

22<sup>nd</sup> Conference of the International Society of Forensic Genetics

Pre-congress seminar: mtDNA typing - EMPOP

Copenhagen, August 20<sup>th</sup>, 2007

---



---

# EMPOP Release 2 Towards Standardization on the Use of Haplogroups in Forensic Science

Walther Parson<sup>1</sup>, Anita Brandstätter<sup>1</sup>, Alexander Röck<sup>2</sup>

<sup>1</sup> Institute of Legal Medicine, Innsbruck Medical University

<sup>2</sup> Institute of Mathematics, University of Innsbruck

Austria



# EMPOP Network Analysis



Welcome to MyEMPOP!  
[Logout](#)

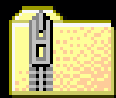
[MYEMPOP](#) | [QUERY](#) | [NETWORK](#) | [DOWNLOADS](#) | [CONTRIBUTORS](#) | [HELP](#)

[MyProfile](#)  
[MyHistory](#)

[AllHistory](#)  
[AllUsers](#)

## Downloads

File	Info
<a href="#">DNW.zip</a>	Tool for drawing quasi-median networks
<a href="#">AUT273_spec.emp</a>	emp example file



dnw.zip



dnw.exe



glut32.dll

C:/Network/



AUT273\_spec.emp

Microsoft Excel - AUT273\_spec.emp

O2

	A	B	C	D	E	F	G
1	# This is an example for the emp format. Find details in Brandstätter et al. (2006) FSI doi:10.1016/j.for						
2	# Ambiguous positions (sequence heteroplasmy) are resolved by reporting the dominant sequence v						
3	# Sample name	# Haplogroup	# Number	# Polymorphisms			
4	AUT0500000	K1a	1	73G	263G	309.1C	315.1C
5	AUT0500001	?	1	235G	263G	309.1C	309.2C
6	AUT0500002	H*	1	263G	309.1C	309.2C	315.1C
7	AUT0500003	J1c	1	73G	185A	188G	228A
8	AUT0500004	J1c	1	73G	185A	189G	228A
9	AUT0500005	H*	1	263G	315.1C	340T	523DEL
10	AUT0500006	K2b	1	73G	151T	152C	199C
11	AUT0500007	U5b	1	73G	150T	263G	315.1C
12	AUT0500008	H*	1	73G	187T	263G	315.1C
13	AUT0500009	H*	1	189G	263G	315.1C	16519C
14	AUT0500010	H1c	1	263G	315.1C	477C	16093C
15	AUT0500011	H5	1	146C	195C	263G	309.1C
16	AUT0500012	U4	1	73G	150T	195C	263G
17	AUT0500013	H1c	1	263G	309.1C	315.1C	477C



# Executing EMPOP Network



MyProfile  
MyHistory

AllHistory  
AllUsers

## NETWORK - STEP 1 OF 4

Please specify the program you wish to execute. Currently only network is available.

☒ [Network analysis](#)

GO TO STEP 2



MyProfile  
MyHistory

AllHistory  
AllUsers

## NETWORK - STEP 2 OF 4 - DATAFILE & PARAMETERS

Please specify the input file and the parameters for network.  
If you wish to view the filters click on the following link:

[view filtered positions](#)

[Input file](#)

Durchsuchen...

### Network analysis - Parameters

[Filter](#)

[Region](#)

GO TO STEP 3

Filter files

[EMPOPspeedy](#)

[EMPOPall](#)

[unfilt](#)

16051	A>R	G>R	#	A
16078	A>R	G>R	#	A
16086	C>Y	T>Y	#	T
16092	C>Y	T>Y	#	T
16093	C>Y	T>Y	#	T
16111	C>H	T>H	A>H	# C
16114	C>Y	T>Y	#	C
16124	C>Y	T>Y	#	T
16126	C>Y	T>Y	#	T
16129	A>R	G>R	#	G
16140	C>Y	T>Y	#	T
16145	A>R	G>R	#	G
16147	C>Y	T>Y	#	C
16148	C>Y	T>Y	#	C
16150	C>Y	T>Y	#	C
16163	A>R	G>R	#	A
16166	A>a	->a	#	A
16172	C>Y	T>Y	#	T



# Executing EMPOP Network



MyProfile  
MyHistory

AllHistory  
AllUsers

## NETWORK - STEP 3 OF 4 - CHECK INPUT

Please check your input from the previous step.  
Press "SUBMIT INPUT" if you wish to execute network.  
Press "EDIT INPUT" if you wish to change parameter(s) and/or input file.

Input file AUT273\_spec.emp

### Parameters

Filter EMPOPspeedy

Region 16024-16569



MyProfile  
MyHistory

AllHistory  
AllUsers

## NETWORK - STEP 4 OF 4 - RESULT

Thank you for using network.  
It took 2 updates  $\approx$  16 seconds.

Use the link below to download the result file.

