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OMICS Group International is a pioneer and leading science event organizer, which publishes around 400 open access journals and conducts over 300 Medical, Clinical, Engineering, Life Sciences, Pharma scientific conferences all over the globe annually with the support of more than 1000 scientific associations and 30,000 editorial board members and 3.5 million followers to its credit.

OMICS Group has organized 500 conferences, workshops and national symposiums across the major cities including San Francisco, Las Vegas, San Antonio, Omaha, Orlando, Raleigh, Santa Clara, Chicago, Philadelphia, Baltimore, United Kingdom, Valencia, Dubai, Beijing, Hyderabad, Bengaluru and Mumbai.



Evaluation of Ethni-Test in the assessment of individual ethnic origin for forensic and medical applications

M. Al Salih

President, Medical Laboratory Director
& Forensic Technical Leader

Measuring Race and Ethnicity: Why and How?

Margaret A. Winker, MD

JAMA. 2004;292(13):1612-1614. doi:10.1001/jama.292.13.1612.

Abstract:

Race and ethnicity are constantly evolving concepts, deceptively easy to measure and used ubiquitously in the biomedical literature, yet slippery to pinpoint as definitive individual characteristics. A current dictionary definition of race is “a family, tribe, people, or nation belonging to the same common stock, or a class or kind of people unified by shared interests, habits, or characteristics.”¹ For 154 years, the US government has defined race for its census takers, and for many years census takers then defined it for US residents. The terms used reflect the nation’s changing demographics and increasing recognition of human diversity. The 1850 enumerators used a form that assumed a default race of white, with a checkmark indicating nonwhites as black or mulatto, with additional indications for free or slave.² Indian was added as a category in 1860. Since 1960, individuals have been able to specify their own race and ethnicity, and by 2000 the census enumerated 126 racial and ethnic categories.³



Why Measure Race/Ethnicity?



Applications for Ethnicity Testing

- ❖ Search for personalized genetic history (PGH)
- ❖ Used to adjust for population and admixture stratification
- ❖ Used to de-convolute environmental and genetic effects from complex diseases
- ❖ Important in medical risk analysis & personalized medicine
- ❖ Used in admixture mapping for socio political purposes
- ❖ Used in forensic investigations



Unusual Use of DNA Aided in Serial Killer Search

The New York Times: By NICHOLAS WADE

Published: June 3, 2003

In what appears to be the first use of DNA to extract details of a criminal suspect's appearance, investigators in the case of the Louisiana serial killer shifted their focus away from white suspects after an analysis of tissue from one of the crime scenes determined that the killer was probably black, the developer of the genetic test says.

DNA evidence has come into widespread use to identify individuals, but the identifying pieces of DNA are not part of the genes and have no influence on a person's physical makeup. Experts have long recognized that as knowledge of the human genome advances, other information could be extracted from DNA samples, including physical traits like race. The developer of the test used in Louisiana, Dr. Mark Shriver, a geneticist at Pennsylvania State University, said investigators had been searching for a white man, based on profiling information suggesting that most serial killers are white.

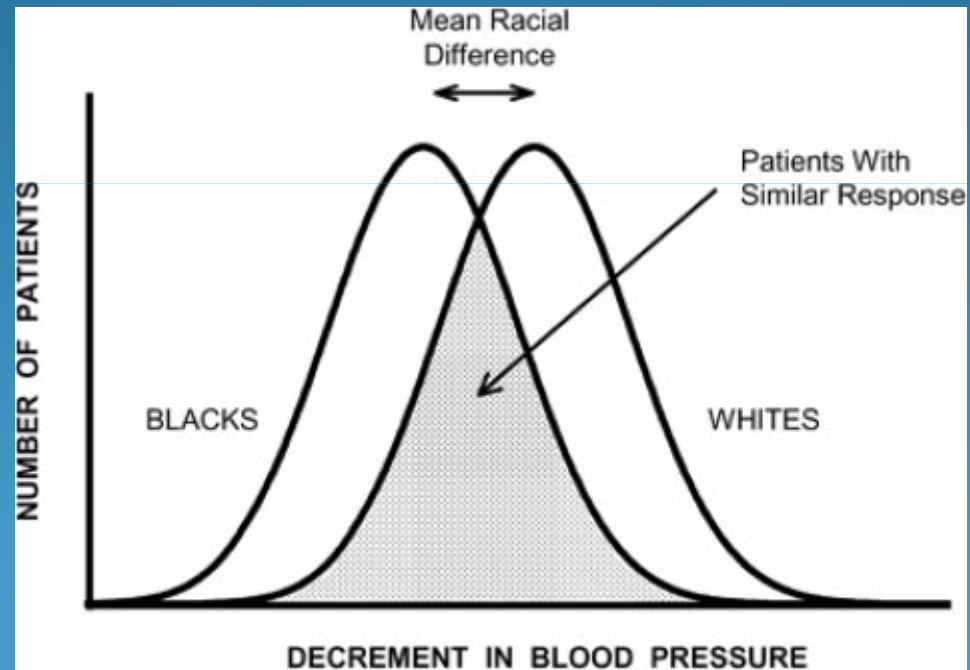
But then they sent DNA samples to DNAPrint Genomics, a company in Sarasota, Fla., that owns the rights to Dr. Shriver's test. Of 20 samples tested, Dr. Shriver said, only one was linked to the suspect, and the company was not told which. It typed the crime scene sample as being 85 percent African ancestry and 15 percent American Indian.

Police sketch of serial killer based on eyewitness accounts (left) and actual murder suspect, Derrick Todd Lee (right).
Courtesy Lafayette Parish Sherriff's Office and F.B.I.





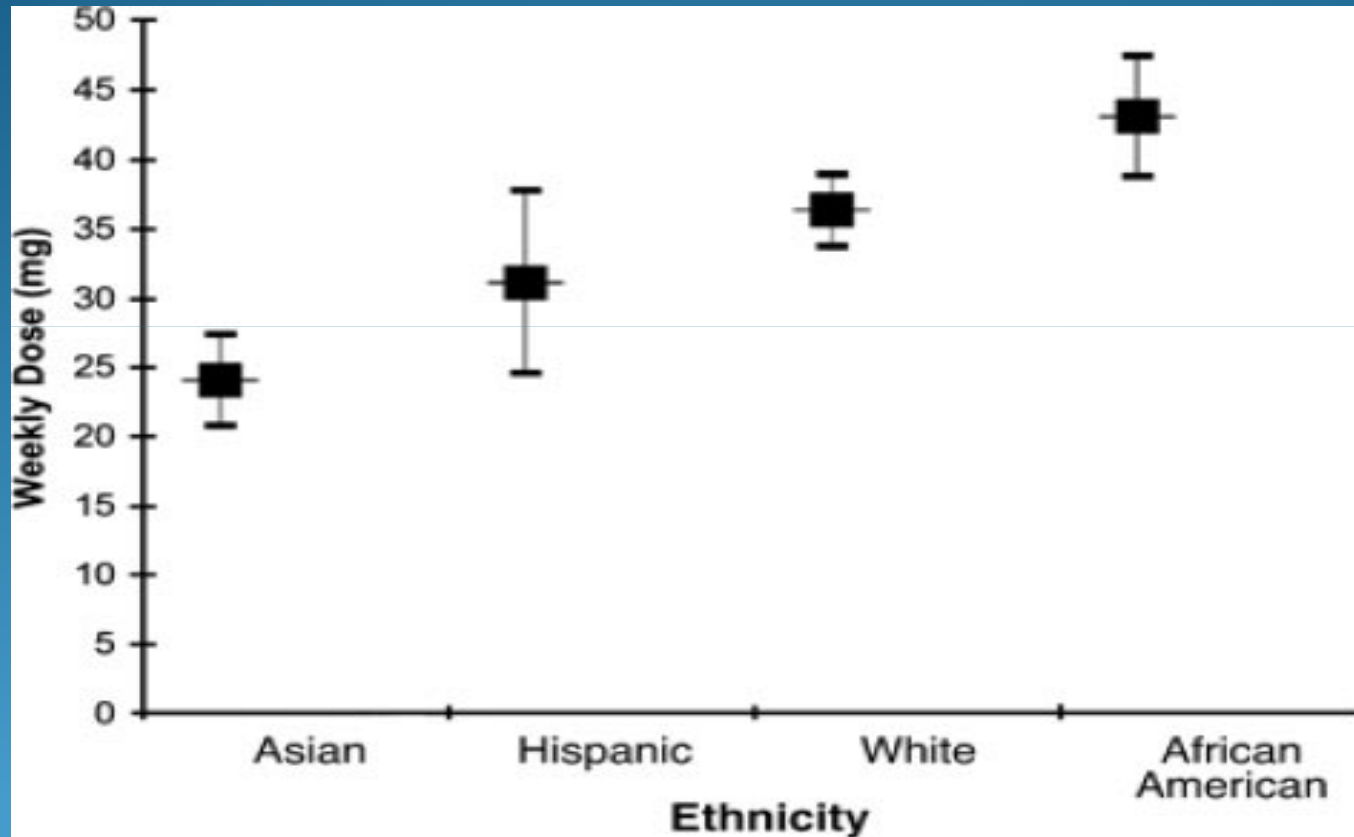
BP Measurements following admin of Antihypertensive drugs Circulation 2008:118 (1383-1393)





Average Warfarin dose requirements

Circulation 2008;118 pp1384.





