



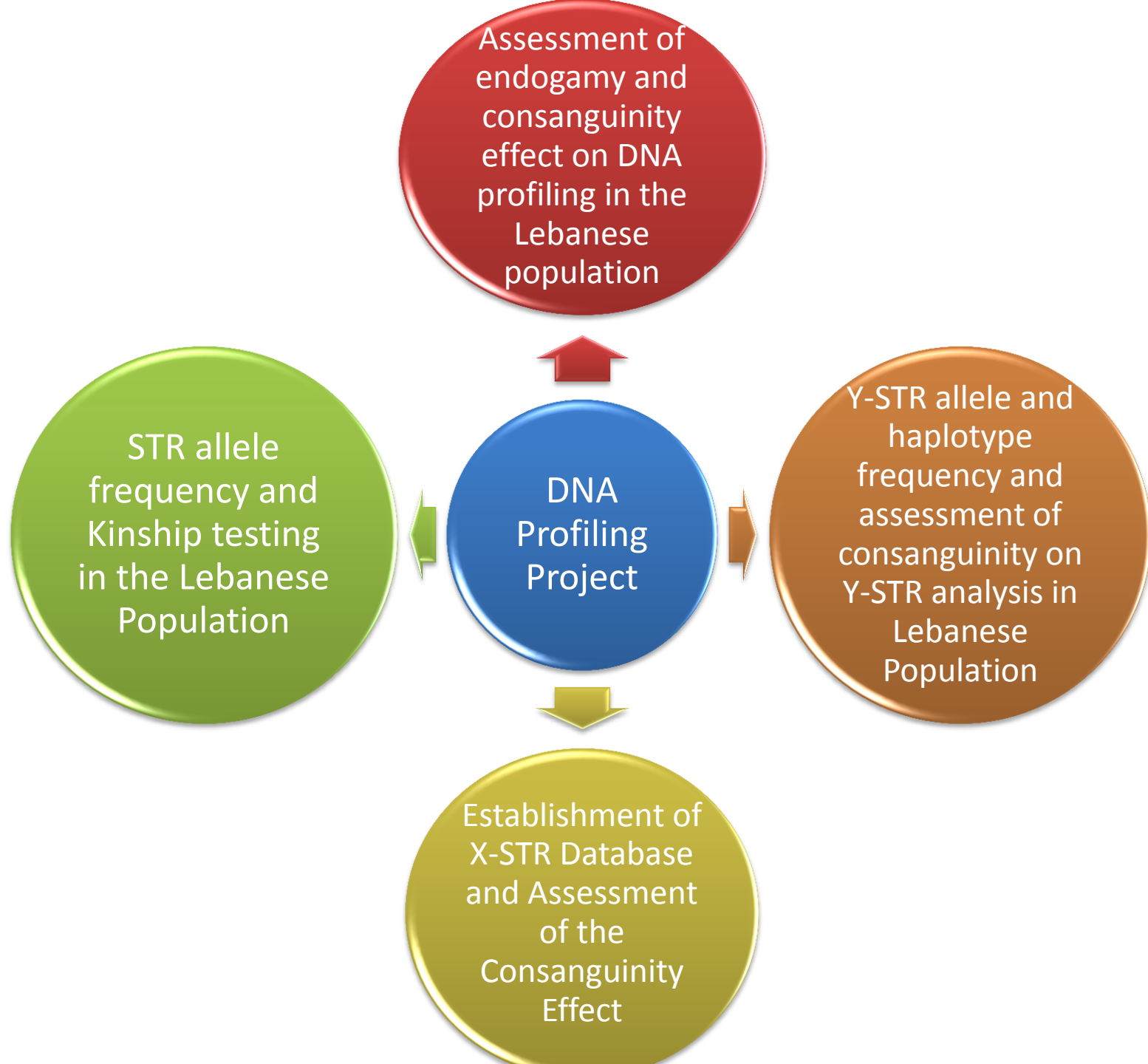
# **PowerPlex® Y23 discriminating power in stringent endogamous and consanguineous situations**

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Beirut Lebanon***

***Promega Webinar  
July 12, 2012***

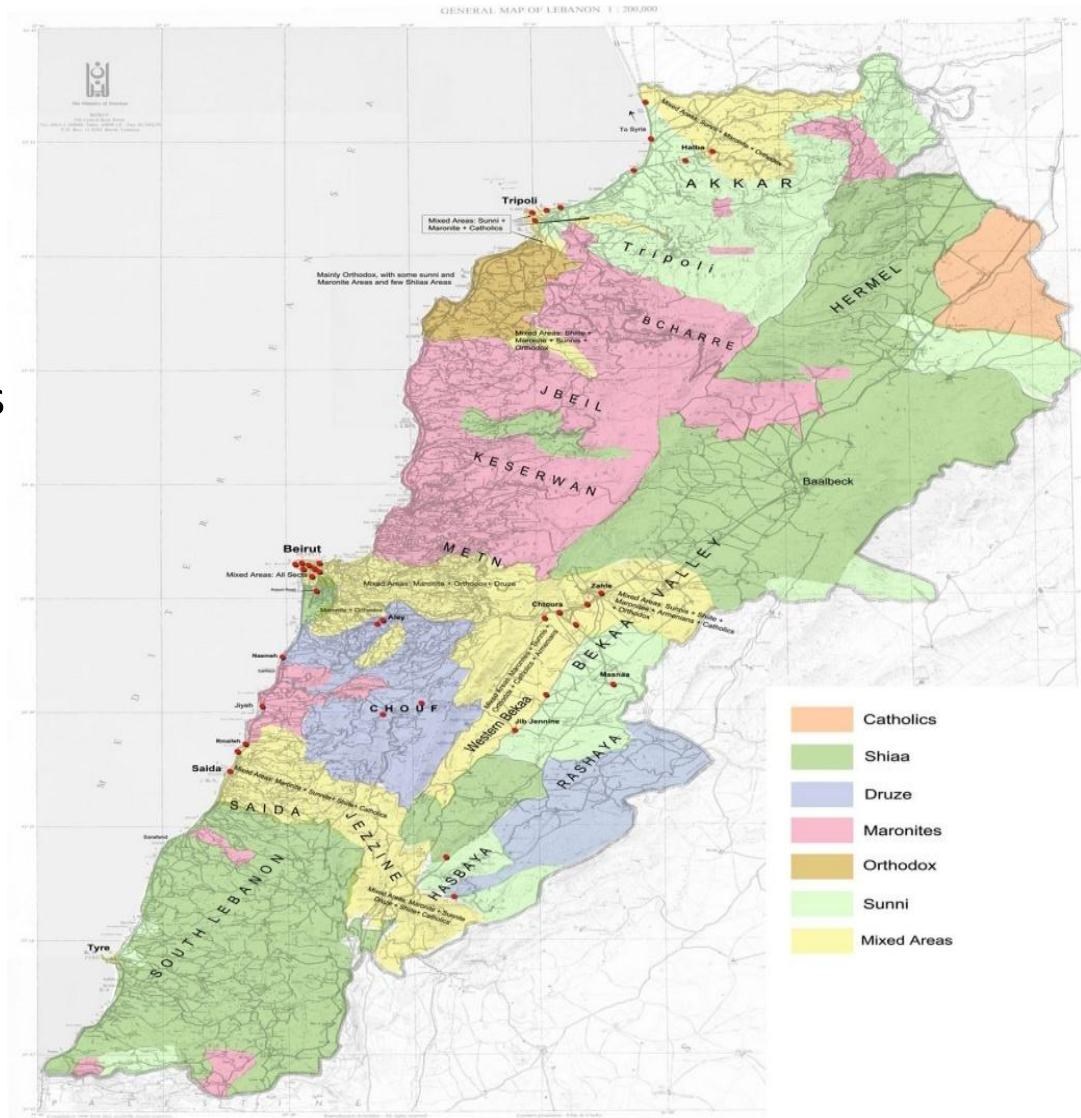


# Lebanon

- Isolated sub-populations:  
**18 Religion communities:**
  - **60.3% Muslim**
  - **39.7% Christian**
- No Y-STR frequency database has been done previously.
- High rate of consanguinity and endogamy:

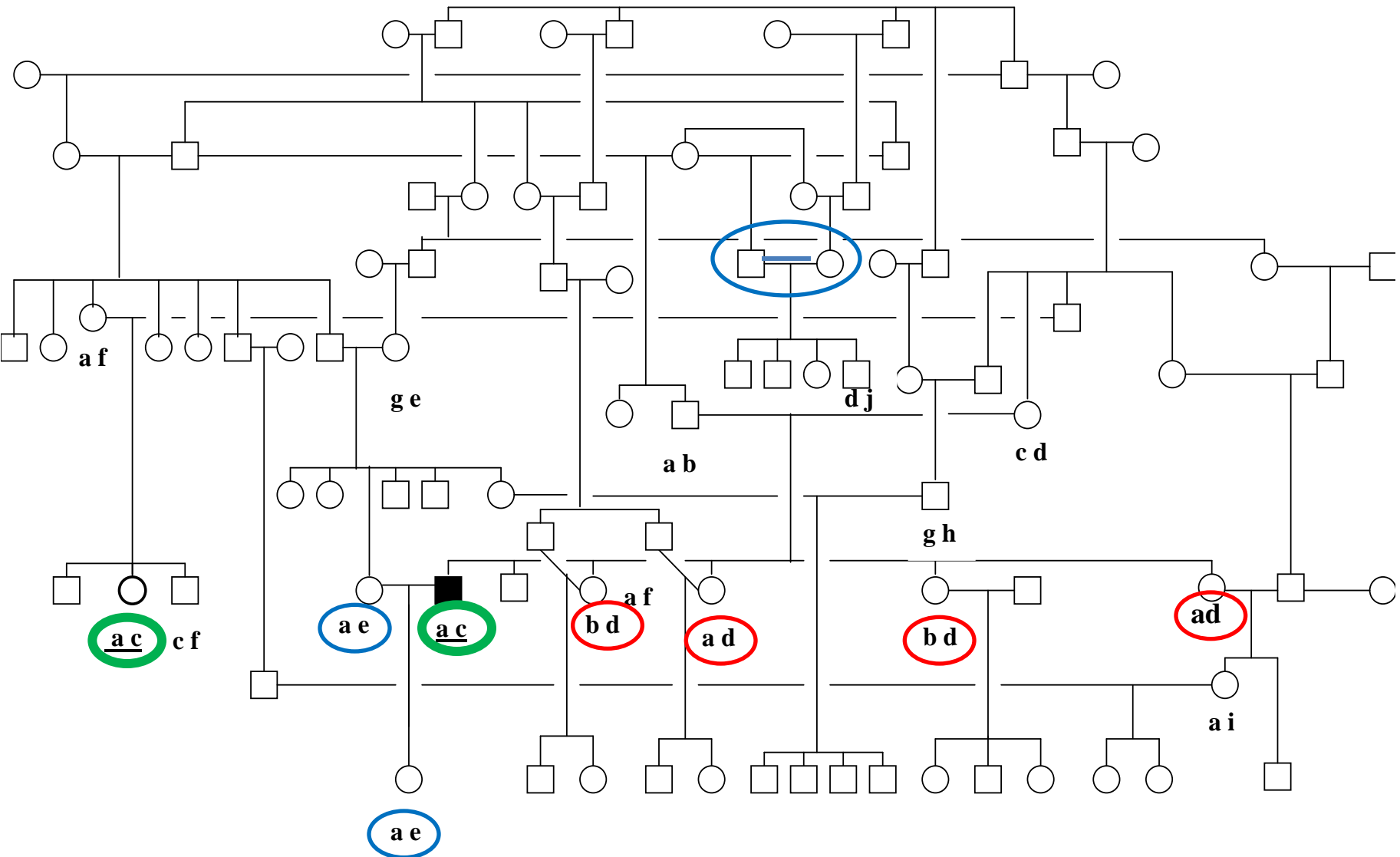
**Average endogamy rate: 88 %**

**Consanguinity rate: 31-35%**



# An Example of Genetic Effect of Consanguinity

## HLA matching for Bone Marrow Transplantation





# Example of Genetic Seclusion

## Familial Mediterranean Fever

FMF in Lebanon  
I Mansour *et al*

Table 2 Allele frequency<sup>1</sup> among the Lebanese communities

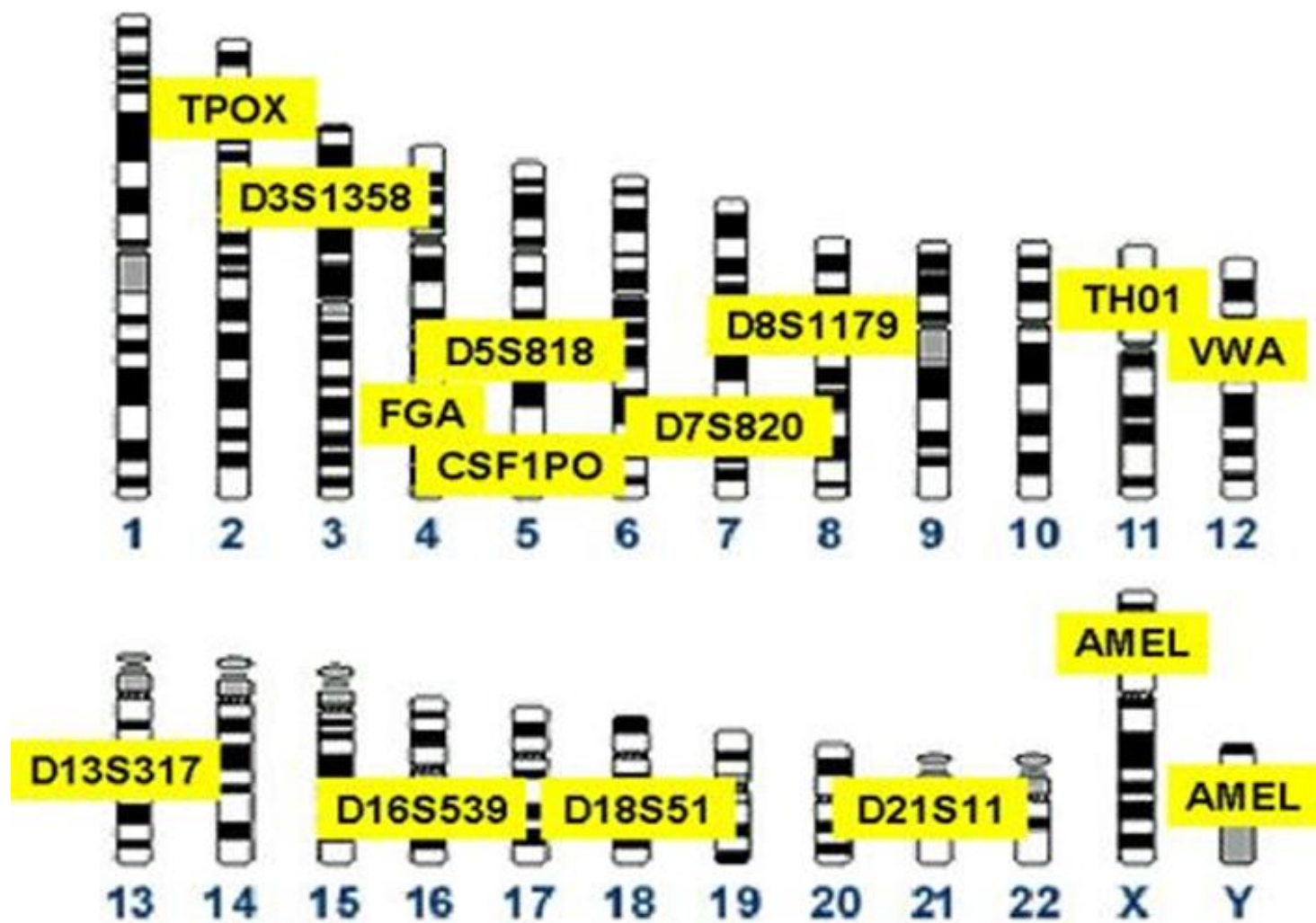
		M694V %	V726A %	M694I %	E148Q %	M680I %	None %
Total alleles	n = 143	27	20	9	8	5	33
Total Moslem <sup>2</sup>	n = 75	32	19	12	9	5	25
Total Christian <sup>2</sup>	n = 64	22	22	6	5	5	41
Druze <sup>2</sup>	n = 9	0	11	56	33	0	0
Chiite <sup>2</sup>	n = 28	50	11	11	0	4	21
Sunnite <sup>2</sup>	n = 36	25	22	0	8	8	36
Armenian <sup>2</sup>	n = 12	83	0	0	0	8	8
Maronite <sup>2</sup>	n = 16	0	13	6	0	6	75
Greek Orthodox <sup>2</sup>	n = 8	0	25	37	13	0	25
Greek Catholic <sup>2</sup>	n = 6	0	50	0	0	17	33
Syriac <sup>2</sup>	n = 6	33	67	0	0	0	0

<sup>1</sup>Only one of the allele was counted in patients homozygous by descent. The E167D and F479L mutations were found in one patient showing the V726A/E167D/F479L genotype but alleles were not determined. The K695R was also found in one patient; <sup>2</sup>Patients with parents of different origins are not shown.

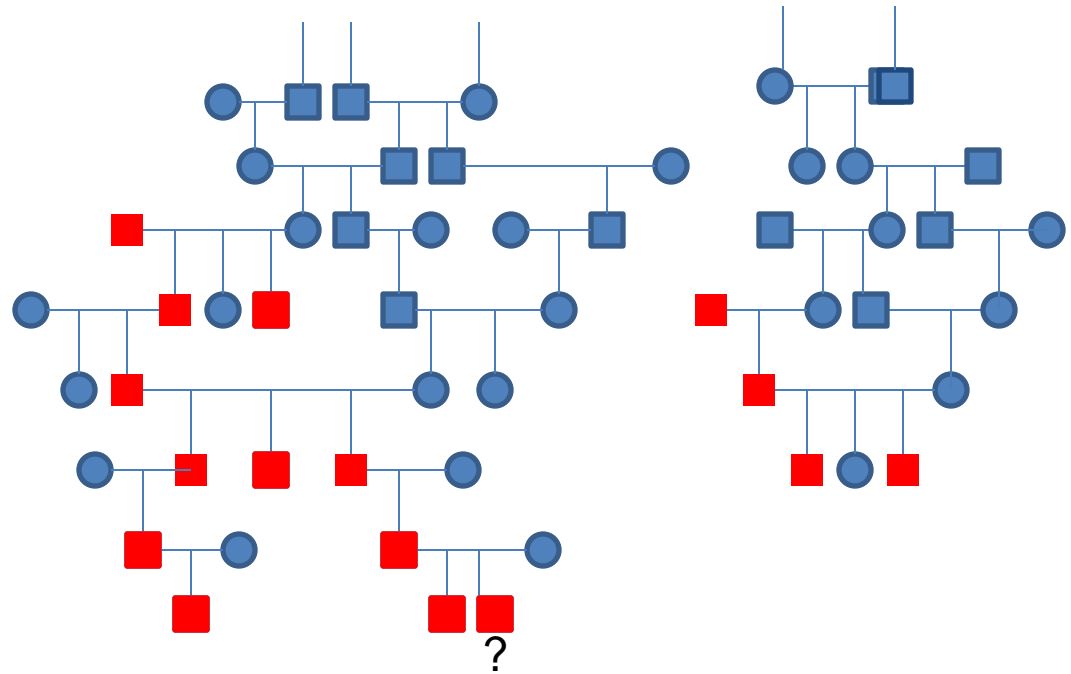
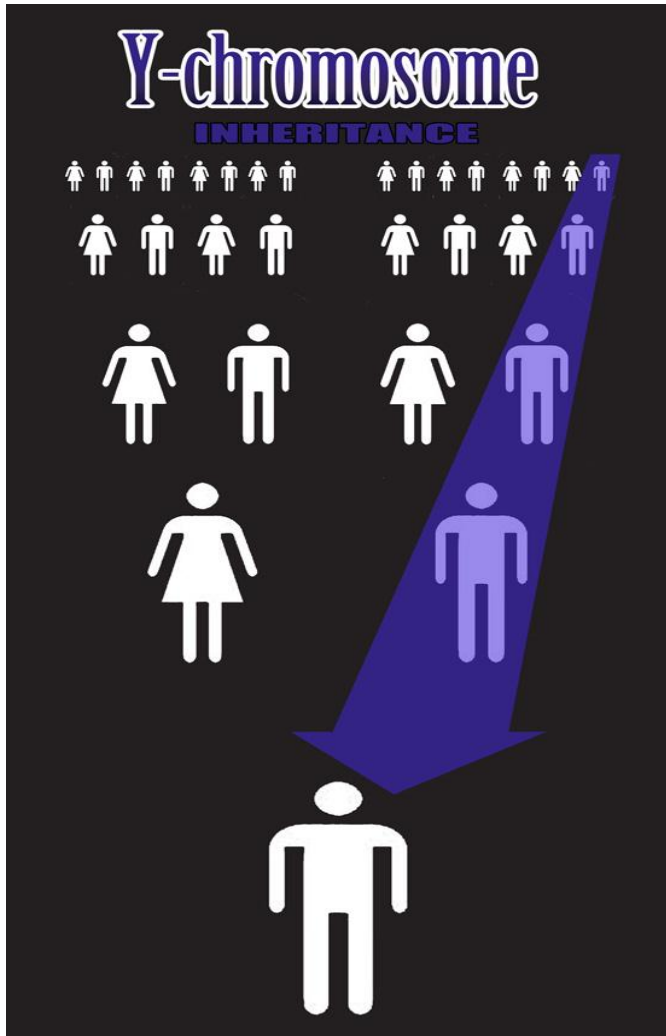
## Effect of Consanguinity and Endogamy

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- In specific populations, genetic relatedness can be very pronounced.
- Increases the chances of finding a DNA profile that might represent more than one individual.