[20070811074722\\_AUT273\\_spec\\_EMPOPspeedy\\_16024-16569.zip](#)  
(zip, 8.581 kb)

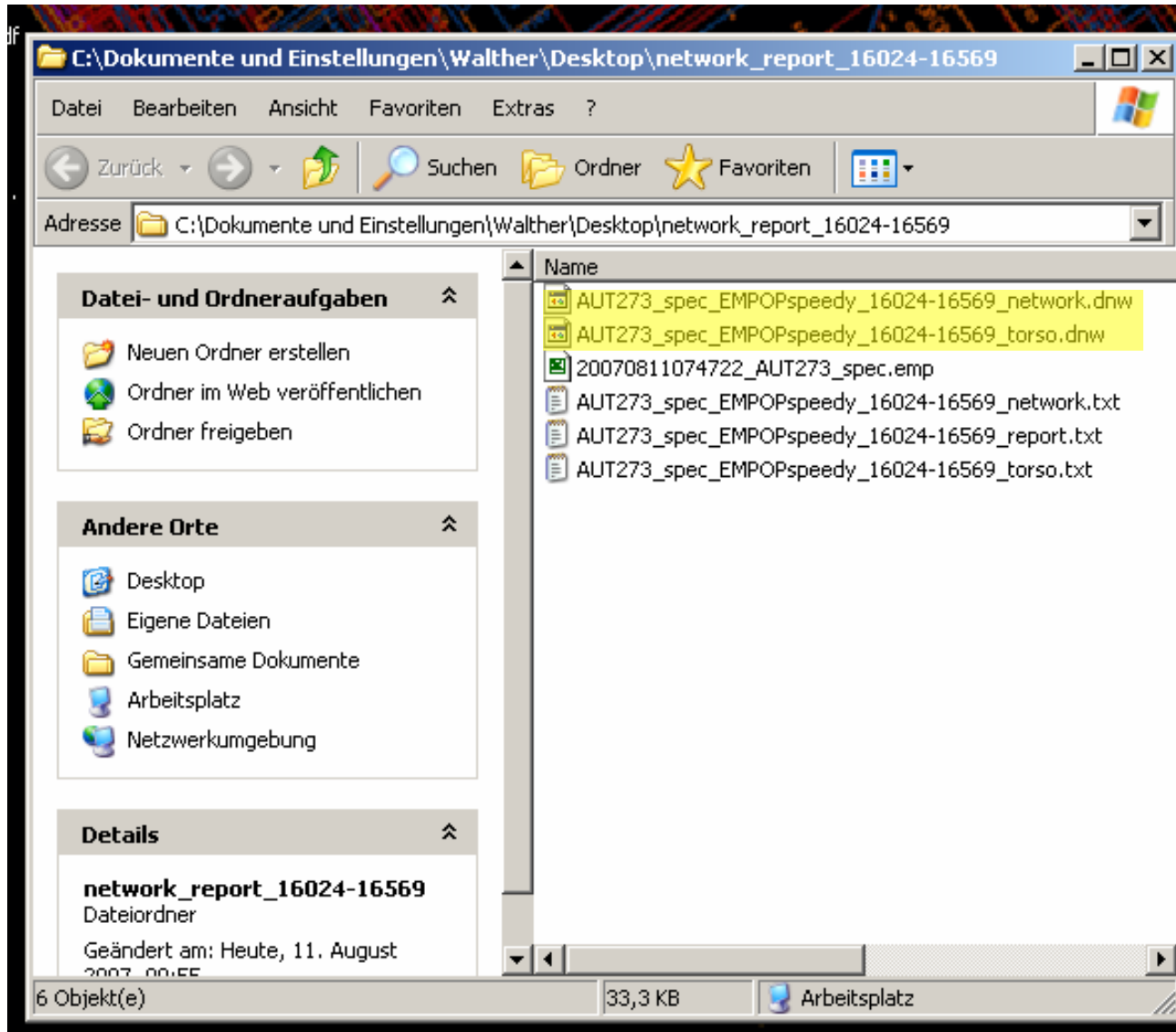


20070811074722\_A...

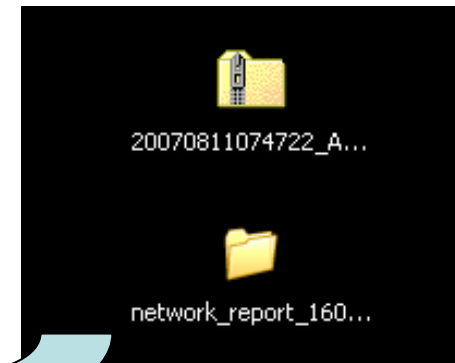


network\_report\_160...