BiDil: Assessing a Race-Based Pharmaceutical

Howard Brody, et al. *Annals of F. Medicine* (2006)

Abstract

Isosorbide and hydralazine in a fixed-dose combination (BiDil) has provoked controversy as the first drug approved by the Food and Drug Administration marketed for a single racial-ethnic group, African Americans, in the treatment of congestive heart failure. Family physicians will be better prepared to counsel their patients about this new drug if they understand a number of background issues. The scientific research leading to BiDil's approval tested the drug only in African American populations, apparently for commercial reasons, so the drug's efficacy in other



How do you Measure Race/ Ethnicity?



Methods Used in Measuring Ethnicity

- ❖ Creation of laws defining who is who?
1850-1860: White and non-white.
- ❖ Self-claimed reports of ethnicity? Problems:
 - a. Errors.
 - b. Large # of ethnicity groups (126 in Yr 2000)
- ❖ Genetic systems to infer ethnicity or ancestry:
Several systems: Problems: Limited on validation and availability of allele frequency databases.



Genetic Systems for Inferring Ethnicity

- ❖ STR
- ❖ Autosomal SNPs
- ❖ AIM-INDELS
- ❖ Y-chromosome markers (STR, SNP)
- ❖ Mitochondrial DNA sequences
- ❖ Optical Emission Spectroscopy for chemical hair analysis



STR vs Autosomal SNPs

Characteristics	STR	Auto SNPs
Power of discrimination	High	Low
Admixture resolution	high	Low
Mutation rate	High	Low
FST Values	Low	High
Database availability	High	Low
# of markers needed	Small	Large
Disease association	None-low	High



*ARID5B Genetic Polymorphisms Contribute to Racial
Disparities in the Incidence and Treatment Outcome of
Childhood Acute Lymphoblastic Leukemia*

*Heng Xu, Cheng Cheng, Meenakshi Devidas, Deqing Pei, Yiping Fan, Wenjian Yang, Geoff Neale, Paul Scheet,
Esteban G. Burchard, Dara G. Torgerson, Celeste Eng, Michael Dean, Frederico Antillon, Naomi J. Winick,
Paul L. Martin, Cheryl L. Willman, Bruce M. Camitta, Gregory H. Reaman, William L. Carroll, Mignon Loh,
William E. Evans, Ching-Hon Pui, Stephen P. Hunger, Mary V. Relling, and Jun J. Yang*

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SNPs in Pharmacogenomics?

1 Project

2 Setup

3 Data

4 Results

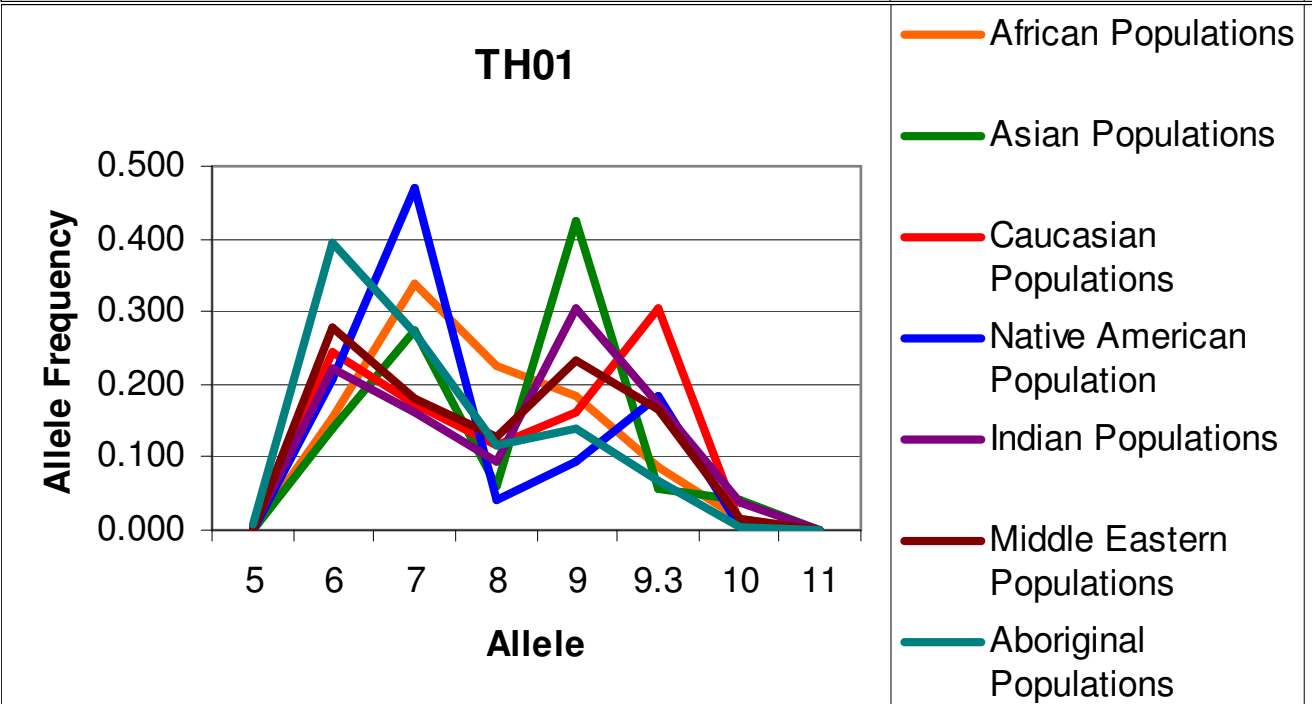
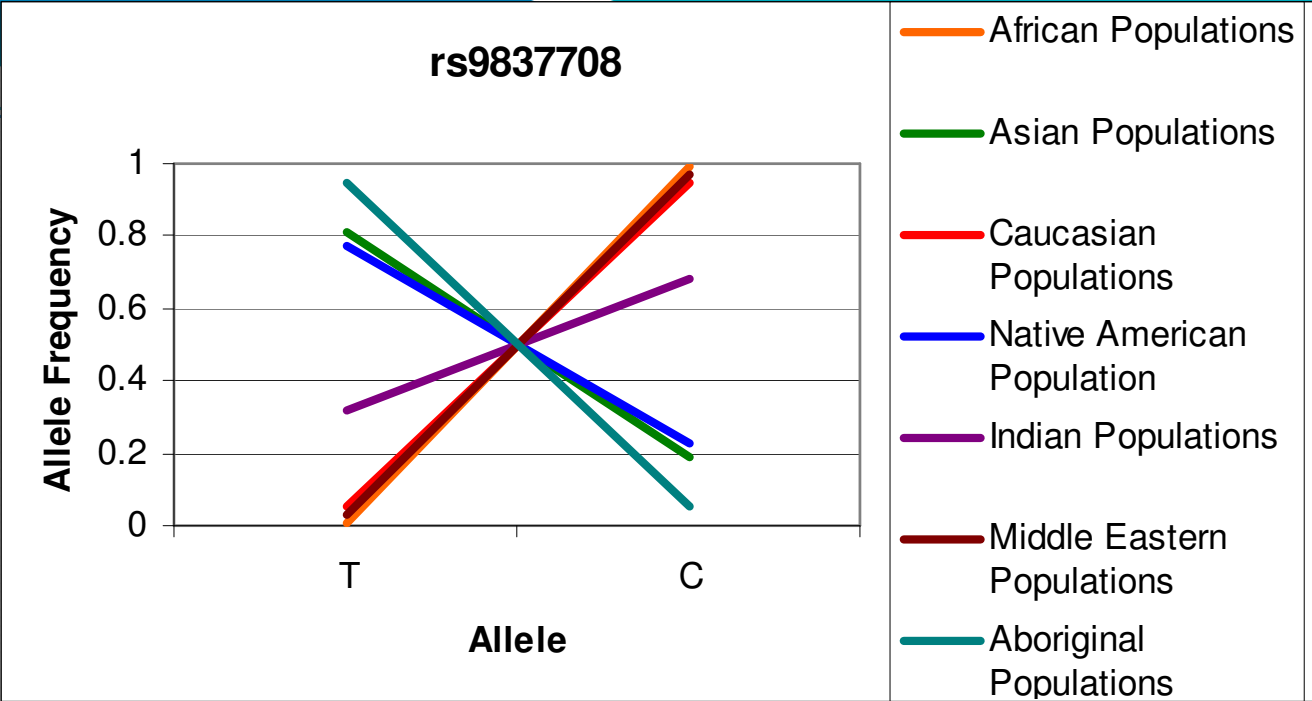
Results

Summary Report

Detailed Report

Export

Sample ID	CYP2D6	CYP2D6_ex9 Hs00010001_Cn	-1584C>G C_32407252_30	100C>T C_11484460_40	137_138i C_324072
NA17217	*1/*41	2	G/G	G/G	-/-
NA17221	*1/*2Ax2, *1x2/*2A	3	G/C	G/G	-/-
NA17223	*1/*1	2	G/G	G/G	-/-
NA17225	*4/*4	2	G/G	A/A	-/-
NA17227	*1/*9	2	G/G	G/G	-/-
NA17228	*1/*1	2	G/G	G/G	-/-
NA17232	*2A/*2Ax2	3	C/C	G/G	-/-
NA17237	*1/*4	2	G/G	A/G	-/-
NA17239	*1/*4	2	G/G	A/G	-/-
NA17242	*1/*1	2	G/G	G/G	-/-



Definitions

Inferred Genetic Ancestry: In inferred genetic ancestry we usually consider 40-60 generations back.