# Limitations: Inherited in a Patrilineal manner



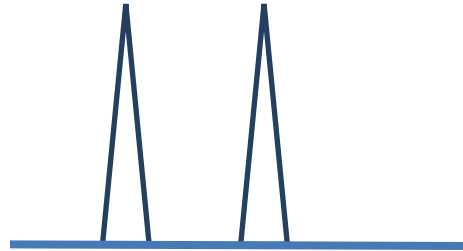


# Masking of male autosomal DNA

Using Autosomal STR

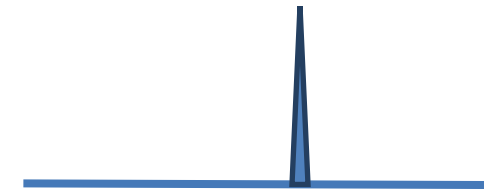
Using Y-chromosome STR

Female (victim) DNA  
Profile

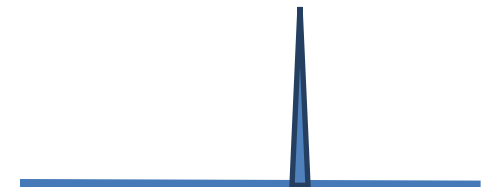
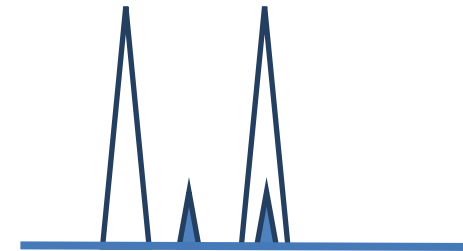


NO SIGNAL

Male (perpetrator) DNA  
profile



DNA profile from crime  
scene



## Rape Case Encountered: Y-Profile

System	Vaginal Swab	Suspect 1	Suspect 2	Suspect 3
DYS 456	15	15	14	15
DYS 389I	14	14	13	14
DYS 390	25	25	22	25
DYS 389II	32	32	29	32
DYS 458	17	17	15	17
DYS 19	14	14	16	14
DYS 385 a/b	16-17	16-17	12-13	16-17
DYS 393	13	13	13	13
DYS 391	10	10	10	10
DYS 439	12	12	13	12
DYS 635	21	21	21	21
DYS 392	11	11	11	11
Y GATA H4	11	11	11	11
DYS 437	14	14	14	14
DYS 438	10	10	10	10
DYS 448	19	19	20	19

## Specific Aim

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### Target Solutions:

- Establish Y-STR allele and **haplotype** frequencies for Lebanese Population.
- Assessment of **Consanguinity** and **Endogamy** effect on Y-STR analysis.

# Sample Distribution

502 unrelated male individuals representative of the Lebanese population

	<b>Expected</b>	<b>Tested</b>
- Muslim Sunnite	27.2%	28.1%
- Muslim Shiite	26.8%	27.1%
- Christian Maronite	21.4%	21.7%
- Christian Catholic	5.0%	5.2%
- Christian Orthodox	7.4%	5.8%
- Druze	5.7%	6.3%
- Armenian Orthodox	2.8%	3.2%
- Armenian Catholic	0.8%	0.6%
- Alawite	0.8%	0.4%
- Others	2.0%	1.6%

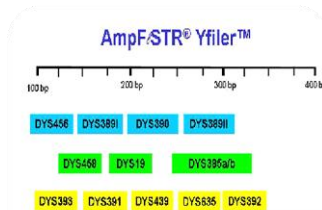
Ministry of Interior and Municipalities (2009)

# Experimental Design



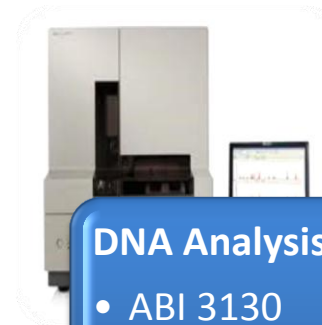
## DNA Extraction

- PCI
- Salting Out



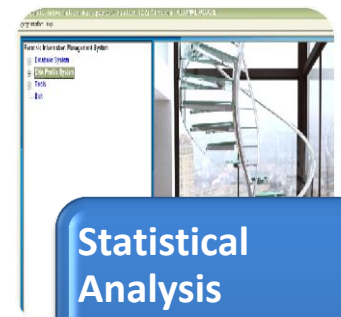
## PCR multiplex:

- Y-filer ABI (17 systems)
- Y-23 PowerPlex Promega



## DNA Analysis:

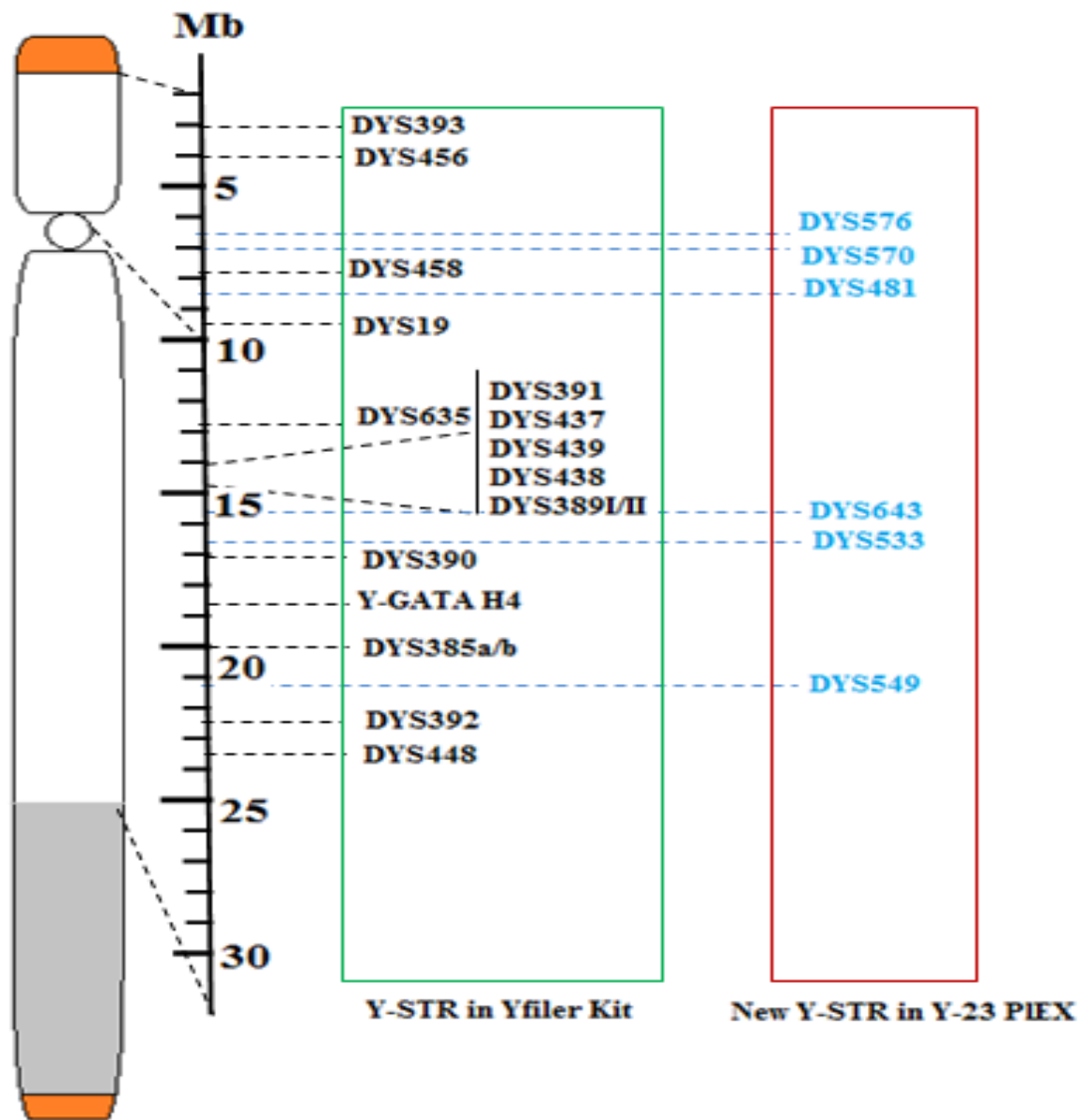
- ABI 3130 Genetic analyzer



## Statistical Analysis

- Using in-house System (FIMS)

# Y-STR systems



## Results

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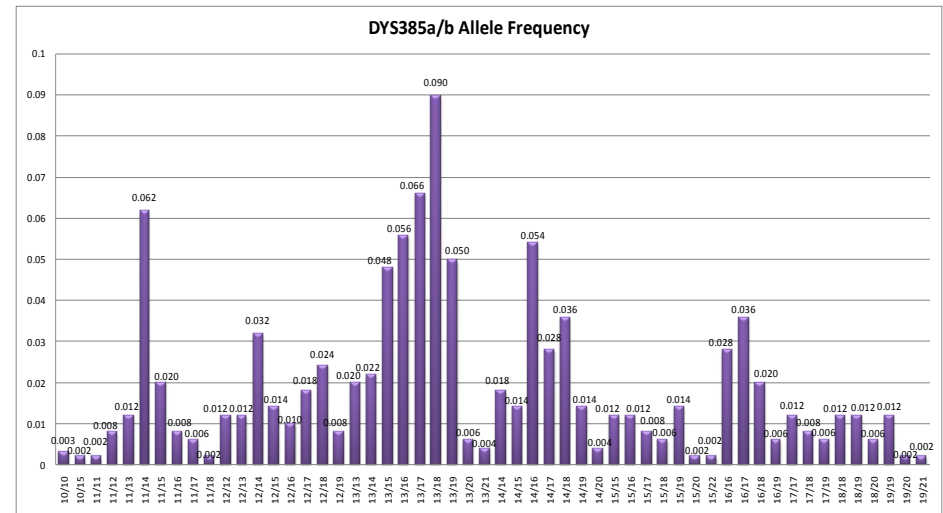
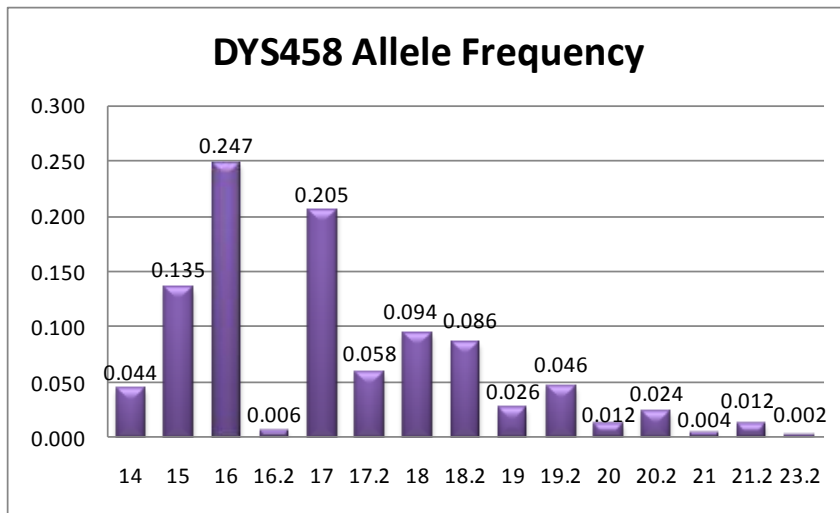
- Y-STR allele frequencies
- Y-STR population genetics
- Y-STR haplotype frequencies
- Resolving common haplotypes using Y-23 Kit
- Detecting false positivity in villages
- Resolving common haplotypes in villages using Y-23 Kit.