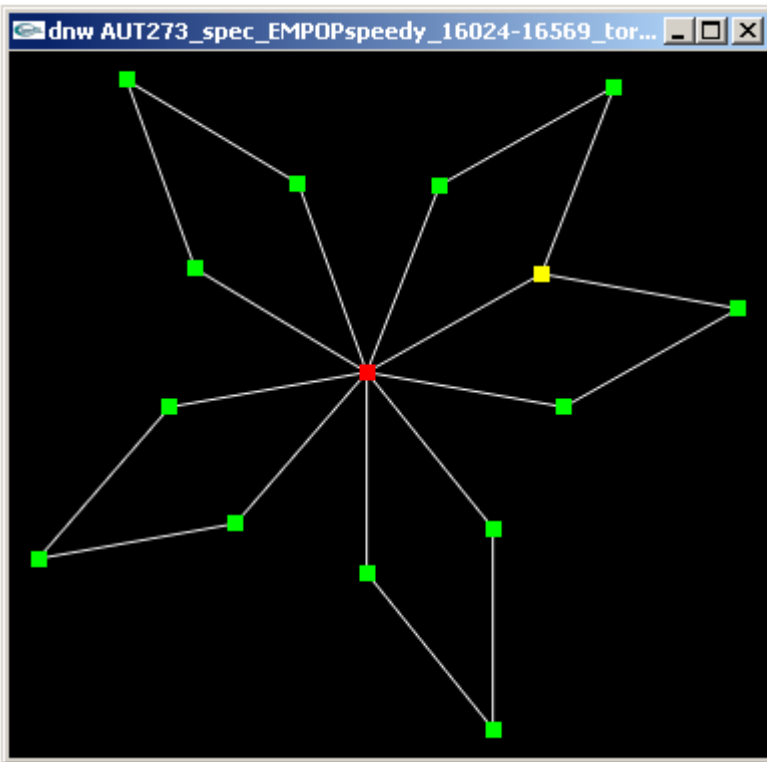
# Drawing the Network



C:/Network/



# Drawing the Network – Torso HVS-I



```
C:\WINDOWS\system32\cmd.exe - dnm AUT273_spec_EMPOPspeedy_16024-16569_torso

C:\Network>dnm AUT273_spec_EMPOPspeedy_16024-16569_torso
Screen resolution 1400x1050
Use left/right mouse button to move nodes/edges or press
escape key to exit,
directional keys to translate network,
page up/down key to zoom,
home directional key to reset,
b for bounding box,
c/C for scaling circles,
d/D for scaling dots,
e for writing EPS to <datasetname>.eps,
E for writing EPS to <datasetname>.eps with title,
f/F for scaling fontsize,
g for writing FIG to <datasetname>.fig,
G for writing FIG to <datasetname>.fig with title,
l for displaying labels,
m for displaying medians,
o/O for scaling origin,
q/Q for scaling quads,
r/R for rotating (in initial position),
s for saving new layout to <datasetname>.dnw,
t for draft/drawing.
```

**Ausführen**

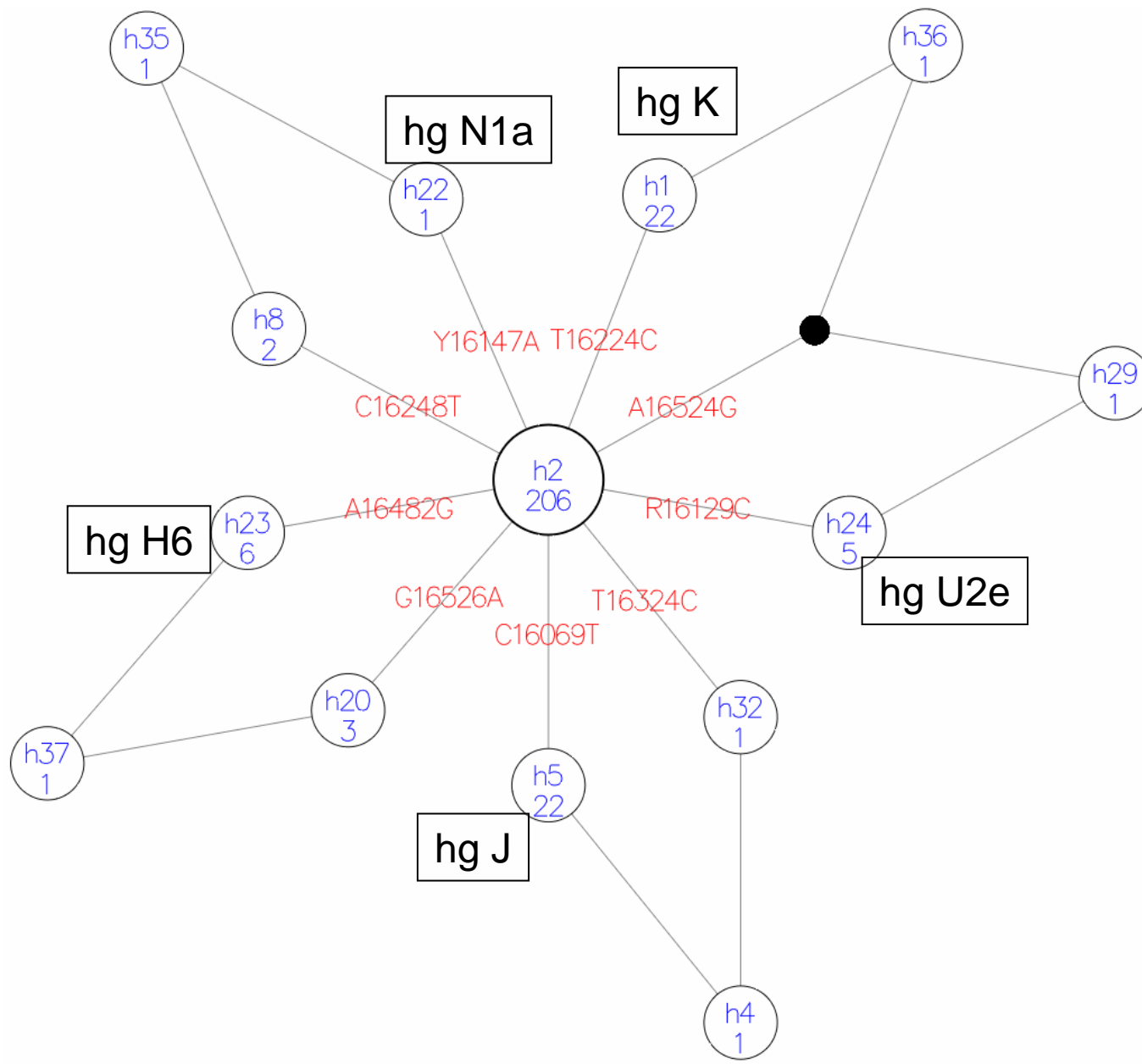
Geben Sie den Namen eines Programms, Dokuments oder einer Internetres...