Inferred Genetic Ethnicity: For inferring genetic ethnicity we usually consider 4-6 generations back.



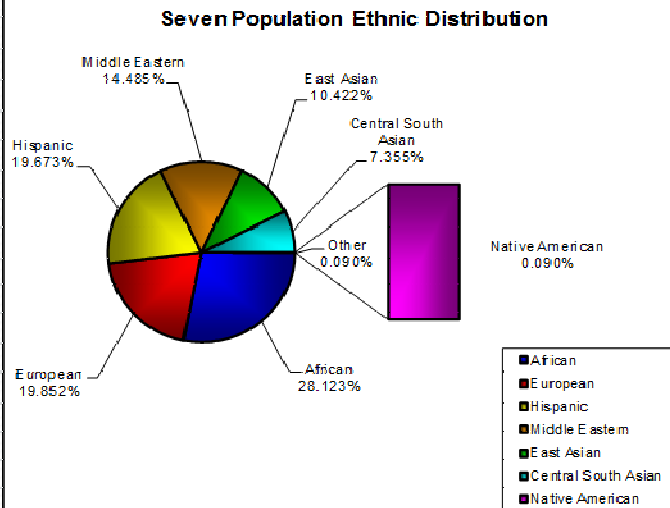
Short Tandem Repeat (STR) Advanced Ethnic Distribution Certificate

Case Number
Unknown

Unknown,

We have tested fifteen STR locations present on your DNA and have critically compared this data to worldwide databases. Your DNA profile and probabilities of genetic ethnicity are located below.

DNA PROFILE	
LOCUS	ALLELE
D3S1358	15,17
TH01	7,7
D21S11	28,31,2
D18S51	15,16
Penta E	5,14
D5S818	11,13
D19S317	9,12
D7S820	8,9
D16S539	10,13
CSF1PO	11,11
Penta D	9,14
vWA	16,18
D8S1179	11,12
TPOX	9,10
FGA	22,25



OVERALL ETHNIC DISTRIBUTION	
African	28.12%
European	19.85%
Hispanic	19.67%
Middle Eastern	14.49%
East Asian	10.42%
Central South Asian	7.35%
Native American	0.09%

The STR test performed on your DNA indicates that you are most likely to be of African origin.

The pie chart and ethnic distribution table to the left represents your overall major ethnic distribution probability. The percentages are representative of that particular ethnicity, based upon a specified 7 population distribution.

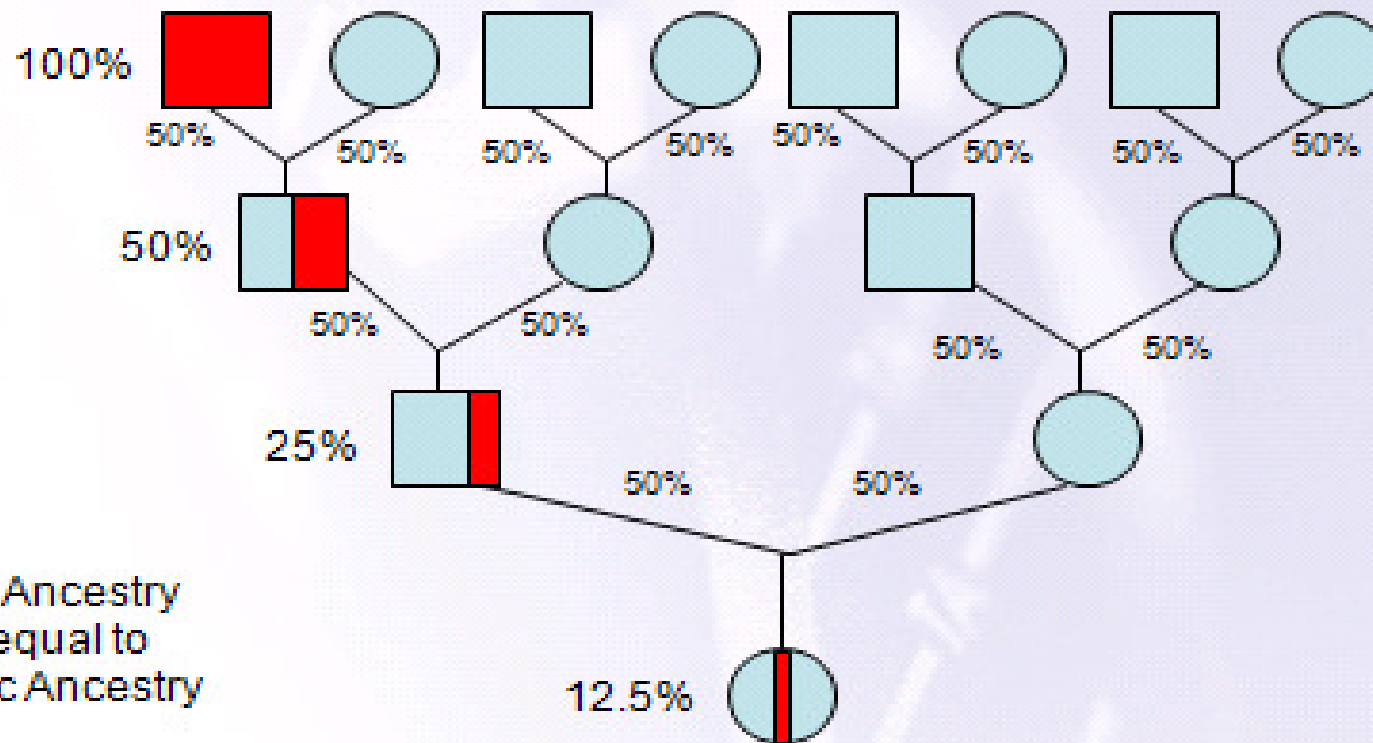
I certify with my signature the authenticity and accuracy of this certificate.

Dr. M. Al. Salih
DNA Reference Laboratory Director

Unknown
Date

NATIVE AMERICAN DISTRIBUTION	EUROPEAN DISTRIBUTION	AFRICAN DISTRIBUTION	EAST ASIAN DISTRIBUTION	CENTRAL SOUTH ASIAN DISTRIBUTION	LATIN AMERICAN DISTRIBUTION						
Huichol (Choles)	61.35%	European United Kingdom	21.36%	Central West African	29.09%	Southeast Asian Continental	43.38%	Shona	57.53%	Central South American	88.65%
Michigan Natives	32.39%	Central East European	15.78%	South African	28.44%	Southeast Asian Islander	41.85%	Vanya	19.99%	Northern South American	5.66%
Mayan, Surul, Karitania	3.96%	Far Eastern European Russian	15.45%	Central East African	19.34%	Chinese	8.75%	Dravidian	16.52%	Caribbean Hispanic	3.17%
Minnesota Natives	2.28%	West Continental European	12.89%	North African	11.73%	Japanese	1.84%	East Asian Thai	3.06%	Southern South American	1.01%
Apache	0.02%	South Eastern European	12.18%	North Coastal West African	11.41%	Mongolian	1.42%	Pakistani-Afghanistan	1.54%	Central American	0.88%
Sallishan	0.00%	East European Anatolia	10.78%			Taiwanese Filipino	1.19%	Somali-Peoples	0.83%	Mexican	0.63%
Saskatchewan	0.00%	European Scandinavian	7.75%			Korean	0.91%	Nepali	0.46%		
Navajo	0.00%	European Iberian	3.81%			Spanish	0.67%	Bengal-Asian, Indian	0.07%		
The table above specifies the closest your Native American ethnicity is to the data. By comparing your DNA profile to the data, you reveal 27 African ethnicities in the Ethnic Distribution Chart. The closest match is Central West African (29.09%). Other matches include South African (28.44%), Central East African (19.34%), North African (11.73%), and North Coastal West African (11.41%).	The table above specifies the closest your European ethnicity is to the data. By comparing your DNA profile to the data, you reveal 27 European ethnicities in the Ethnic Distribution Chart. The closest match is European United Kingdom (21.36%). Other matches include Central East European (15.78%), Far Eastern European Russian (15.45%), West Continental European (12.89%), South Eastern European (12.18%), East European Anatolia (10.78%), European Scandinavian (7.75%), and European Iberian (3.81%).	The table above specifies the closest your East Asian ethnicity is to the data. By comparing your DNA profile to the data, you reveal 43 East Asian ethnicities in the Ethnic Distribution Chart. The closest match is Southeast Asian Continental (43.38%). Other matches include Southeast Asian Islander (41.85%), Chinese (8.75%), Japanese (1.84%), Mongolian (1.42%), Taiwanese Filipino (1.19%), Korean (0.91%), and Spanish (0.67%).	The table above specifies the closest your Central South Asian ethnicity is to the data. By comparing your DNA profile to the data, you reveal 7 Central South Asian ethnicities in the Ethnic Distribution Chart. The closest match is Shona (57.53%). Other matches include Vanya (19.99%), Dravidian (16.52%), East Asian Thai (3.06%), Pakistani-Afghanistan (1.54%), Somali-Peoples (0.83%), Nepali (0.46%), and Bengal-Asian, Indian (0.07%).	The table above specifies the closest your Latin American ethnicity is to the data. By comparing your DNA profile to the data, you reveal 12 Latin American ethnicities in the Ethnic Distribution Chart. The closest match is Central South American (88.65%). Other matches include Northern South American (5.66%), Caribbean Hispanic (3.17%), Southern South American (1.01%), Central American (0.88%), and Mexican (0.63%).							
				MIDDLE EASTERN DISTRIBUTION							
				Mesopotamian	61.20%						
				Levantine Jewish	25.89%						
				Asian Arabian	8.90%						
				Levantine Arab	4.00%						