## 2) Y-STR population genetics: Gene Diversity

Most polymorphic markers: **DYS458** and **DYS385a/b**

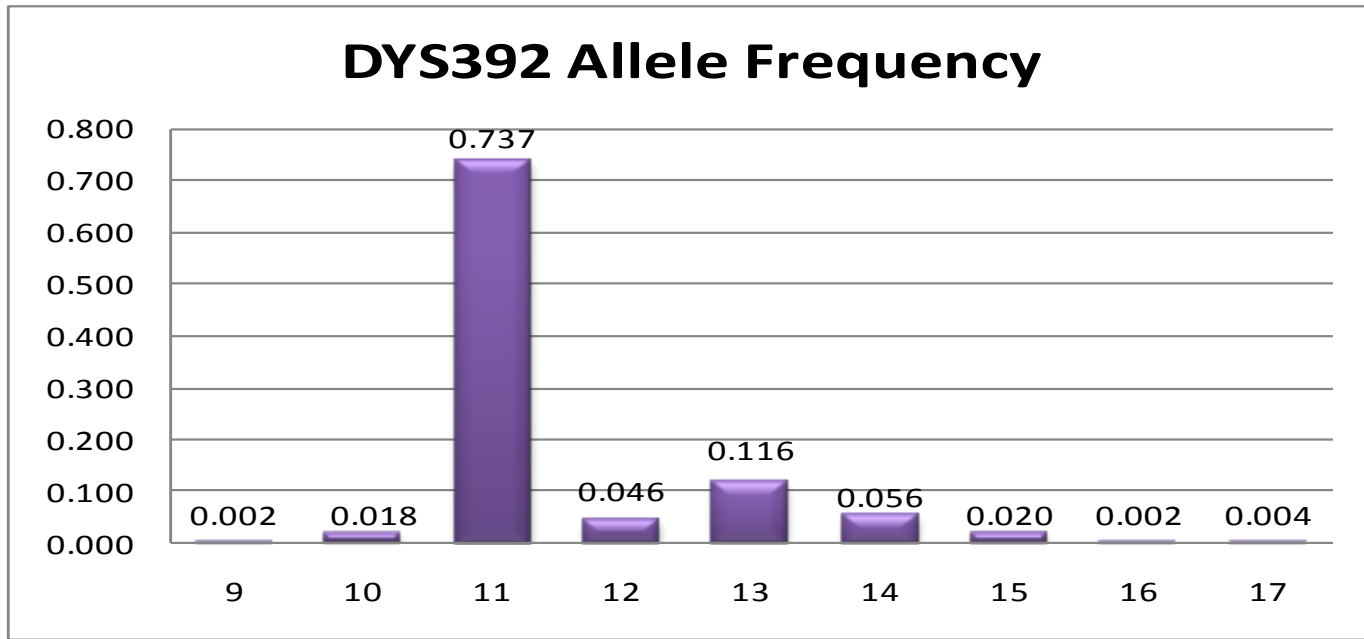


Lebanese population gene diversity(L-GD) compared as to the Y-STR systems in the **Caucasian population (C-GD)**.

	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y-GATA-H4	DYS385a/b
L-GD*	0.63	0.58	0.75	0.68	0.50	0.44	0.59	0.56	0.65	0.64	0.67	0.65	0.86	0.78	0.56	0.92
Rank	10	12	4	5	15	16	11	13	7	9	6	8	2	3	14	1
C-GD*	0.50	0.52	0.68	0.71	0.55	0.60	0.38	0.58	0.59	0.65	0.60	0.72	0.78	0.64	0.60	0.84
Rank	15	14	5	4	13	8	16	12	11	6	10	3	2	7	9	1

# Gene Diversity

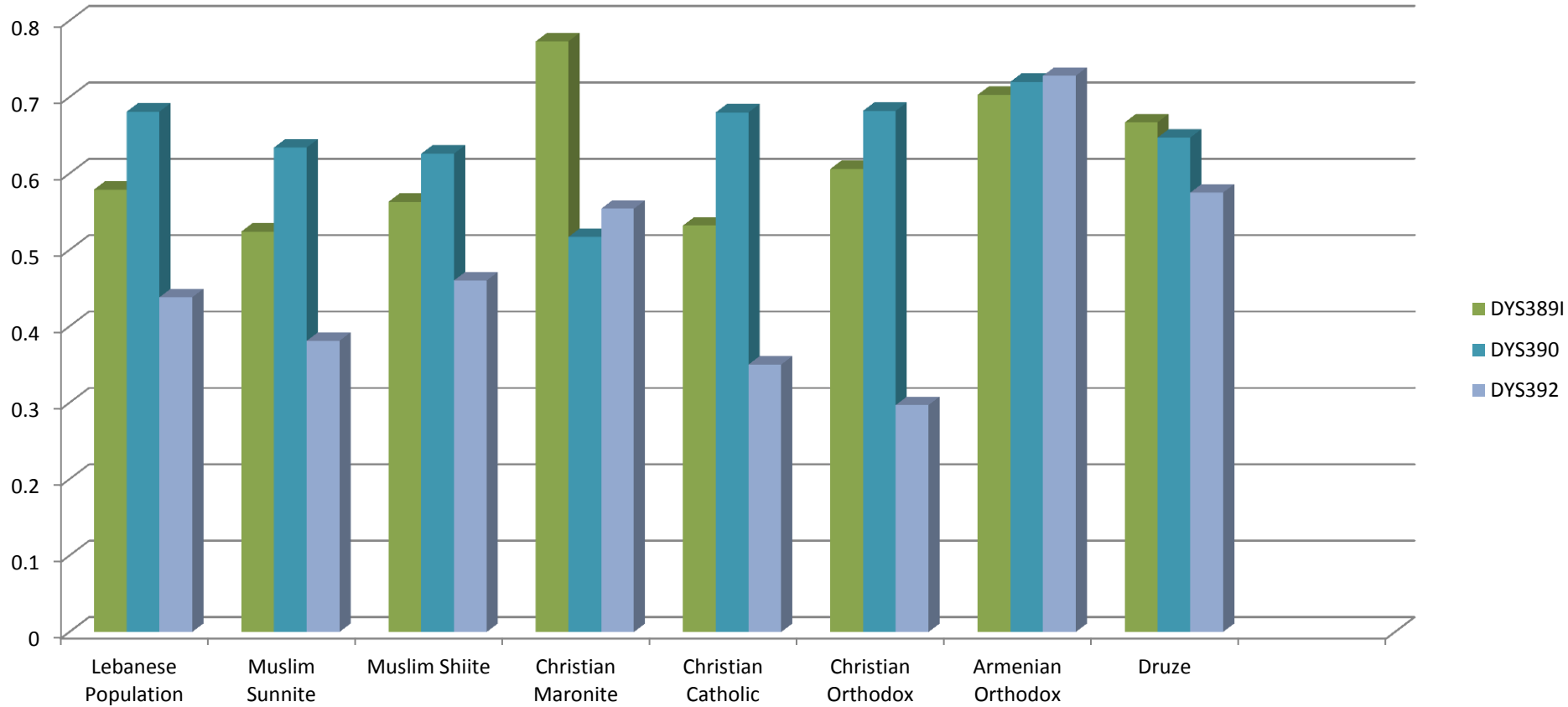
Least polymorphic markers: DYS392



Lebanese population gene diversity(L-GD) compared as to the Y-STR systems in the **Caucasian population (C-GD)**.

	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y-GATA-H4	DYS385a/b
L-GD*	0.63	0.58	0.75	0.68	0.50	0.44	0.59	0.56	0.65	0.64	0.67	0.65	0.86	0.78	0.56	0.92
Rank	10	12	4	5	15	16	11	13	7	9	6	8	2	3	14	1
C-GD*	0.50	0.52	0.68	0.71	0.55	0.60	0.38	0.58	0.59	0.65	0.60	0.72	0.78	0.64	0.60	0.84
Rank	15	14	5	4	13	8	16	12	11	6	10	3	2	7	9	1

# Sub-population Gene Diversity



**Gene diversity for Y-STR systems in different Lebanese sub-populations.**

# Locus Power of Discrimination

Power of discrimination for individual systems in Lebanese and Caucasian populations

	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y-GATA-H4	DYS385a/b
L-PD*	0.67	0.49	0.67	0.60	0.56	0.37	0.51	0.47	0.57	0.56	0.59	0.56	0.79	0.69	0.47	0.92
C-PD*	0.54	0.52	0.69	0.69	0.55	0.61	0.32	0.59	0.58	0.65	0.63	0.73	0.77	0.63	0.59	0.82

\*L-PD: Lebanese Power of Discrimination; \*C-PD: Caucasian Power of Discrimination

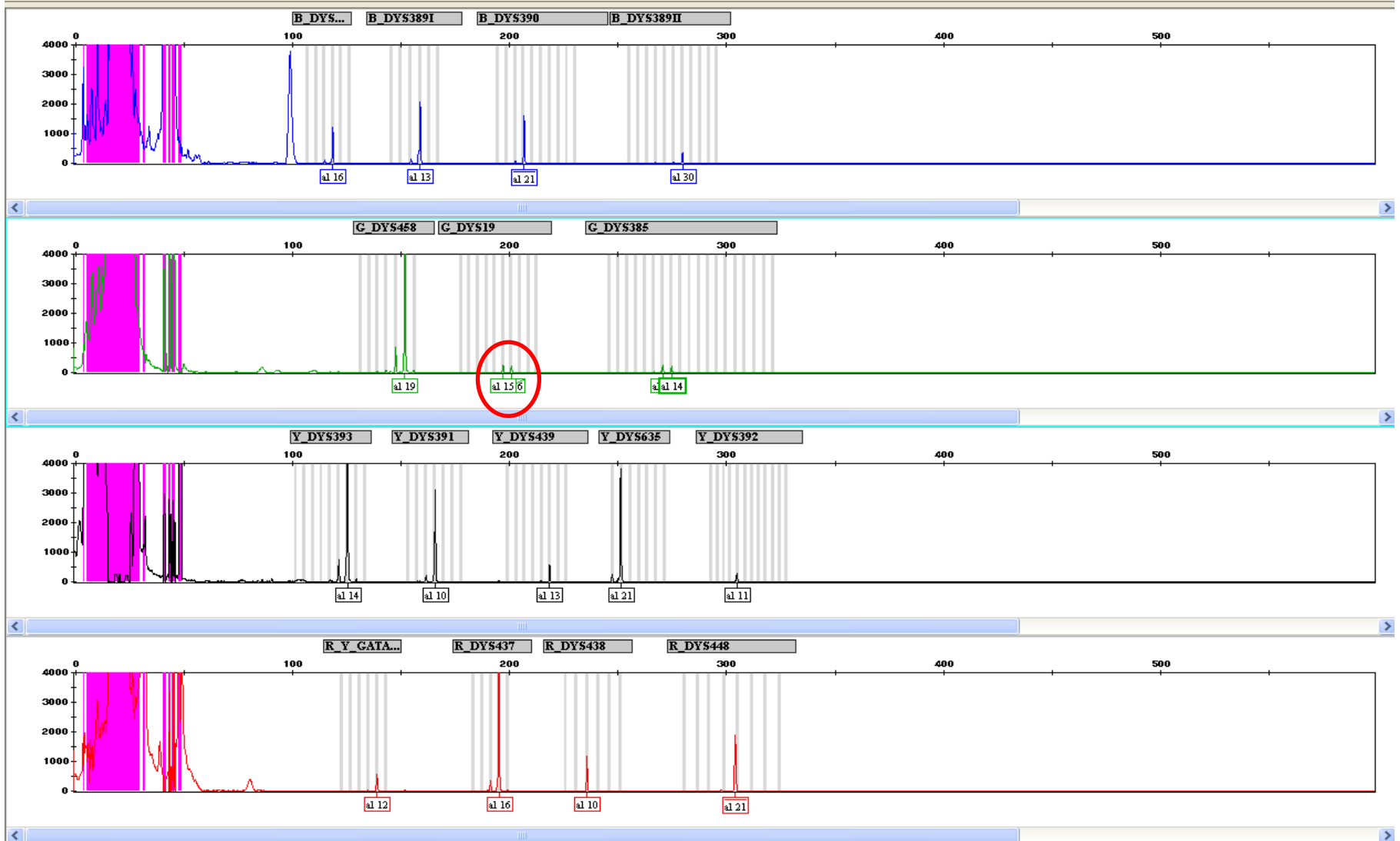
## Rare and new Alleles

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	Rare Allele	New Allele
DYS456	Allele 12 (n=1)	
DYS635	Allele 17 (n=2)	
DYS438		Allele 7 (n=2)