Öffnen:

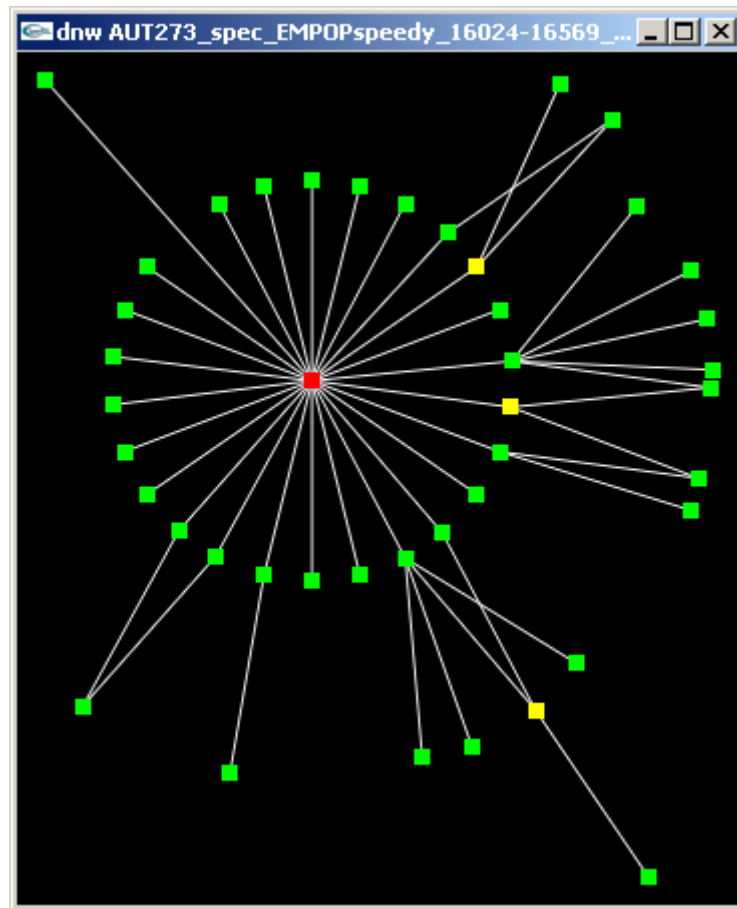
```
C:\WINDOWS\system32\cmd.exe

C:\Network>dnm AUT273_spec_EMPOPspeedy_16024-16569_torso
```

# HVS-I Torso Interpretation



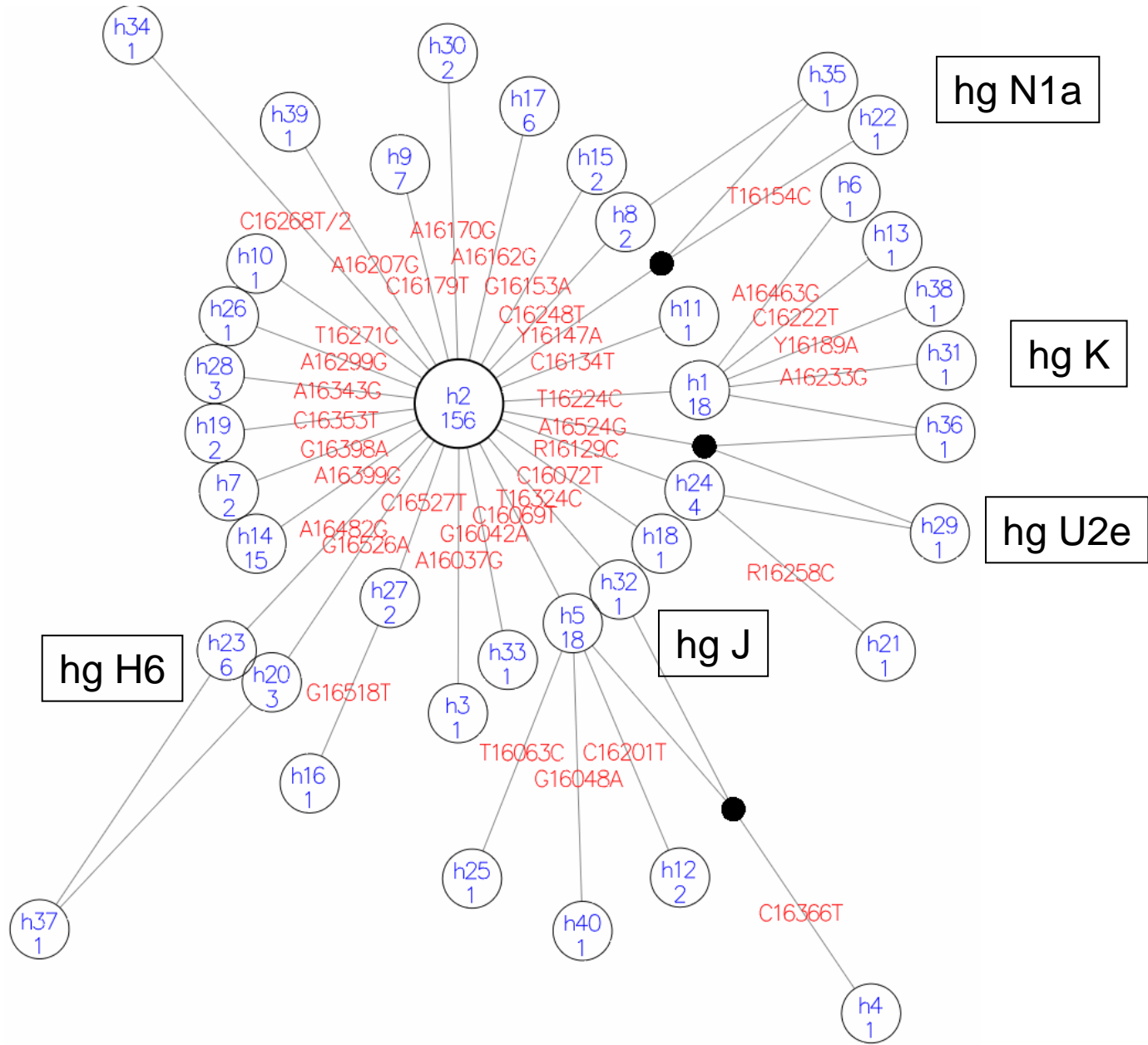
# Drawing the Network - entire HVS-I Network



```
C:\Network>dnw AUT273_spec_EMPOPspeedy_16024-16569_network_
```