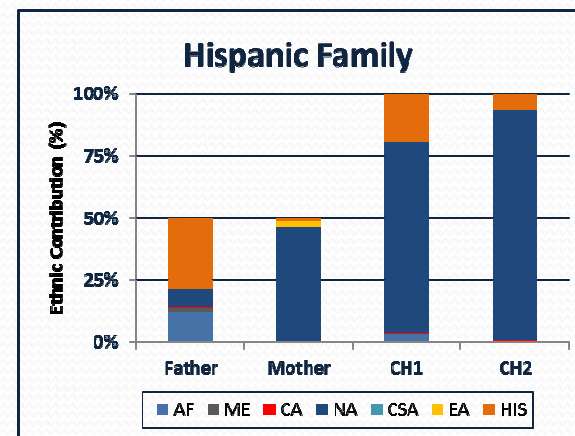
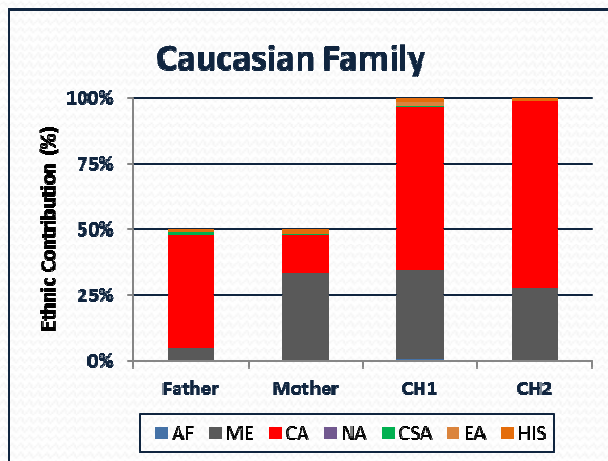
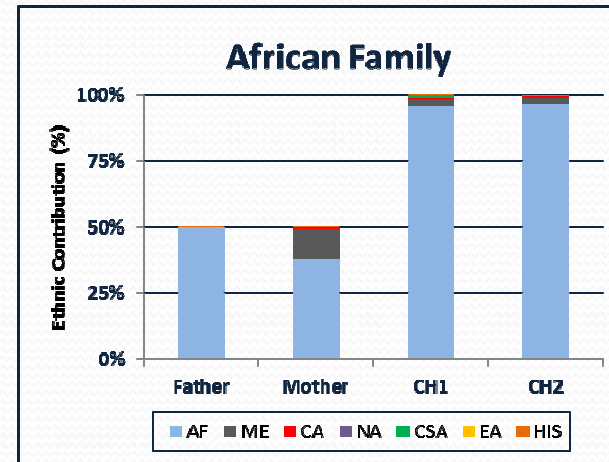
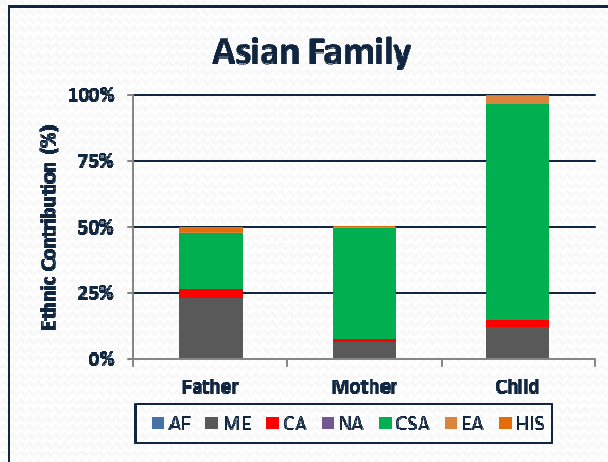
- Each generation one half of genome is passed down
- Lose 50% of genetic information / generation



Actual Ancestry
Not equal to
Genetic Ancestry

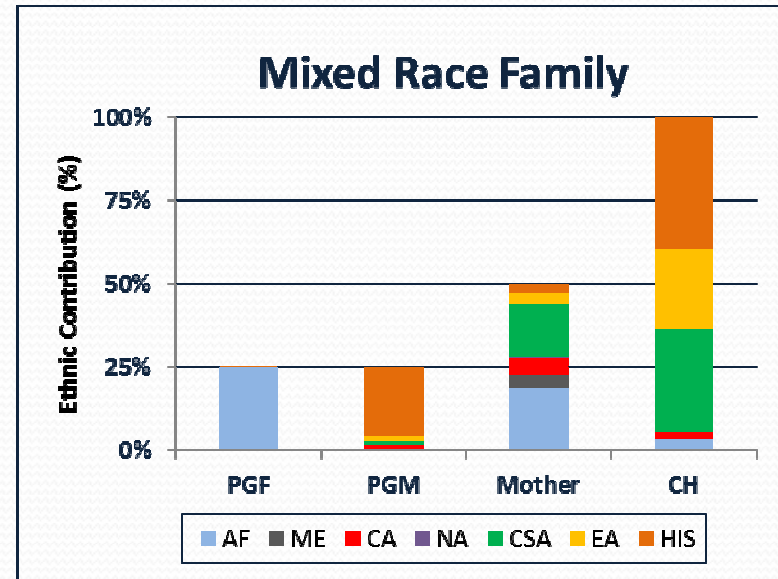
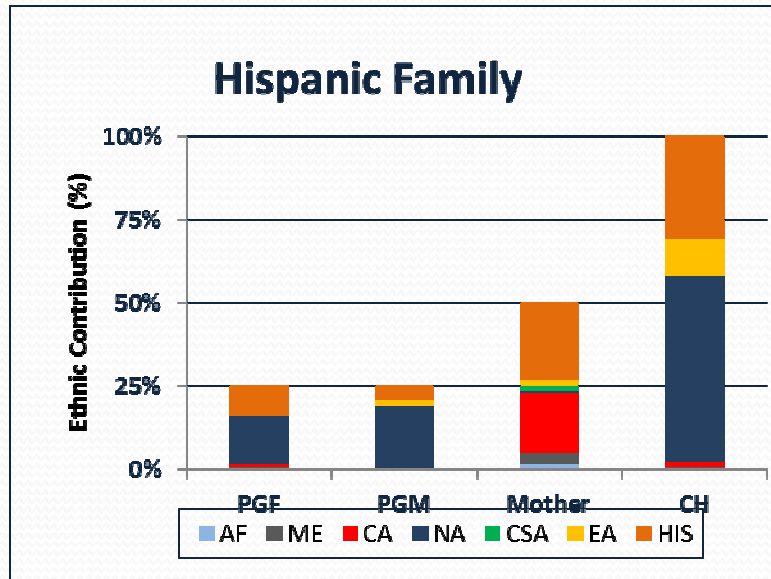
Validation Studies