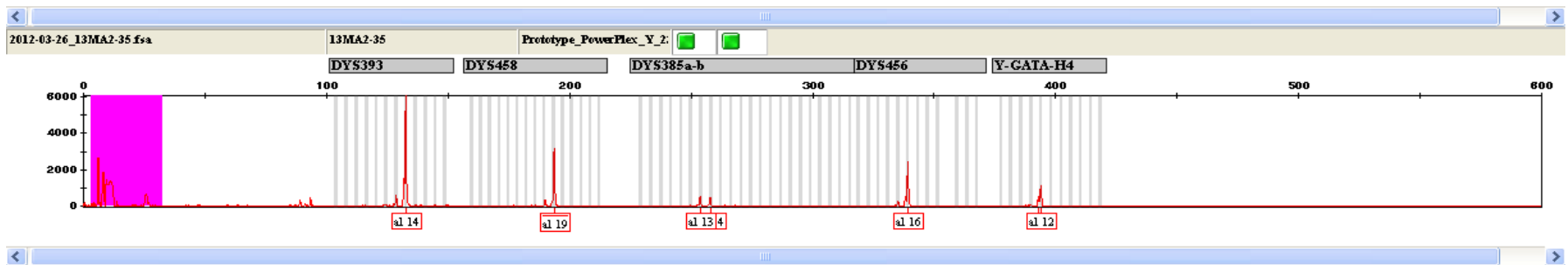
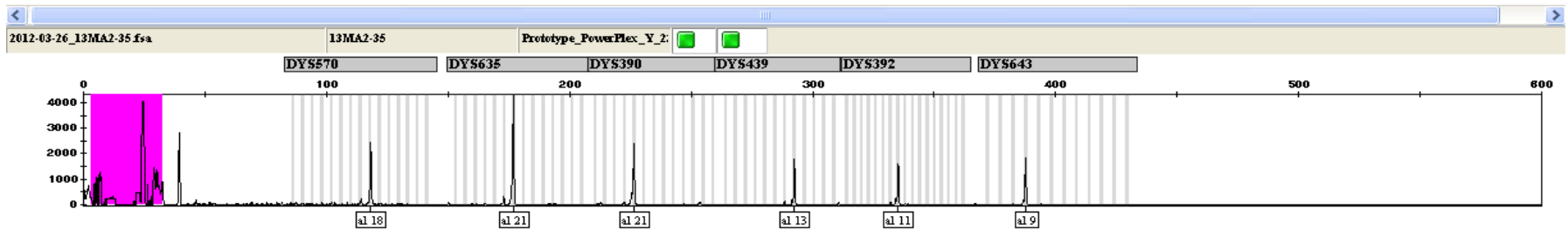
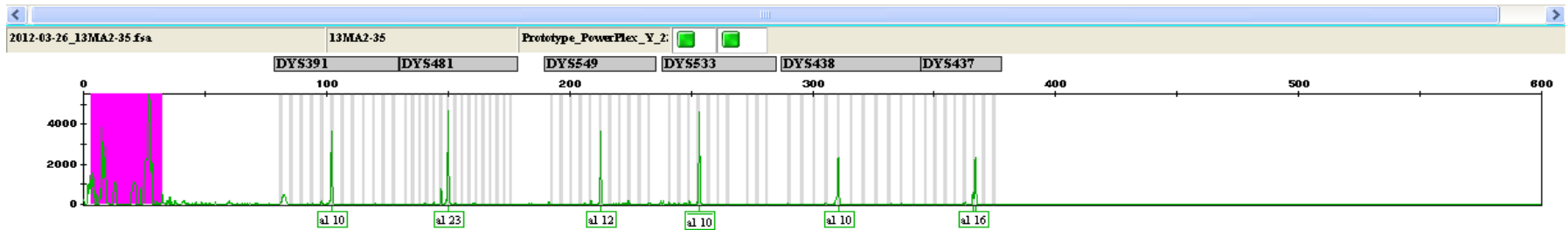
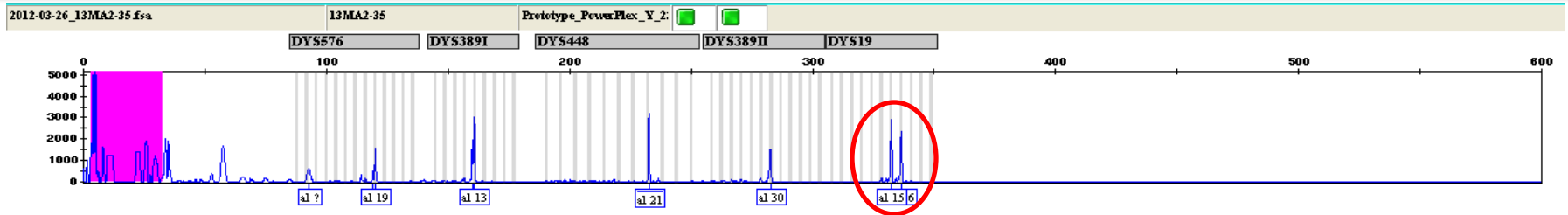
**Rare and new alleles observed in the Lebanese population.**

# Particular Profiles using Y-filer



Duplication of the DYS19 system

# Particular Profiles confirmed using Y-23



Duplication of the DYS19 system

# Particular Profiles using Y-filer



Possible allele dropout at DYS448 system



# Particular Profiles confirmed using Y-23



DYS448 system null allele

Lebanese Y-STR allele frequencies and population genetics

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**Heterogeneity among Lebanese sub-populations.**

### 3) “Y-Filer” Haplotype Frequencies

Out of 502 tested individuals

- **431 unique haplotypes**

Haplotype frequency for a profile occurring 1 time: 0.0059% (1 in 16976)

- **2 groups of 5 individuals each, all individuals have the same haplotype**

Haplotype frequency for a profile occurring 5 times: 0.018% (1 in 5362)

- **3 groups of 3 individuals each, all individuals have the same haplotype**

Haplotype frequency for a profile occurring 3 times: 0.012% (1 in 7866)

- **26 groups of 2 individuals each, all individuals have the same haplotype**

Haplotype frequency for a profile occurring 2 times: 0.0094% (1 in 10536)

Community	Region																
Muslim-Shiite	South Lebanon	15	13	23	29	16	14	13/15	12	10	11	23	11	11	15	9	21
Muslim-Shiite	South Lebanon	15	13	23	29	16	14	13/15	12	10	11	23	11	11	15	9	21
Christian Maronite	South Lebanon	15	13	22	29	15	15	13/15	12	10	11	20	11	11	15	9	21
Christian Maronite	North Lebanon	16	14	24	31	16	14	19/19	13	11	12	20	11	12	14	10	19
Christian Maronite	North Lebanon	16	14	24	31	16	14	19/19	13	11	12	20	11	12	14	10	19

## Comparison of Haplotype Frequencies

Religious community	Haplotype frequency (occurrence)	Match frequency in total population	Match frequency in sub-population
<b>Muslim Sunnite</b> (n = 141)	2	1/10536	1/2983
<b>Muslim Shiite</b> (n = 136)	5	1/5363	1/1868
<b>Christian Maronite</b> (n = 109)	3	1/7866	1/1704
<b>Christian Maronite</b> (n = 109)	5	1/5363	1/1168
<b>Druze</b> (n = 32)	2	1/10536	1/683

Rate of match of the most frequent haplotype in the total population and in the subpopulation where each haplotype occurred

# Statistical calculations on “Y-filer” data in Lebanese population

<b>Statistical Indices</b>	<b>Lebanese Population</b>	<b>Caucasian Population</b>
<b>Haplotype Diversity (HD)</b>	0.9995	0.9999
<b>Unique Haplotype (UHD)</b>	431 (total 502)	714 (total 778)
<b>Random Match Probability (RMP)</b>	0.0004%	0.0001%
<b>Discrimination Capacity</b>	<b>92.0%</b>	<b>95.8%</b>

**Statistical indices for the Lebanese and Caucasian populations**

## “Y-filer” Haplotype Frequencies

- **High haplotype match most probably due to high co-ancestry rate and the discriminating power of the kit used.**
- **Higher match probability within sub-populations than when crossing a profile against Lebanese dataset.**

**Resolving common haplotypes using Y-23 Kit**





## Second group of 5 individuals sharing a “Y-filer” haplotype

	Individual 1	Individual 2	Individual 3	Individual 4	Individual 5
DYS456	16	16	16	16	16
DYS389I	14	14	14	14	14
DYS390	24	24	24	24	24
DYS389II	31	31	31	31	31
DYS458	16	16	16	16	16
DYS19	14	14	14	14	14
DYS385a/b	19-19	19-19	19-19	19-19	19-19
DYS393	13	13	13	13	13
DYS391	11	11	11	11	11
DYS439	12	12	12	12	12
DYS635	20	20	20	20	20
DYS392	11	11	11	11	11
Y GATA H4	12	12	12	12	12
DYS437	14	14	14	14	14
DYS438	10	10	10	10	10
DYS448	19	19	19	19	19
DYS576	18	17	19	18	18
DYS481	26	25	26	26	26
DYS549	12	12	12	12	12
DYS533	13	13	13	13	13
DYS570	19	19	20	20	20
DYS643	12	12	12	12	12

**Comment** Y-23 was able to differentiate 5 individuals with same haplotype into 4 different haplotypes.

## First group of 3 individuals sharing a “Y-filer” haplotype

	Individual 1	Individual 2	Individual 3
DYS456	15	15	15
DYS389I	13	13	13
DYS390	23	23	23
DYS389II	29	29	29
DYS458	16	16	16
DYS19	15	15	15
DYS385a/b	12-14	12-14	12-14
DYS393	13	13	13
DYS391	10	10	10
DYS439	11	11	11
DYS635	21	21	21
DYS392	11	11	11
Y GATA H4	12	12	12
DYS437	14	14	14
DYS438	10	10	10
DYS448	19	19	19
DYS576	16	17	17
DYS481	24	24	24
DYS549	14	14	14
DYS533	12	12	12
DYS570	13	13	13
DYS643	11	11	11
<b>Comment</b>	<b>Y-23 differentiated one individual from the other two</b>		

## Second group of 3 individuals sharing a “Y-filer” haplotype

	Individual 1	Individual 2	Individual 3
DYS456	15	15	15
DYS389I	13	13	13
DYS390	23	23	23
DYS389II	30	30	30
DYS458	18	18	18
DYS19	14	14	14
DYS385a/b	13-19	13-19	13-19
DYS393	12	12	12
DYS391	10	10	10
DYS439	12	12	12
DYS635	21	21	21
DYS392	11	11	11
Y GATA H4	11	11	11
DYS437	14	14	14
DYS438	10	10	10
DYS448	20	20	20
DYS576	17	17	17
DYS481	25	24	24
DYS549	13	13	13
DYS533	11	11	11
DYS570	17	17	17
DYS643	9	9	9
<b>Comment</b>	<b>Y-23 differentiated one individual from the other two</b>		

### Third group of 3 individuals sharing a “Y-filer” haplotype

	Individual 1	Individual 2	Individual 3
DYS456	15	15	15
DYS389I	13	13	13
DYS390	22	22	22
DYS389II	29	29	29
DYS458	15	15	15
DYS19	15	15	15
DYS385a/b	13-15	13-15	13-15
DYS393	12	12	12
DYS391	10	10	10
DYS439	11	11	11
DYS635	20	20	20
DYS392	11	11	11
Y GATA H4	11	11	11
DYS437	15	15	15
DYS438	9	9	9
DYS448	21	21	21
DYS576	17	17	17
DYS481	23	25	25
DYS549	13	13	13
DYS533	11	15	11
DYS570	17	19	18
DYS643	6	6	6
<b>Comment</b>	<b>Y-23 differentiated 3 individuals who had a common haplotype</b>		