# HVS-I Network Interpretation

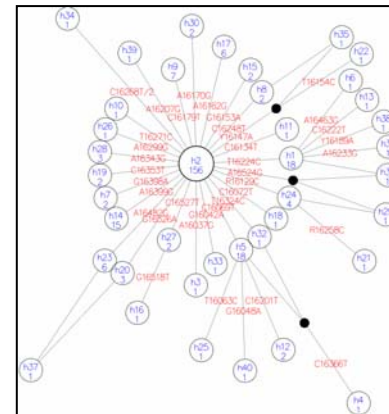
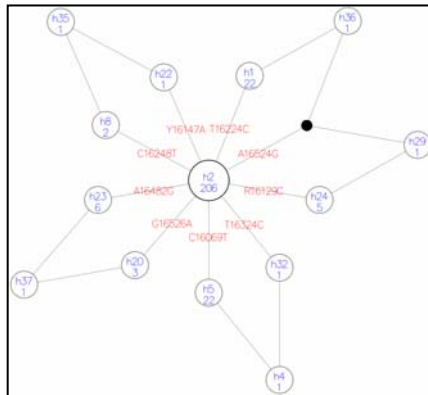


# Torso - Network

**torso** – network without the individual haplotypes; rough overview, less complex

**network** – contains all unfiltered mutations; harbours all most parsimonious reconstructions for any tree

Highlights clerical errors, phantom mutations, unusual alignment, reference bias, wrong reference sequence

Available online at [www.sciencedirect.com](http://www.sciencedirect.com)

ScienceDirect

Molecular Phylogenetics and Evolution 42 (2007) 256–271

MOLECULAR  
PHYLOGENETICS  
AND  
EVOLUTION

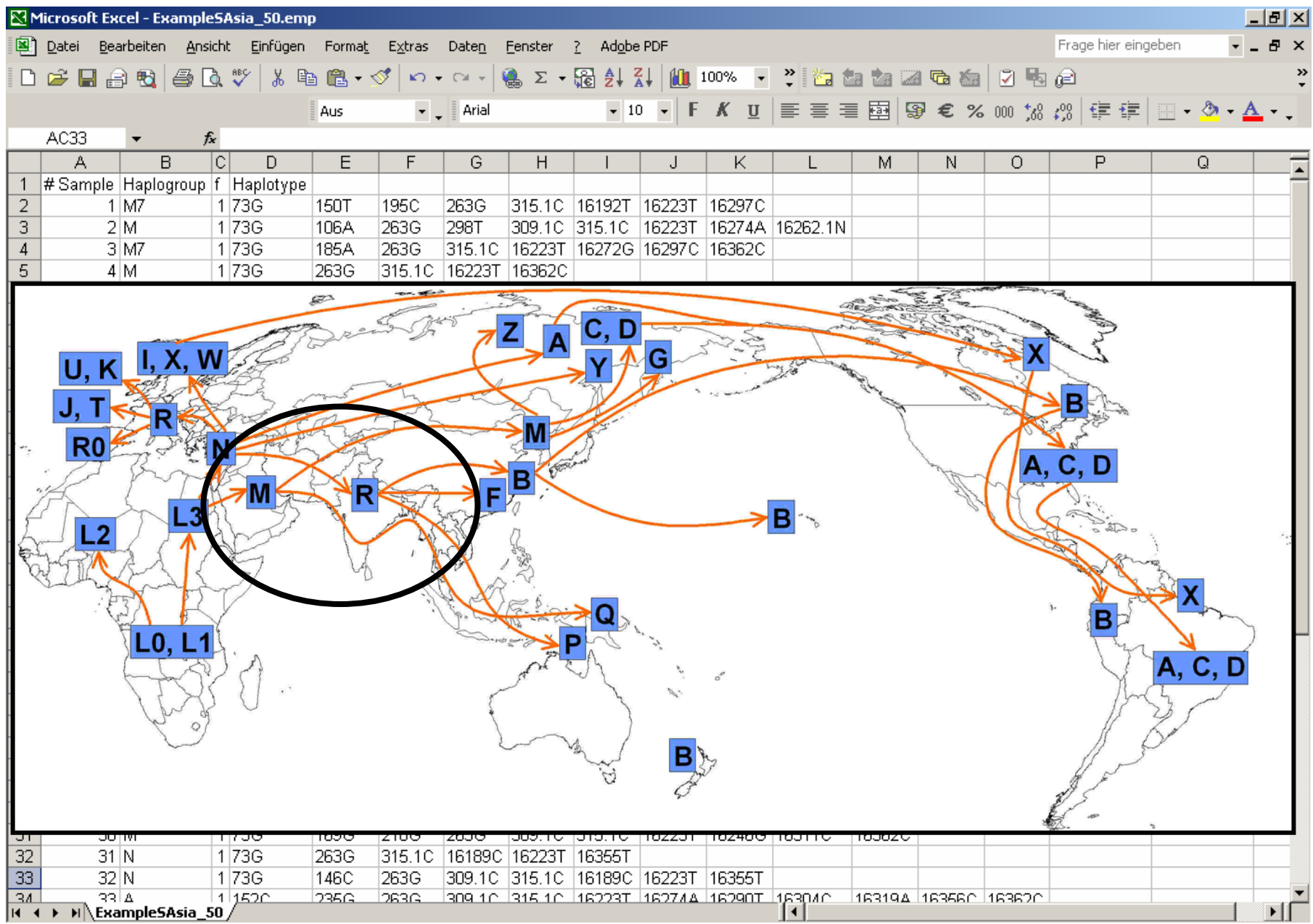
[www.elsevier.com/locate/ympev](http://www.elsevier.com/locate/ympev)