Contribution of genetic ethnicity from Parents to children

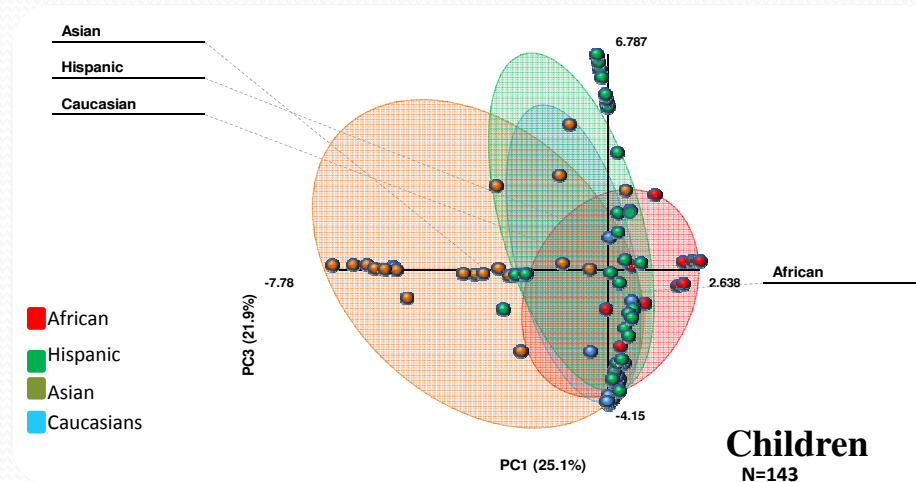
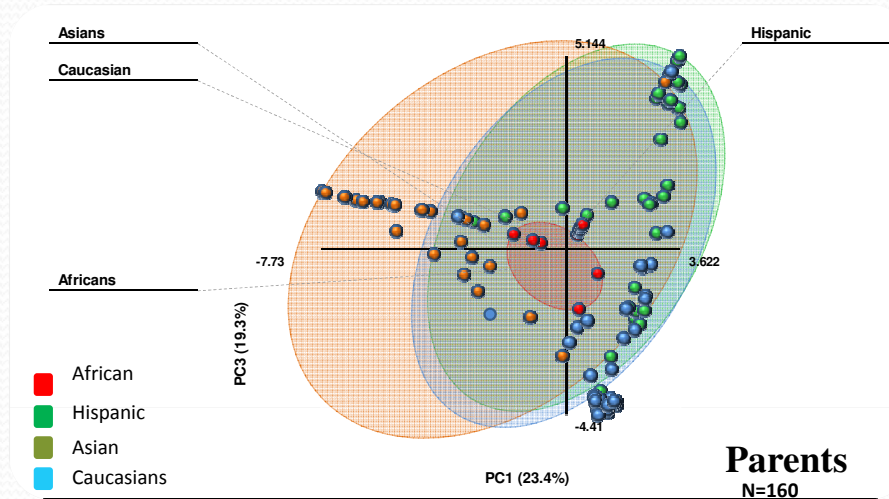


Validation Studies

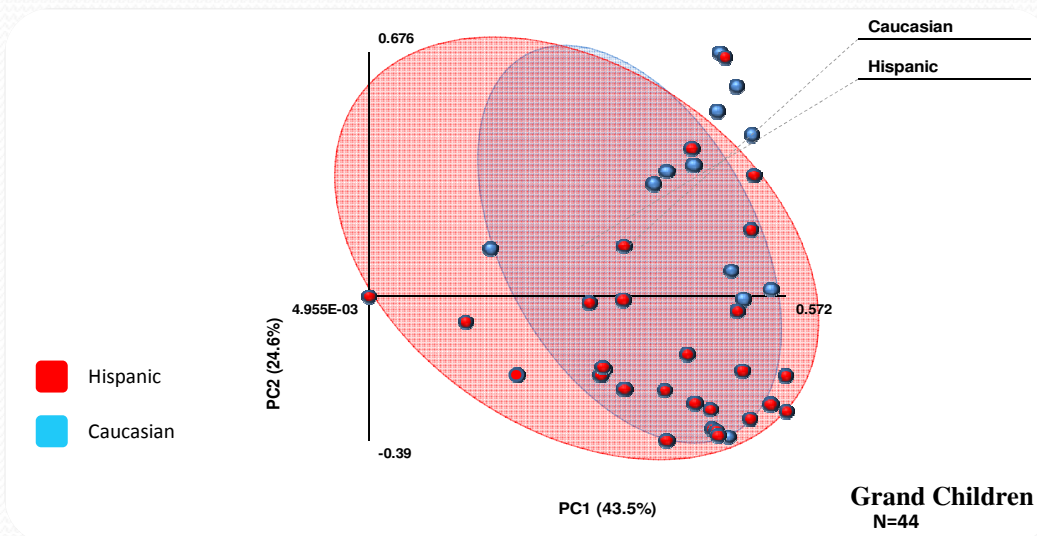
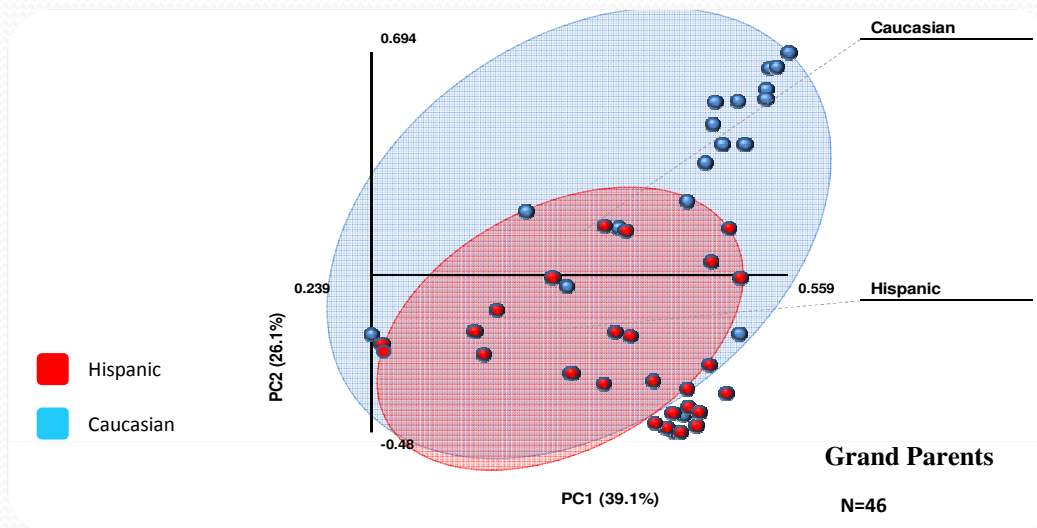
Contribution of genetic ethnicity from grandparents to grandchildren



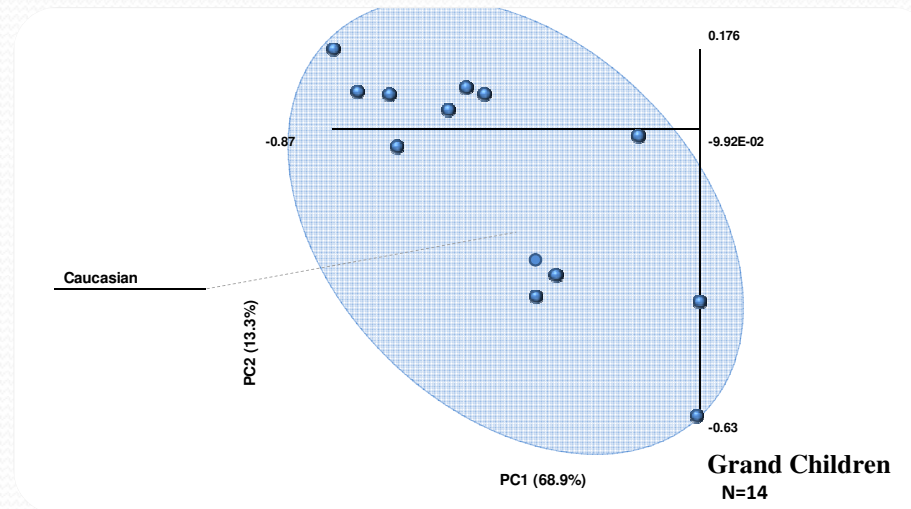
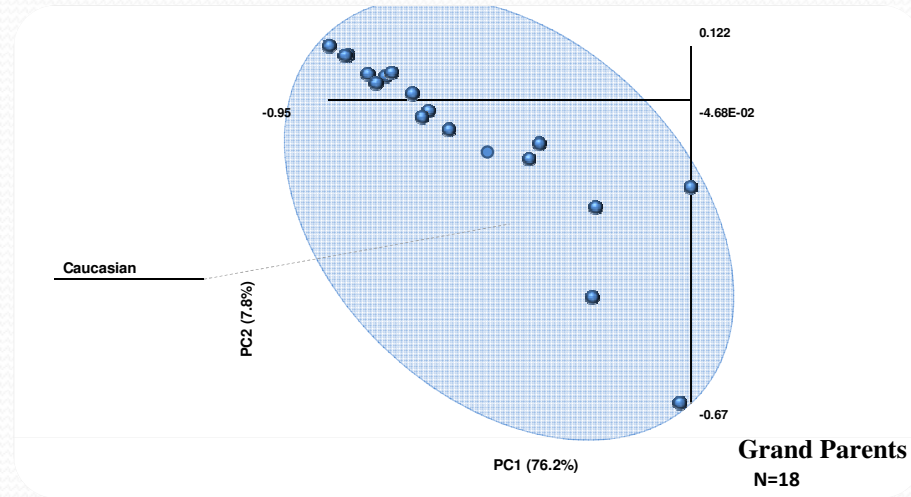
PLS comparison between Parents and their children for Africans, Asians, Caucasians and Hispanics