26 groups of 2 individuals each



18 groups were resolved or differentiated

	Individual 1	Individual 2
DYS456	15	15
DYS389I	14	14
DYS390	25	25
DYS389II	29	29
DYS458	16	16
DYS19	15	15
DYS385a/b	12-18	12-18
DYS393	11	11
DYS391	10	10
DYS439	11	11
DYS635	23	23
DYS392	14	14
Y GATA H4	11	11
DYS437	16	16
DYS438	10	10
DYS448	19	19
DYS576	18	17
DYS481	24	24
DYS549	13	14
DYS533	12	12
DYS570	15	16
DYS643	11	11

## 16 versus 23 systems

<b>Statistical Indices</b>	<b>Using Y-filer</b>	<b>Using Y-23</b>
<b>Haplotype Diversity (HD)</b>	<b>0.9995</b>	<b>0.9999</b>
<b>Unique Haplotype (UHD)</b>	<b>431</b> (total 502)	<b>478</b> (total 502)
<b>Random Match Probability (RMP)</b>	0.0004	0.0001
<b>Discrimination Capacity</b>	<b>92.0%</b>	<b>97.6%</b>

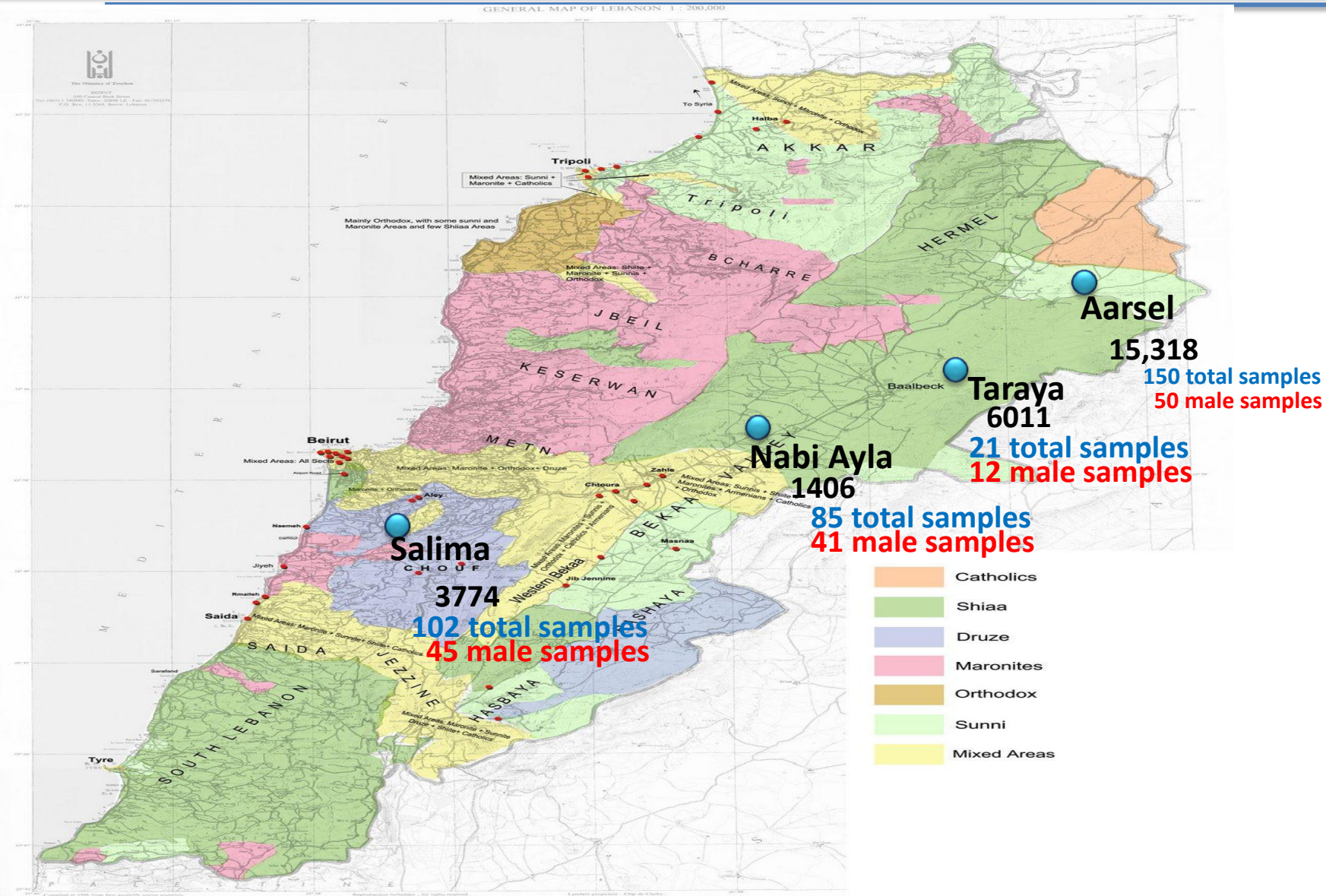
## Specific Aims

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### Target Solutions:

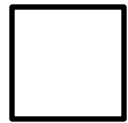
- Establish Y-STR allele and haplotype frequencies for Lebanese Population .
- **Assessment of Consanguinity and Endogamy effect on Y-STR analysis.**

# Assessment of Consanguinity and Endogamy





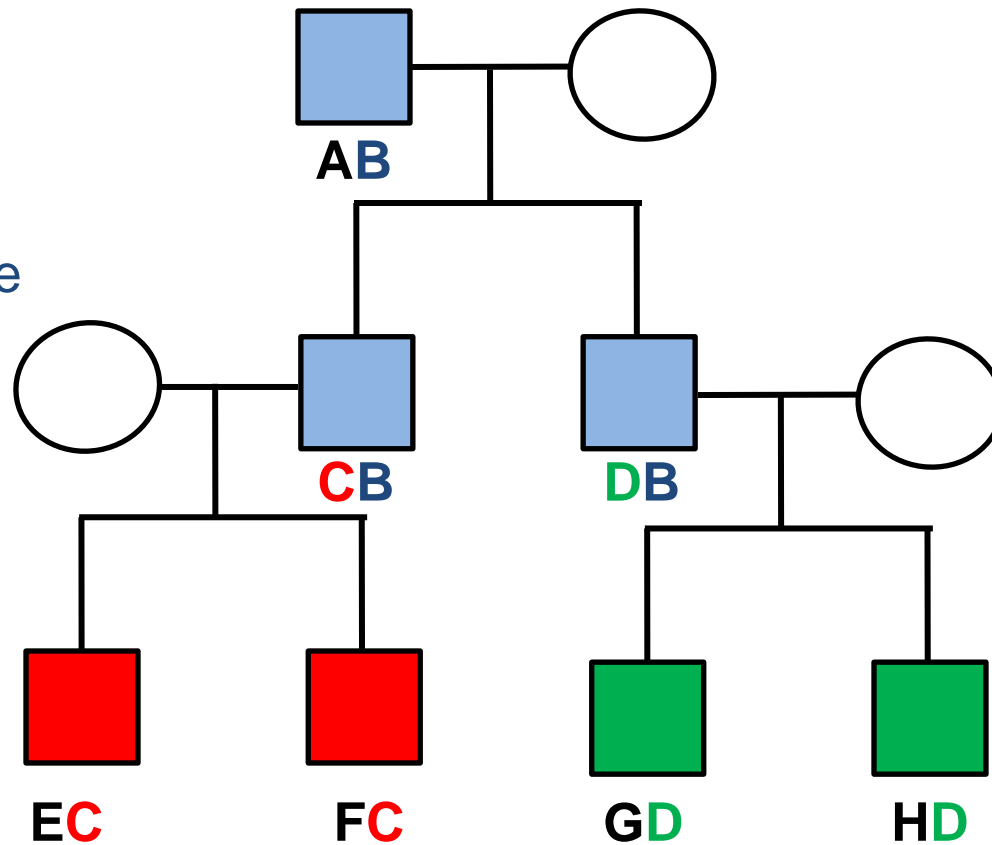
# Origin of family names within a village



**AB**

A Forename

B Family name



A) Taraya 6011 inhabitants (1 Family Name)  
12 male Samples/4 haplotypes

Systems	6 Individuals	1 Individual	1 Individual	4 Individuals
DYS 456	15	15	15	15
DYS 389I	14	14	14	14
DYS 390	25	24	25	25
DYS 389II	32	32	32	32
DYS 458	17	17	17	17
DYS 19	14	14	14	14
DYS 385 a/b	16-17	16-17	17-17	16-17
DYS 393	13	13	13	13
DYS 391	10	10	10	10
DYS 439	12	12	12	12
DYS 635	21	21	21	22
DYS 392	11	11	11	11
Y GATA H4	11	11	11	11
DYS 437	14	14	14	14
DYS 438	10	10	10	10
DYS 448	19	19	19	19

Different haplotypes observed in Taraya village

B) Aarsal 15,318 inhabitants  
50 male samples/30 haplotypes

System	9 Individuals	4 Individuals	4 Individuals	3 Individuals	2 Individuals	2 Individuals
DYS456	16	17	14	16	16	16
DYS398I	13	13	14	13	13	13
DYS390	24	23	24	24	24	24
DYS389II	30	29	31	30	30	30
DYS458	15	16	17.2	15	15	15
DYS19	14	15	14	14	14	14
DYS385a/b	18-19	13-16	13-18	18-19	18/19	18/19
DYS393	13	12	12	13	13	14
DYS391	10	9	11	10	10	10
DYS439	14	12	11	13	13	14
DYS635	22	22	22	22	22	22
DYS392	11	11	11	11	11	11
Y-GATA H4	13	12	11	13	13	13
DYS437	14	14	14	14	14	14
DYS438	10	9	10	10	10	10
DYS448	19	21	20	19	19	19

**Different haplotypes observed in Aarsal village with extrapolation results for a haplotype occurring 9 times in this dataset**

**Extrapolation of Haplotype available 9 times:  $4\% < p < 32\%$**

C) Nabi Aayla 1406 habitants  
41 male samples/11 haplotypes

System	15 individuals	8 individual	5 individual	4 individual	3 individual
DYS456	15	16	15	14	15
DYS389I	14	13	14	12	13
DYS390	23	22	24	23	23
DYS389II	31	30	30	28	31
DYS458	15	15	15	17	15
DYS19	15	14	14	14	15
DYS385a/b	12-17	13-17	11-13	12-19	12-17
DYS393	12	11	12	12	12
DYS391	10	10	10	10	10
DYS439	11	11	12	11	11
DYS635	21	22	23	21	21
DYS392	11	14	13	11	11
Y GATA H4	11	12	13	11	11
DYS437	14	15	15	16	14
DYS438	9	10	12	9	9
DYS448	19	19	19	22	19

**Different haplotypes observed in Nabi-Aayla village with extrapolation results for a haplotype occurring 15 times in this dataset**