# Translating DNA data tables into quasi-median networks for parsimony analysis and error detection

Hans-Jürgen Bandelt <sup>a,\*</sup>, Arne Dür <sup>b</sup>



# Let's test an example – 50 sequences from South Asia



# HVS-II Torso

## Report...

## Haplotype designation in the network:

h1: 1 3 4 6 8 9 11 12 15 16 21 24 29 31 32 33 34 35 36 37 41 45 46 47 48 49 50

## h2: 2

### h3: 5 7

#### h4: 10 14 27 28

## h5: 13 19 20 23 39 42 44

## h6: 17

## h7: 18

## h8: 22

## h9: 25

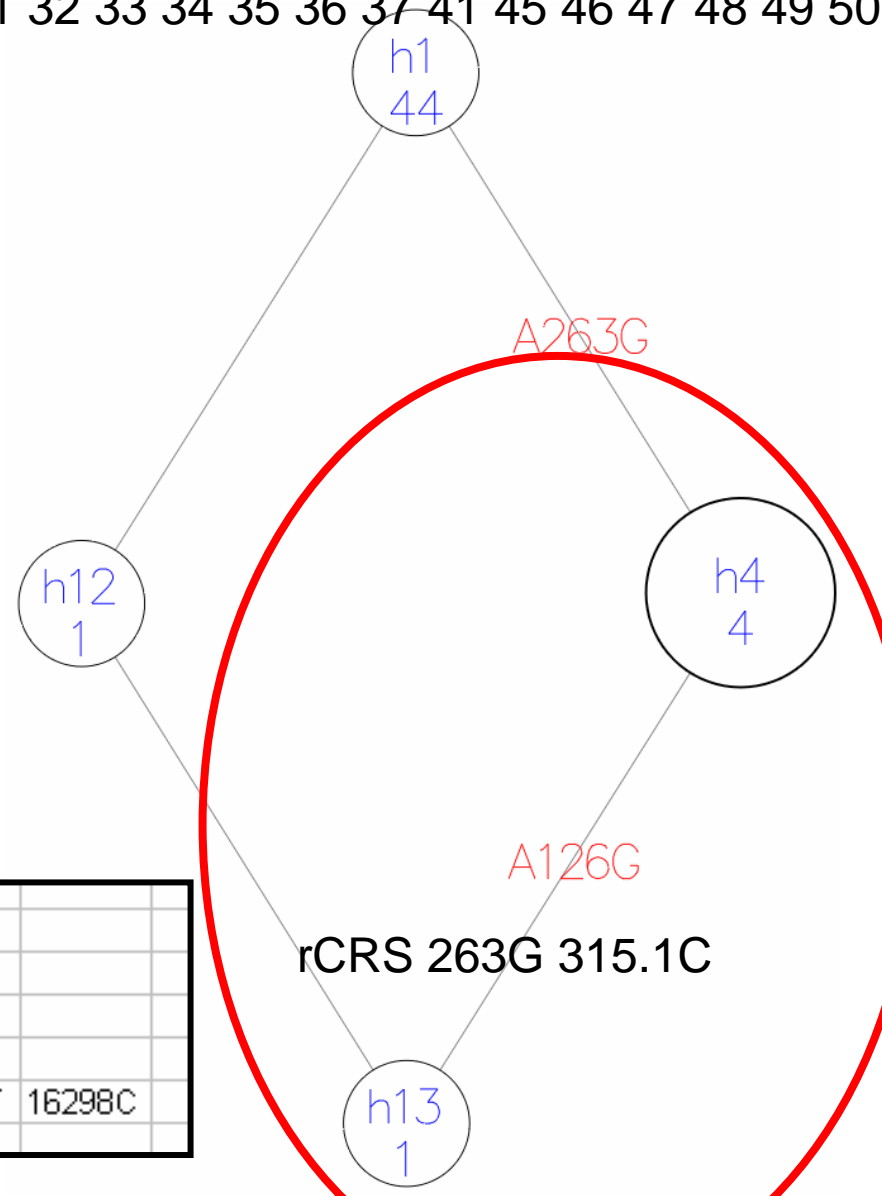