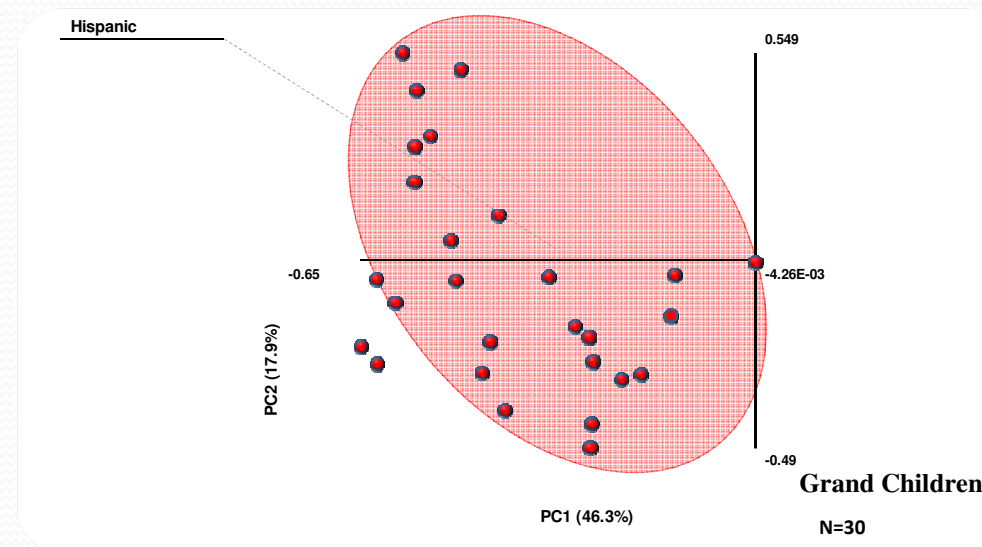
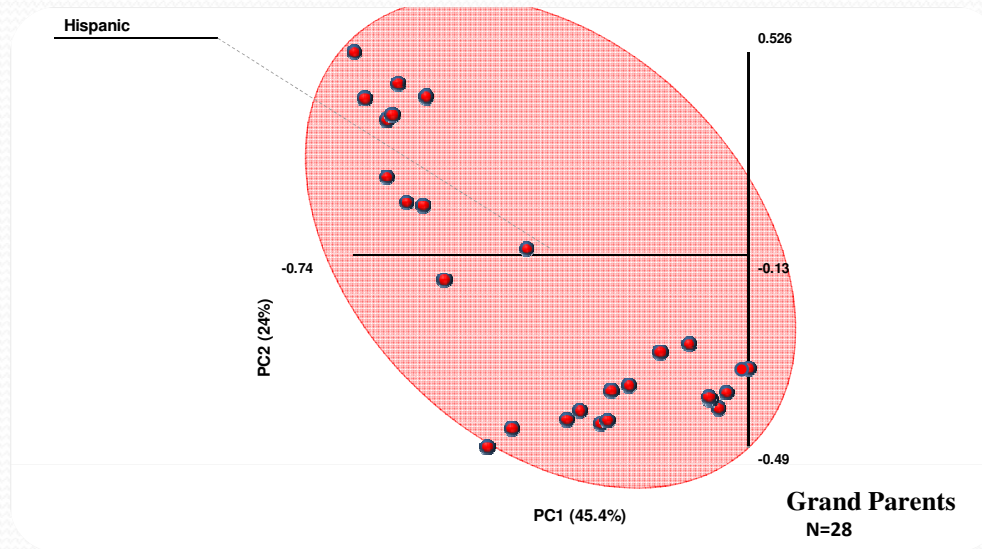
PLS comparison between Grandparents and their Grandchildren for both Caucasians and Hispanics



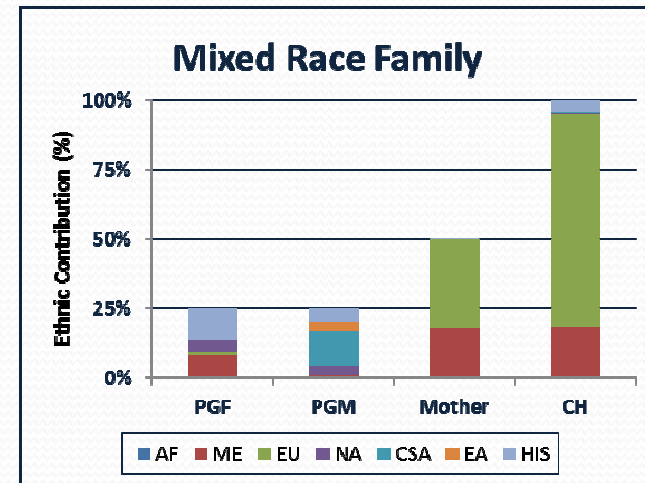
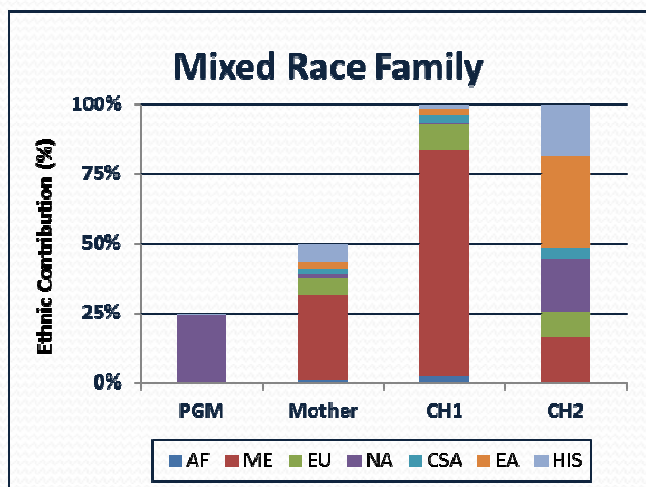
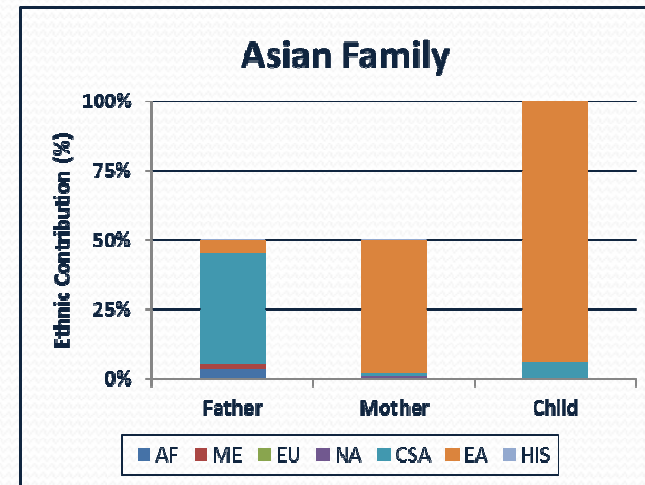
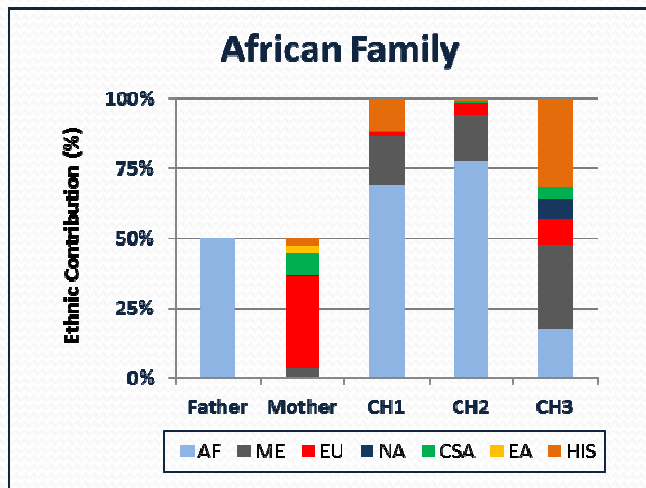
PCA comparison between Caucasians Grandparents and their Grandchildren