**Extrapolation of Haplotype available 15 times: 7% < p < 44.6%**

## D) Salima 3774 inhabitants 45 male samples/17 haplotypes

System	14 Individuals	5 Individuals	4 Individuals	3 Individuals	3 Individuals	2 Individuals	2 Individuals	2 Individuals	2 Individuals
DYS456	16	15	16	15	15	15	16	16	17
DYS389I	13	13	13	13	13	13	12	13	13
DYS390	25	22	24	22	24	25	23	25	23
DYS389II	29	29	30	30	32	29	28	29	28
DYS458	17	15	15	15	16	17	17	17	17
DYS19	14	15	13	14	16	14	15	14	17
DYS385a/b	12-14	13-17	15-17	13-17	11-14	12-14	14-18	13-14	14-16
DYS393	12	11	13	11	13	12	12	12	13
DYS391	10	10	10	10	11	10	10	10	10
DYS439	11	12	13	12	11	11	13	11	11
DYS635	23	23	23	23	24	23	24	23	21
DYS392	13	13	11	14	11	13	11	13	13
Y GATA H4	11	12	12	12	13	11	11	11	11
DYS437	15	15	14	14	14	15	15	15	14
DYS438	11	10	10	10	11	11	11	11	9
DYS448	20	19	20	19	19	20	19	20	19

**Different haplotypes observed in Salima village with extrapolation results for a haplotype occurring 14 times in this dataset**

**Extrapolation of Haplotype available 14 times: 13.4% < p < 48.8%**

# Matching haplotypes in villages v/s Lebanese dataset

DYS456	16	16
DYS398I	13	13
DYS390	24	24
DYS389II	30	30
DYS458	15	15
DYS19	14	14
DYS385a/b	18-19	18-19
DYS393	14	14
DYS391	10	10
DYS439	14	14
DYS635	22	22
DYS392	11	11
Y-GATA H4	13	13
DYS437	14	14
DYS438	10	10
DYS448	19	19
Community	Muslim Shiite	Muslim Shiite
Frequency	Aarsal Village (n=2)	Lebanese dataset (n=1)

**A profile match between a village that occurred once with a profile in the dataset**

DYS456	16	16
DYS398I	13	13
DYS390	25	25
DYS389II	29	29
DYS458	19	19
DYS19	14	14
DYS385a/b	12-14	12-14
DYS393	12	12
DYS391	10	10
DYS439	11	11
DYS635	23	23
DYS392	13	13
Y-GATA H4	11	11
DYS437	15	15
DYS438	11	11
DYS448	20	20
Community	Druze	Muslim Shiite
Frequency	Salima Village (n=1)	Lebanese dataset (n=1)

**A profile match between a village that occurred once with a profile in the dataset**

**Only two matches.**

# Using Y-23 to resolve matching haplotypes between villages and Lebanese dataset

DYS456	16	16
DYS398I	13	13
DYS390	24	24
DYS389II	30	30
DYS458	15	15
DYS19	14	14
DYS385a/b	18-19	18-19
DYS393	14	14
DYS391	10	10
DYS439	14	14
DYS635	22	22
DYS392	11	11
Y-GATA H4	13	13
DYS437	14	14
DYS438	10	10
DYS448	19	19
<b>DYS576</b>	18	18
<b>DYS481</b>	22	22
<b>DYS549</b>	12	12
<b>DYS533</b>	11	11
<b>DYS570</b>	20	20
<b>DYS643</b>	11	9
Frequency	Aarsal Village (n=2)	Lebanese dataset (n=1)

DYS456	16	16
DYS398I	13	13
DYS390	25	25
DYS389II	29	29
DYS458	19	19
DYS19	14	14
DYS385a/b	12-14	12-14
DYS393	12	12
DYS391	10	10
DYS439	11	11
DYS635	23	23
DYS392	13	13
Y-GATA H4	11	11
DYS437	15	15
DYS438	11	11
DYS448	20	20
<b>DYS576</b>	18	17
<b>DYS481</b>	22	22
<b>DYS549</b>	13	13
<b>DYS533</b>	12	12
<b>DYS570</b>	15	15
<b>DYS643</b>	10	10
Frequency	Salima Village (n=1)	Lebanese dataset (n=1)

## Compare haplotype frequencies between village and Lebanese dataset

Lebanese villages	Occurance	Rate of match in village	Rate of match in total population
<b>Nabi-Aayla</b>	15	1/195	1/16976
<b>Nabi-Aayla</b>	8	1/316	1/16976
<b>Salima</b>	14	1/224	1/16976
<b>Salima</b>	2	1/959	1/10626
<b>Aarsal</b>	7	1/341	1/16976
<b>Aarsal</b>	3	1/638	1/16976

**Comparison of haplotype frequencies in villages and Lebanese population**



## Statistical calculations on “Y-filer” data in Lebanese villages

Population sample	Haplotype Diversity (HD)	Unique Haplotype (UHD)	Random Match Probability (RMP)	Discrimination Capacity (DC)
<b>Aarsal village</b>	0.956	25 in 50 tested	0.043	64.0%
<b>Salima village</b>	0.883	8 in 45 tested	0.117	37.7%
<b>Nabi-Aayla village</b>	0.816	6 in 41 tested	0.184	26.8%
<b>Lebanese population</b>	0.9995	431 in 502 tested	0.0004	92.0%

**Statistical indices for the different tested villages and comparison to the Lebanese populations**

# Comparison of HD between sub-population and villages

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Community	Haplotype Diversity
Muslim Sunnite Sub-population (n=141)	0.9998
Aarsal Village (Muslim Sunnite) (n=40)	0.957
Muslim Shiite Sub-population (n=136)	0.9982
Nabi Aayla Village (Muslim Shiite) (n=41)	0.816
Druze Sub-population (n=32)	1
Salima Village (Druze) (n=45)	0.883

## Detecting false positivity in villages

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- **Non-related individuals had common Y-STR haplotypes.**
- **Current number of Y-STR loci not enough to completely resolve forensic and kinship studies.**
- **Match probability increased in religiously and geographically isolated regions.**

# **Resolving common haplotypes in villages using Y-23 Kit**

# Taraya Village

	6 Individuals	1 Individual	1 Individual	4 Individuals
DYS456	15	15	15	15
DYS389I	14	14	14	14
DYS390	25	24	25	25
DYS389II	32	32	32	32
DYS458	17	17	17	17
DYS19	14	14	14	14
DYS385a/b	16-17	16-17	17-17	16-17
DYS393	13	13	13	13
DYS391	10	10	10	10
DYS439	12	12	12	12
DYS635	21	21	21	22
DYS392	11	11	11	11
Y GATA H4	11	11	11	11
DYS437	14	14	14	14
DYS438	10	10	10	10
DYS448	19	19	19	19
<b>DYS576</b>	15	15	15	15
<b>DYS481</b>	23	24	23	23
<b>DYS549</b>	12	12	12	12
<b>DYS533</b>	10	10	10	10
<b>DYS570</b>	21	21	20	21
<b>DYS643</b>	12	12	12	12
<b>Comment</b>	<b>Y-23 Kit was not able to further discriminate between individuals of this village</b>			

# Aarsal Village

System	9 Individuals	4 Individuals	4 Individuals	3 Individuals	2 Individuals	2 Individuals
DYS456	16	17	14	16	16	16
DYS398I	13	13	14	13	13	13
DYS390	24	23	24	24	24	24
DYS389II	30	29	31	30	30	30
DYS458	15	16	17.2	15	15	15
DYS19	14	15	14	14	14	14
DYS385a/b	18-19	13-16	13-18	18-19	18/19	18/19
DYS393	13	12	12	13	13	14
DYS391	10	9	11	10	10	10
DYS439	14	12	11	13	13	14
DYS635	22	22	22	22	22	22
DYS392	11	11	11	11	11	11
Y-GATA H4	13	12	11	13	13	13
DYS437	14	14	14	14	14	14
DYS438	10	9	10	10	10	10
DYS448	19	21	20	19	19	19
<b>Comment</b>	<b>Partially Resolved</b>	<b>Not resolved</b>	<b>Partially Resolved</b>	<b>Not resolved</b>	<b>Not resolved</b>	<b>Not resolved</b>

# Aarsal Village

## Group of 9 Individuals

	Individual 1	Individual 2	Individual 3	Individual 4	Individual 5	Individual 6	Individual 7	Individual 8	Individual 9
DYS456	16	16	16	16	16	16	16	16	16
DYS389I	13	13	13	13	13	13	13	13	13
DYS390	24	24	24	24	24	24	24	24	24
DYS389II	30	30	30	30	30	30	30	30	30
DYS458	15	15	15	15	15	15	15	15	15
DYS19	14	14	14	14	14	14	14	14	14
DYS385a/b	18-19	18-19	18-19	18-19	18-19	18-19	18-19	18-19	18-19
DYS393	13	13	13	13	13	13	13	13	13
DYS391	10	10	10	10	10	10	10	10	10
DYS439	14	14	14	14	14	14	14	14	14
DYS635	22	22	22	22	22	22	22	22	22
DYS392	11	11	11	11	11	11	11	11	11
Y GATA H4	13	13	13	13	13	13	13	13	13
DYS437	14	14	14	14	14	14	14	14	14
DYS438	10	10	10	10	10	10	10	10	10
DYS448	19	19	19	19	19	19	19	19	19
DYS576	18	18	18	18	18	18	18	18	18
DYS481	22	22	22	22	22	22	22	22	22
DYS549	12	12	12	12	12	12	12	12	12
DYS533	11	11	11	11	11	11	11	11	11
DYS570	19	19	20	20	20	20	20	20	20
DYS643	11	11	11	11	11	11	11	11	11
Comment	Y-23 Kit differentiated this group of 9 individuals into 4 different haplotypes								

# Aarsal Village

## Group of 4 Individuals

	Individual 1	Individual 2	Individual 3	Individual 4
DYS456	14	14	14	14
DYS389I	14	14	14	14
DYS390	24	24	24	24
DYS389II	31	31	31	31
DYS458	17.2	17.2	17.2	17.2
DYS19	14	14	14	14
DYS385a/b	13-18	13-18	13-18	13-18
DYS393	12	12	12	12
DYS391	11	11	11	11
DYS439	11	11	11	11
DYS635	22	22	22	22
DYS392	11	11	11	11
Y GATA H4	11	11	11	11
DYS437	14	14	14	14
DYS438	10	10	10	10
DYS448	20	20	20	20
<b>DYS576</b>	18	18	18	18
<b>DYS481</b>	26	26	26	26
<b>DYS549</b>	13	13	13	13
<b>DYS533</b>	11	11	11	11
<b>DYS570</b>	18	18	18	18
<b>DYS643</b>	9	9	9	10
<b>Comment</b>	<b>Y-23 Kit was able to differentiate one individual from this group whereby individuals belonged to four different family names.</b>			



## Y-filer and Y-23 Kits Comparison in Aarsal Village

<b>Statistical Indices</b>	<b>Using Y-filer</b>	<b>Using Y-23</b>
<b>Haplotype Diversity (HD)</b>	<b>0.956</b>	<b>0.977</b>
<b>Unique Haplotype (UHD)</b>	<b>25 (total 50)</b>	<b>26 (total 50)</b>
<b>Random Match Probability (RMP)</b>	<b>0.043</b>	<b>0.023</b>
<b>Discrimination Capacity</b>	<b>64.0%</b>	<b>68.0%</b>

# Nabi Aayla Village

System	15 Individuals	8 Individuals	5 Individuals	4 Individuals	3 Individuals
DYS456	15	16	15	14	15
DYS398I	14	13	14	12	13
DYS390	23	22	24	23	23
DYS389II	31	30	30	28	31
DYS458	15	15	15	17	15
DYS19	15	14	14	14	15
DYS385a/b	12-17	13-17	11-13	12-19	12-17
DYS393	12	11	12	12	12
DYS391	10	10	10	10	10
DYS439	11	11	12	11	11
DYS635	21	22	23	21	21
DYS392	11	14	13	11	11
Y-GATA H4	11	12	13	11	11
DYS437	14	15	15	16	14
DYS438	9	10	12	9	9
DYS448	19	19	19	22	19
<b>Comment</b>	<b>Partially Resolved</b>	<b>Not resolved</b>	<b>Partially Resolved</b>	<b>Not resolved</b>	<b>Partially Resolved</b>

