoTree with WATO to Identify Your Potential **Tree Locations** Kitty Cooper - Automated tree building with Genetic Affairs DNAeXplained – Genetic Genealogy - Genetic Affairs Reconstructs Trees from Genetic Clusters – Even Without Your Tree or Common Ancestors Dana Leeds Blog Kitty Cooper's Blog - Automatic Clustering from Genetic Affairs Kitty Cooper's blog - More Clustering Tools! DNAeXplained – AutoClustering by Genetic Affairs Hartley DNA & genealogy - A New Look for AutoClusters Behold Genealogy - Genetic Affairs Clustering at 23andMe Anne's Family History - DNA: experimenting with reports from GeneticAffairs.com **DNAsleuth - Clustering Tools for DNA matches** Genea Musings - Using GeneticAffairs.com to Create DNA Match AutoClusters HistorTree - Analyzing DNA Auto-Clusters with Pedigree Collapse MyHeritage DNA - Introducing AutoClusters for DNA Matches The Genealogy Guys Blog - Genetic Affairs, a New DNA Tool

Matt's Genealogy Blog - Auto-Clustering of DNA Matches





Other AutoCluster implementations.

After the initial launch of AutoCluster in December 2018, a lot of requests were made concerning a version for MyHeritage. Fortunately, MyHeritage decided to license the tool and together with their team, we implemented the AutoCluster in their infrastructure. It was released during Rootstech 2019 in Salt Lake City. The visualization of AutoCluster from MyHeritage has been changed a bit, for instance, the usage of different cluster colors (see Figure 59). In addition, an algorithm was implemented that uses the results of various clustering analyses to find a clustering that produces a chart containing around 100 members. This ensures that the user experience will be more constant. Moreover, this clustering analysis also considers the shared cM between common matches. This implementation has been shown to improve the clustering results of people from <u>endogamous</u> populations (for instance, Ashkenazi or Acadian).



Figure 59. AutoCluster implementation from MyHeritage. Note that the current implementation does not contain the sorting of clusters based on the grey cells, instead, they are sorted based on cluster size.

Another website that now employs the AutoClustering algorithm and visualization is GEDmatch (see Figure 60). This analysis is available for Tier 1 users. GEDmatch has gone a step further by creating an interactive AutoCluster analysis by combining the Multi Kit Analysis with the AutoCluster clustering. As a result, members of a cluster can now be selected for further analysis, for instance, to see which DNA segments are shared.



Figure 60. GEDmatch implementation of the AutoCluster algorithm with selectable clusters and members for downstream analysis by the MKA.

Prices

The free trial provides a total of 200 credits that can be used to try several features (see Figure 61). By purchasing a subscription on our site, several premium features are unlocked as well as the removal of some restrictions.



Figure 61. Free trial vs subscriber membership

The front page holds information concerning the costs of the analyses (see <u>https://www.geneticaffairs.com/prices.html</u> and Figure 62).

What do analyses on Genetic Affairs cost?

- Reclustering, CSV or AutoFastCluster analyses are 50 credits.
- A default AutoCluster analysis using shared matches for FTDNA is 75 credits per search.
- An AutoSegment analysis costs **75** credits per search.
- A manual AutoKinship analysis costs **25** credits per search.
- Rule based analyses* are 50 credits per applied rule and 50 credits for the primary profile
- AutoCluster analyses with the AutoTree or AutoPedigree*. features are 100 credits.
- Hybrid AutoSegment clustering costs 100 credits for 2 datasets*.
- Hybrid AutoSegment clustering costs 125 credits for 3 datasets*.
- Hybrid AutoSegment clustering costs 150 credits for 4 datasets*.
- · AutoLineage requires an active subscription*.
 - * premium feature

Figure 62. Cost of the analyses

Troubleshooting

Sometimes the websites (FTDNA or 23andme) are unreachable, for instance, if they are under maintenance. In this case, a message concerning this error will be mentioned in the regular mail. In addition, in the case of a weekly or monthly search, the delay is saved. When the upcoming search is successful, we use this delay to correct the weekly or monthly interval (which would otherwise shift by one or more days).

In the case of endogamous populations, the AutoCluster might not work properly. This is partly due to the large number of matches which a long time to download. The limited-time slot of a single AutoCluster analysis does not provide for these long downloads. In some cases, selecting the more powerful server will help. If it still doesn't work, alternative methods such as <u>DNAGedcom</u> might be a good option since they also perform clustering of the matches based on shared matches. The difference, however, is that this tool can run on your own computer and doesn't suffer from the time constraints that AutoCluster has.

See our <u>frequently asked questions</u> for more information concerning security.

Contact us

In case of questions, please use the contact form that can be in the right top corner of the members page. Or visit this page directly: <u>https://members.geneticaffairs.com/contactus</u>

The contact page requires your email address and name as well as the question. Last, to prevent spammers, enter the second name of our company in lower case letters under the secret question. The answer to that question is: **affairs**

	Members & Frontpage 🔻	Websites & Profiles 🔻	Analyses 👻	Settings 🔹	Contact us 👻	1 4	ejblom 👻
Contact	Form						
	Please visit our FAQ pay Questions regard	for more information ments and invoking the ing the interpretation suited for our new	concerning p he AutoCluste of the AutoCl	rivacy, securit er analysis. luster analysis	y, credit card are better		
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johndoe@gmail.co	m						
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I have a question re	garding						
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Please provide secre	et code: enter the second wor	d of our company name in	1 lowercase				10
affairs							
Contact US!							10

Figure 63. Contact us page