PCA Cluster comparison between Hispanic Grandparents and their Grandchildren

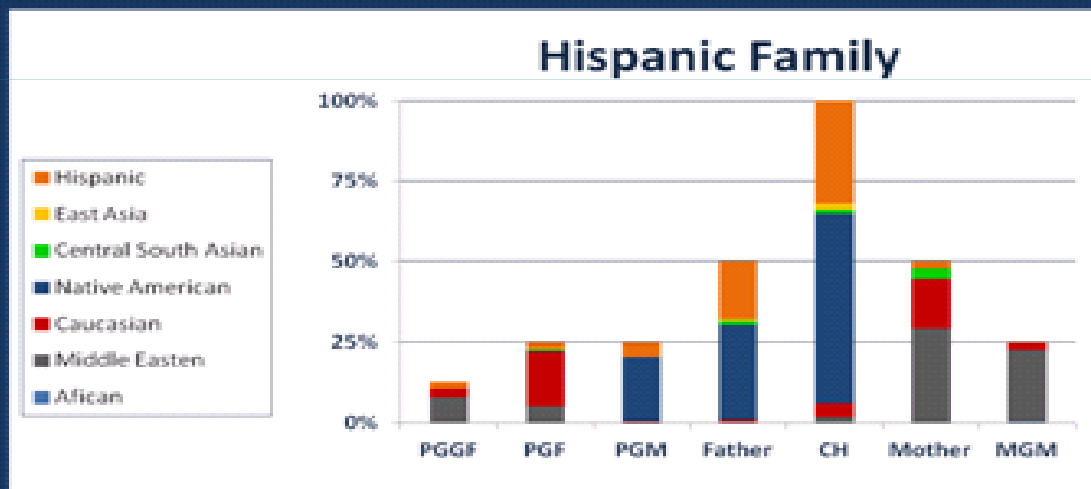


Parents/ children and grandparents/children combinations that do not conform to classical Mendelian genetic inheritance. Discrepancy?



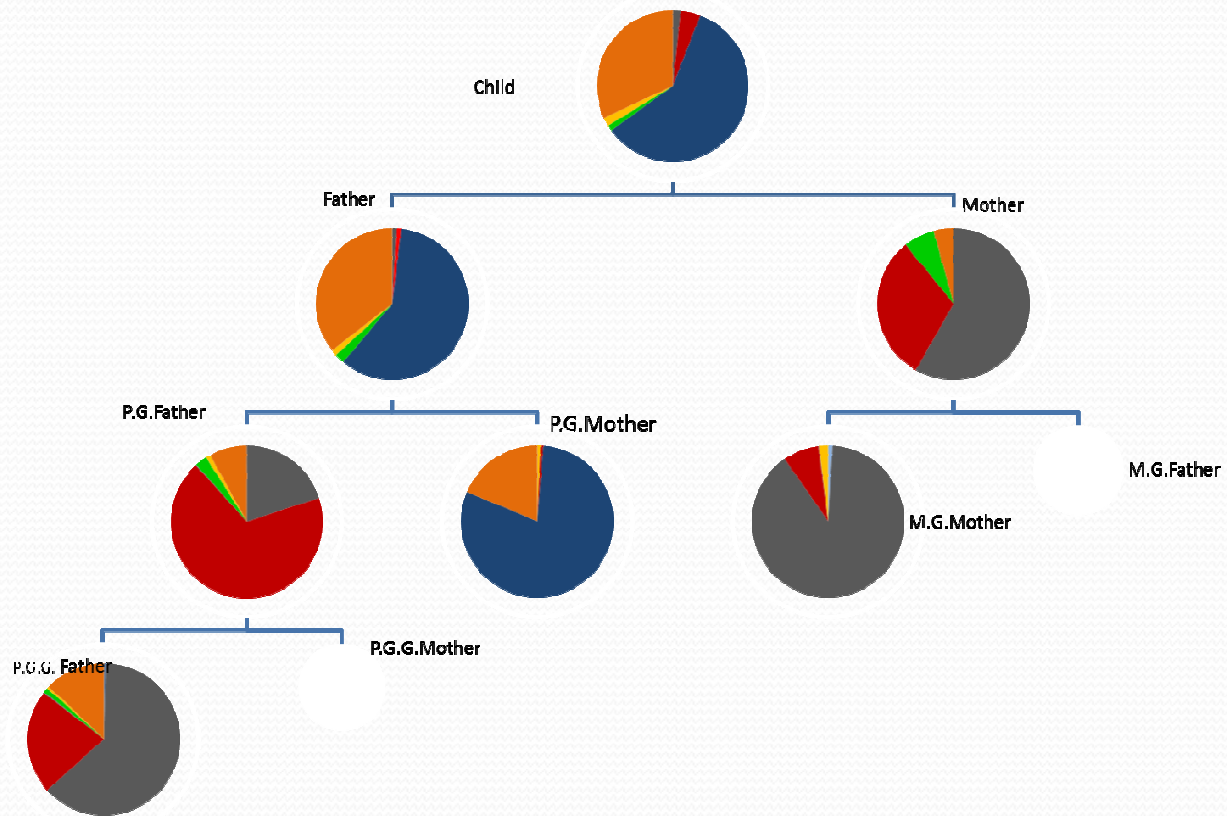


Ethnicity determination in pedigree studies



Discrepancies

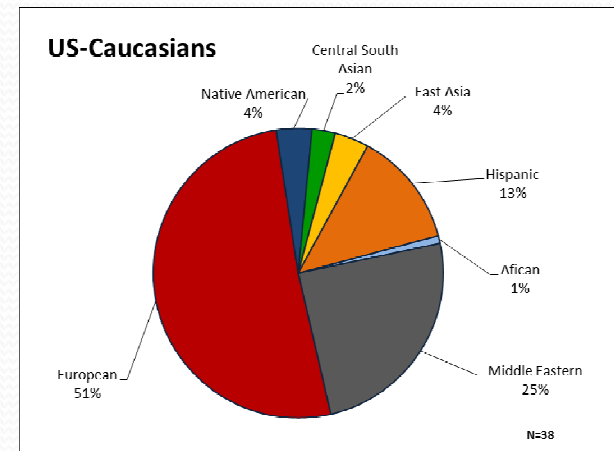
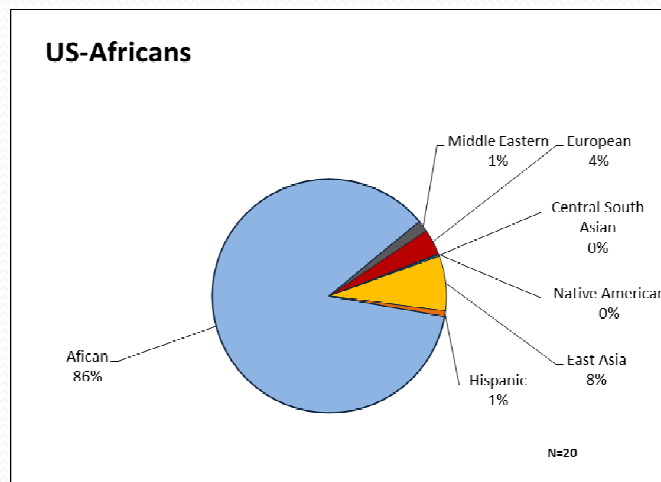
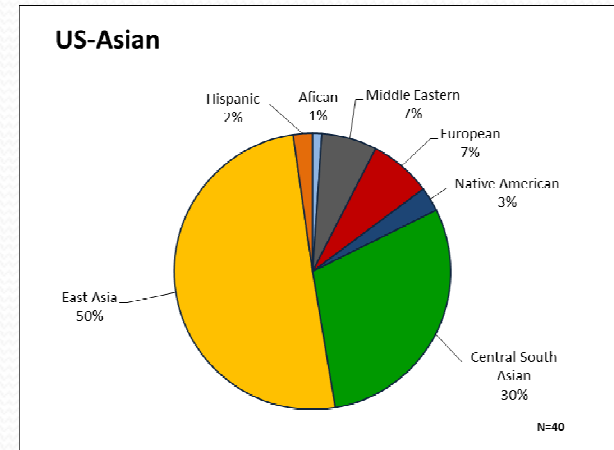
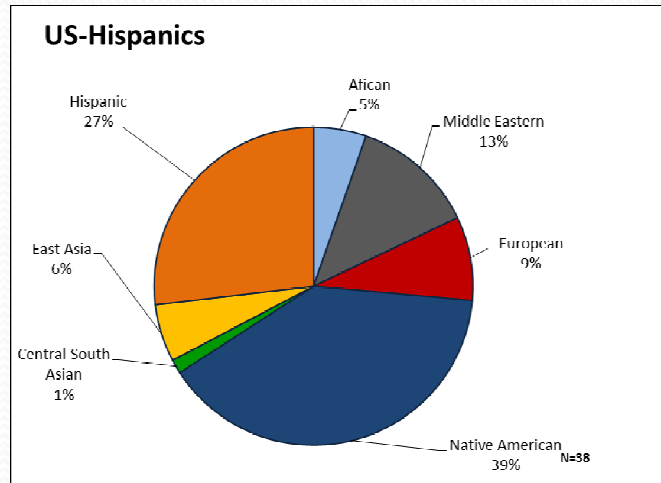
Extended family pedigree studies. Possible role for genetic selection!