# Nabi Aayla Village

## Group of 15 Individuals

DYS456	15	15	15	15	15	15	15	15	15	15	15	15	15	15	15
DYS389I	14	14	14	14	14	14	14	14	14	14	14	14	14	14	14
DYS390	23	23	23	23	23	23	23	23	23	23	23	23	23	23	23
DYS389II	31	31	31	31	31	31	31	31	31	31	31	31	31	31	31
DYS458	15	15	15	15	15	15	15	15	15	15	15	15	15	15	15
DYS19	15	15	15	15	15	15	15	15	15	15	15	15	15	15	15
DYS385a/ b	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17	12-17
DYS393	12	12	12	12	12	12	12	12	12	12	12	12	12	12	12
DYS391	10	10	10	10	10	10	10	10	10	10	10	10	10	10	10
DYS439	11	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS635	21	21	21	21	21	21	21	21	21	21	21	21	21	21	21
DYS392	11	11	11	11	11	11	11	11	11	11	11	11	11	11	11
Y GATA H4	11	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS437	14	14	14	14	14	14	14	14	14	14	14	14	14	14	14
DYS438	9	9	9	9	9	9	9	9	9	9	9	9	9	9	9
DYS448	19	19	19	19	19	19	19	19	19	19	19	19	19	19	19
DYS576	16	16	16	16	16	16	16	16	16	16	16	16	16	16	16
DYS481	24	24	24	24	24	24	24	24	24	24	24	24	24	24	24
DYS549	13	13	13	13	13	13	13	13	13	13	13	13	13	13	13
DYS533	11	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS570	19	19	19	19	19	19	19	19	19	19	19	19	19	19	18
DYS643	9	9	9	9	9	9	9	9	9	9	9	9	9	9	9

**Comment** Y-23 kit was able to differentiate only one individual from this group of 15 individuals belonging to 3 different family names

# Nabi Aayla Village

## Group of 5 Individuals

	Individual 1	Individual 2	Individual 3	Individual 4	Individual 5
DYS456	15	15	15	15	15
DYS389I	14	14	14	14	14
DYS390	24	24	24	24	24
DYS389II	30	30	30	30	30
DYS458	15	15	15	15	15
DYS19	14	14	14	14	14
DYS385a/b	11-13	11-13	11-13	11-13	11-13
DYS393	12	12	12	12	12
DYS391	10	10	10	10	10
DYS439	12	12	12	12	12
DYS635	23	23	23	23	23
DYS392	13	13	13	13	13
Y GATA H4	13	13	13	13	13
DYS437	15	15	15	15	15
DYS438	12	12	12	12	12
DYS448	19	19	19	19	19
<b>DYS576</b>	16	16	16	16	16
<b>DYS481</b>	22	22	22	22	22
<b>DYS549</b>	12	12	12	12	12
<b>DYS533</b>	12	12	12	12	13
<b>DYS570</b>	15	15	15	15	15
<b>DYS643</b>	10	10	10	10	10
<b>Comment</b>	<b>Y-23 was able to differentiate only one individual from this group of individuals with same family name</b>				

# Nabi Aayla Village

## Group of 3 Individuals

	Individual 1	Individual 2	Individual 3
DYS456	15	15	15
DYS389I	13	13	13
DYS390	23	23	23
DYS389II	31	31	31
DYS458	15	15	15
DYS19	15	15	15
DYS385a/b	12-17	12-17	12-17
DYS393	12	12	12
DYS391	10	10	10
DYS439	11	11	11
DYS635	21	21	21
DYS392	11	11	11
Y GATA H4	11	11	11
DYS437	14	14	14
DYS438	9	9	9
DYS448	19	19	19
DYS576	16	16	16
DYS481	25	25	25
DYS549	13	13	13
DYS533	11	11	11
DYS570	20	20	19
DYS643	9	9	9

Comment

Y-23 was able to differentiate only one individual from this group of individuals with same family name

## Y-filer and Y-23 Kits Comparison In Nabi Ayla Village

<b>Statistical Indices</b>	<b>Using Y-filer</b>	<b>Using Y-23</b>
<b>Haplotype Diversity (HD)</b>	<b>0.816</b>	<b>0.839</b>
<b>Unique Haplotype (UHD)</b>	<b>6 (total 41)</b>	<b>9 (total 41)</b>
<b>Random Match Probability (RMP)</b>	<b>0.184</b>	<b>0.161</b>
<b>Discrimination Capacity</b>	<b>26.8%</b>	<b>34.1%</b>



# Salima Village

## Group of 14 Individuals

DYS456	16	16	16	16	16	16	16	16	16	16	16	16	16	16
DYS389I	13	13	13	13	13	13	13	13	13	13	13	13	13	13
DYS390	25	25	25	25	25	25	25	25	25	25	25	25	25	25
DYS389II	29	29	29	29	29	29	29	29	29	29	29	29	29	29
DYS458	17	17	17	17	17	17	17	17	17	17	17	17	17	17
DYS19	14	14	14	14	14	14	14	14	14	14	14	14	14	14
DYS385a/b	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14	12-14
DYS393	12	12	12	12	12	12	12	12	12	12	12	12	12	12
DYS391	10	10	10	10	10	10	10	10	10	10	10	10	10	10
DYS439	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS635	23	23	23	23	23	23	23	23	23	23	23	23	23	23
DYS392	13	13	13	13	13	13	13	13	13	13	13	13	13	13
Y GATA H4	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS437	15	15	15	15	15	15	15	15	15	15	15	15	15	15
DYS438	11	11	11	11	11	11	11	11	11	11	11	11	11	11
DYS448	20	20	20	20	20	20	20	20	20	20	20	20	20	20
DYS576	18	18	18	18	18	18	18	18	18	18	17	17	19	19
DYS481	22	22	22	22	22	22	22	22	22	22	22	22	22	24
DYS549	13	13	13	13	13	13	13	13	13	12	13	13	13	12
DYS533	12	12	12	12	12	12	12	11	11	12	12	12	12	12
DYS570	15	15	15	15	16	16	16	15	15	15	15	16	15	15
DYS643	10	10	10	10	10	10	10	10	10	10	10	10	10	10

**Comment** Y-23 Kit was able to differentiate 14 individuals from three different family names with same haplotype into 8 different haplotypes



## Y-filer and Y-23 Kits Comparison in Salima Village

<b>Statistical Indices</b>	<b>Using Y-filer</b>	<b>Using Y-23</b>
<b>Haplotype Diversity (HD)</b>	<b>0.883</b>	<b>0.965</b>
<b>Unique Haplotype (UHD)</b>	<b>8 (total 45)</b>	<b>13 (total 45)</b>
<b>Random Match Probability (RMP)</b>	<b>0.117</b>	<b>0.034</b>
<b>Discrimination Capacity</b>	<b>37.7%</b>	<b>53.3%</b>

## Most discriminative Y-23 systems in Lebanese population and villages

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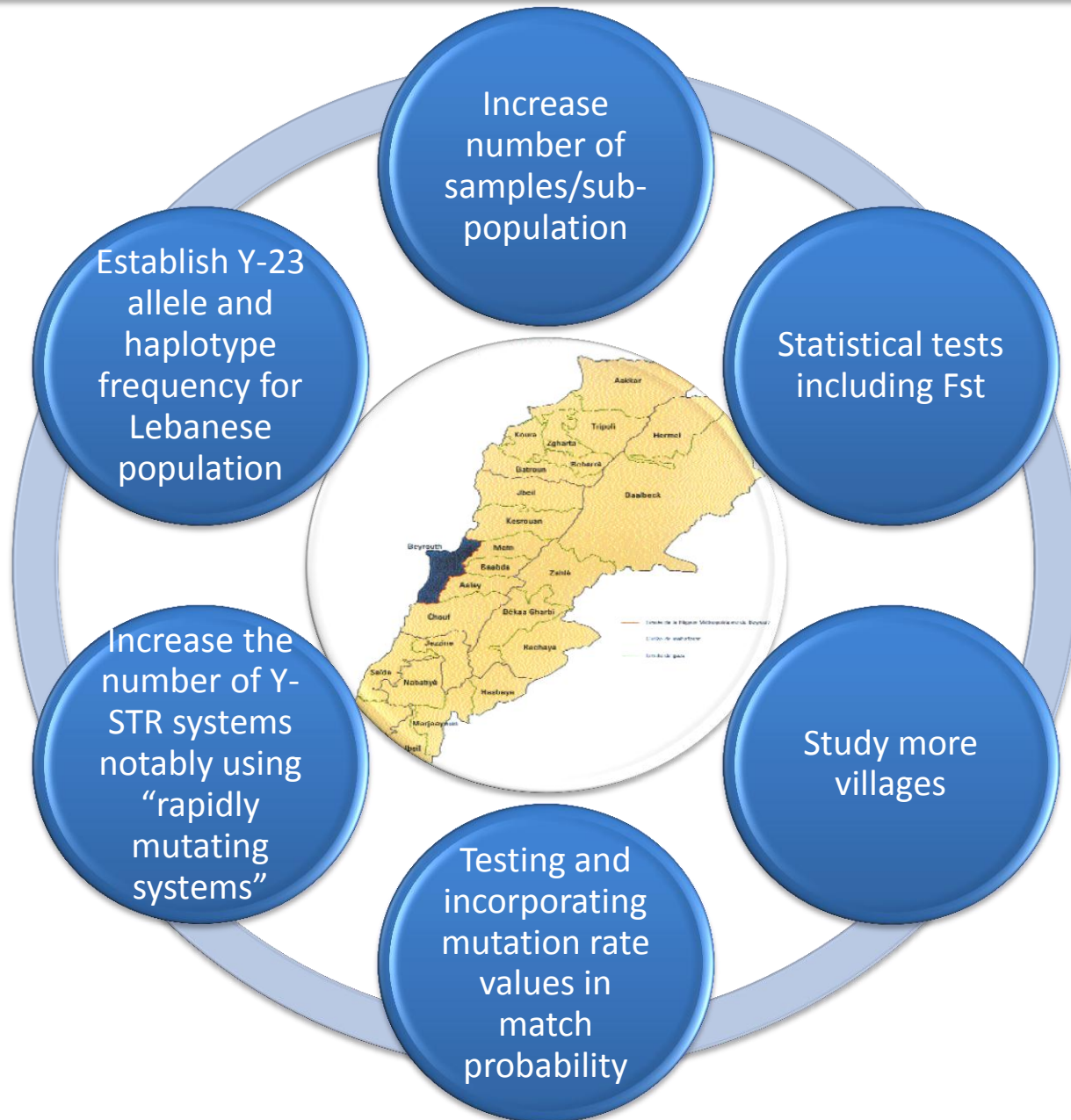
Number of haplotypes differentiated by the genetic system

System	Database	Villages	Total
DYS576	10	3	13
DYS481	10	0	10
DYS549	9	1	10
DYS533	5	2	7
DYS570	10	5	15
DYS643	1	1	2

## Conclusion for using Y-23 Kits

- **Numerous “non-related” individuals with common ancestors are resolved by their Y-23 STR profile.**
- **Common haplotypes are resolved in database much higher than in villages.**

# Future Studies



**Thank You**