# h10: 26

# h1 1: 30

# h12: 38

# h13: 40

# h14: 43



emp-file

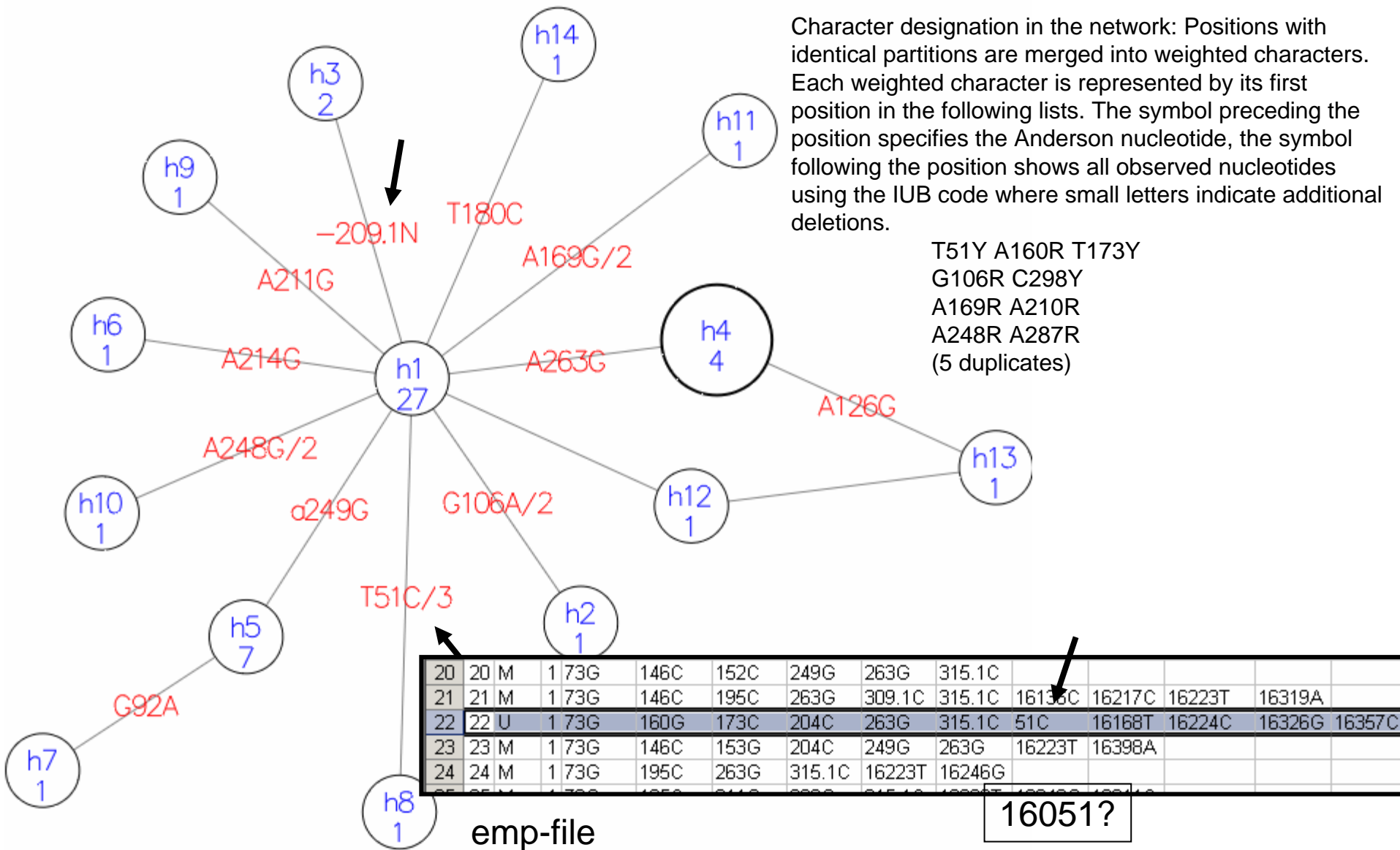
[illegible]

# HVS-II entire Network


Network legends:

Character designation in the network: Positions with identical partitions are merged into weighted characters. Each weighted character is represented by its first position in the following lists. The symbol preceding the position specifies the Anderson nucleotide, the symbol following the position shows all observed nucleotides using the IUB code where small letters indicate additional deletions.

T51Y A160R T173Y  
G106R C298Y  
A169R A210R  
A248R A287R  
(5 duplicates)



# Detailed analysis – unfiltered – 1-100



MyProfile  
MyHistory

AllHistory  
AllUsers

## NETWORK - STEP 2 OF 4 - DATAFILE & PARAMETERS

Please specify the input file and the parameters for network.  
If you wish to view the filters click on the following link:  
[view filtered positions](#)

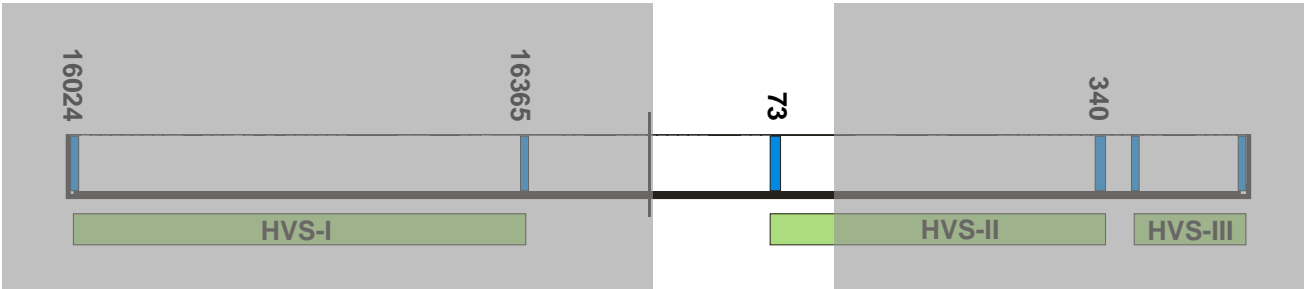
**Input file**

**Network analysis - Parameters**

**Filter**

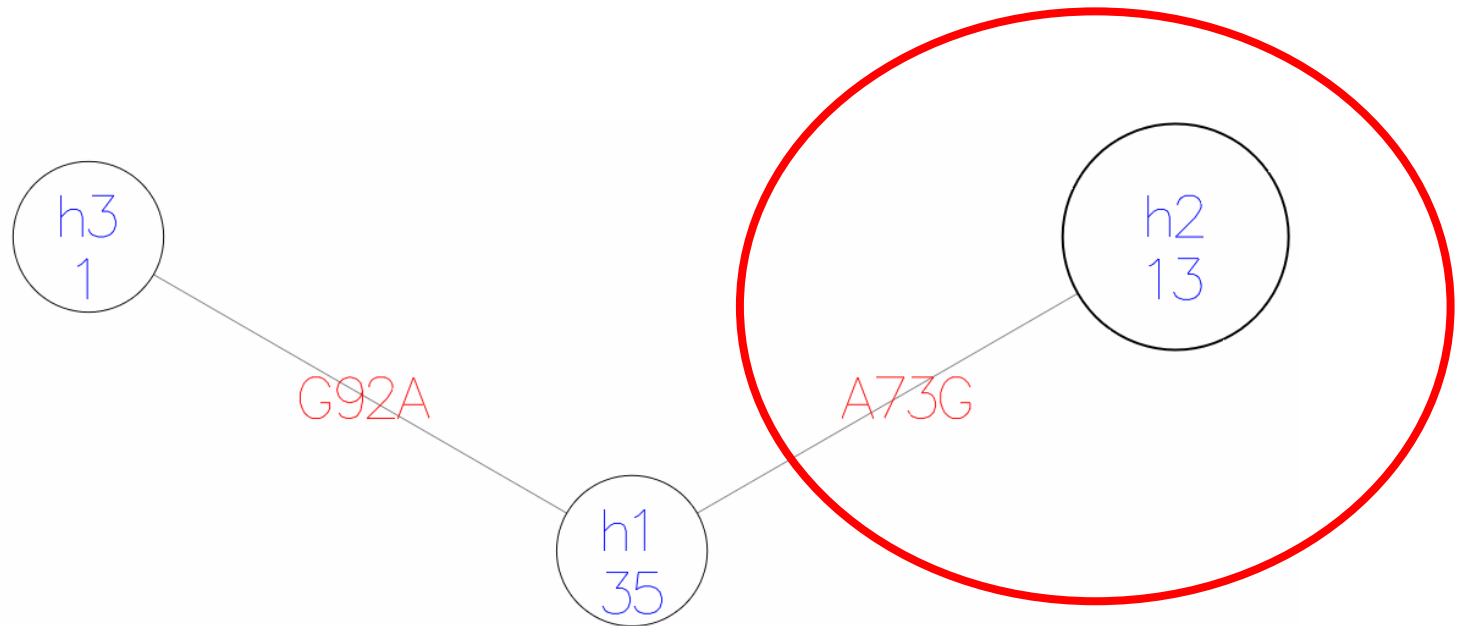
**Region**

1 - 100



The diagram illustrates the HVS (Hypervariable Segment) regions and positions. It shows a horizontal line with three segments labeled HVS-I, HVS-II, and HVS-III. HVS-I is on the left, HVS-II is in the middle, and HVS-III is on the right. Vertical lines indicate specific positions: 16024 and 16365 on HVS-I, 73 on HVS-II, and 340 on HVS-III. A bracket above the line indicates the range 1 - 100.

## Detailed analysis – unfiltered – 1-100



## Report

Haplotype designation in the network:

h1: 1 2 3 4 5 6 7 8 9 11 12 13 16 17 19 20 21 23 24 25 26 29 30 31 32 41 42 43 44 45 46 47 48 49 50

h2: 10 14 15 27 28 33 34 35 36 37 38 39 40

[illegible]



# EMPOP bioinformatics chart