Self-claimed ethnicity vs EthniTest

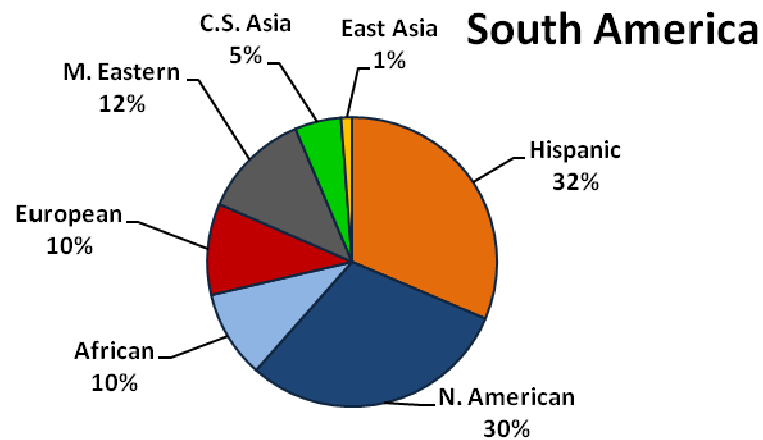
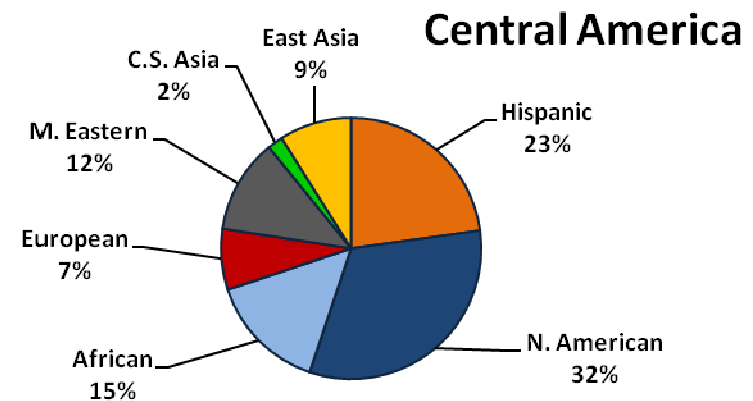
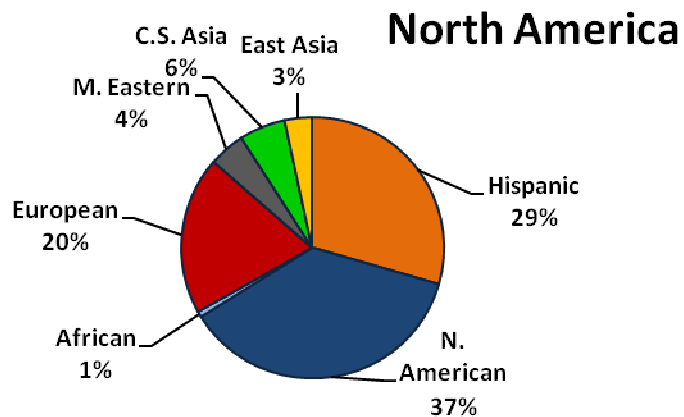
Self-reported race vs Ethnites analysis of major US ethnic populations



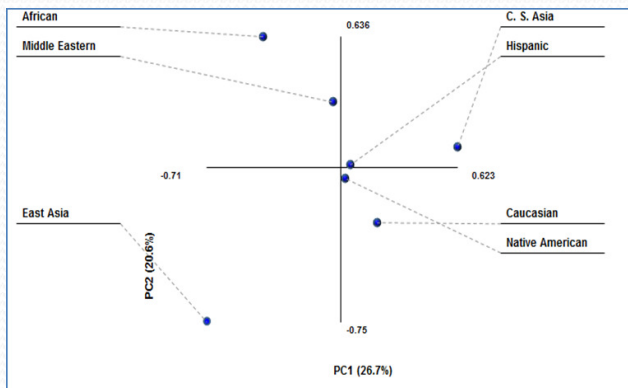
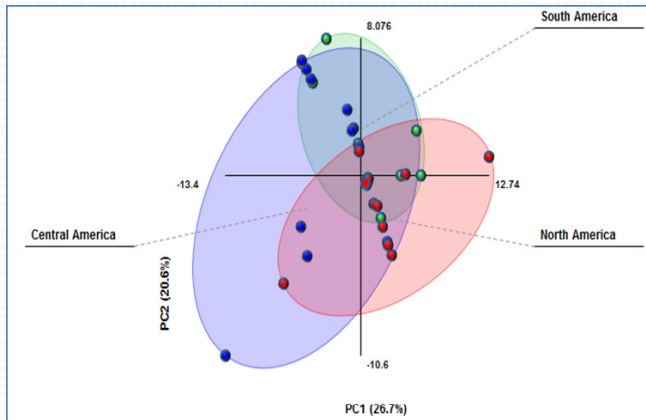


Geographical Distribution of Hispanics and Africans

Individual contributions of various ethnic backgrounds in Hispanic from North, Central & South America

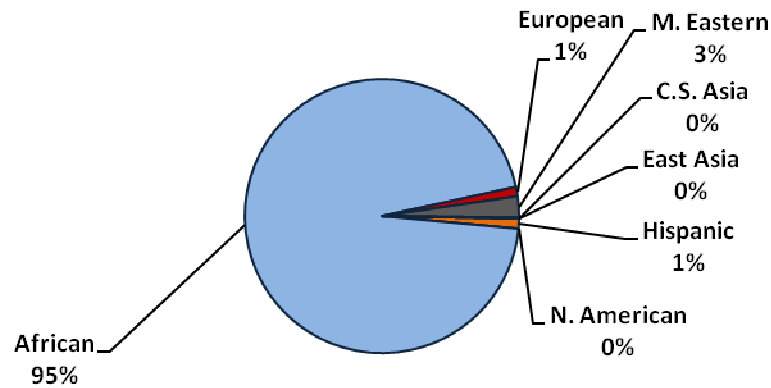


Analysis of self-claimed Hispanics from different geographical locations

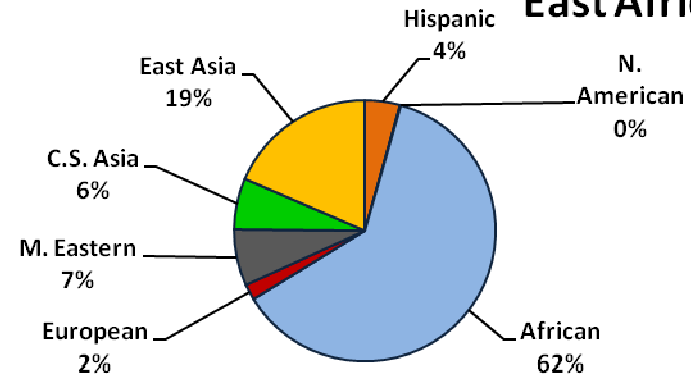


Individual contributions of various ethnic backgrounds in Africans from North, East & West Africa

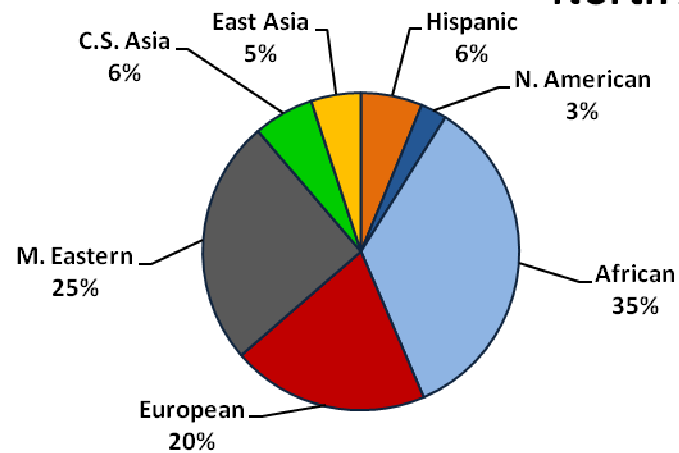
West Africa



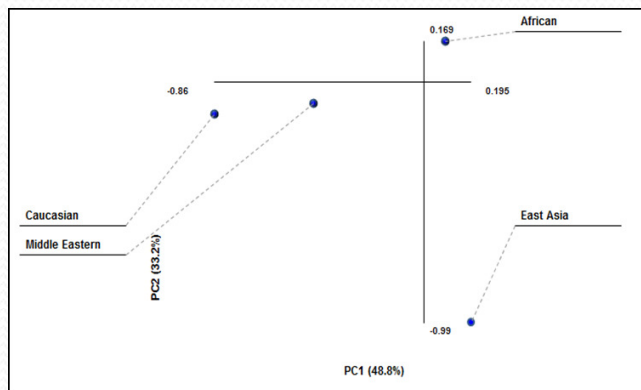
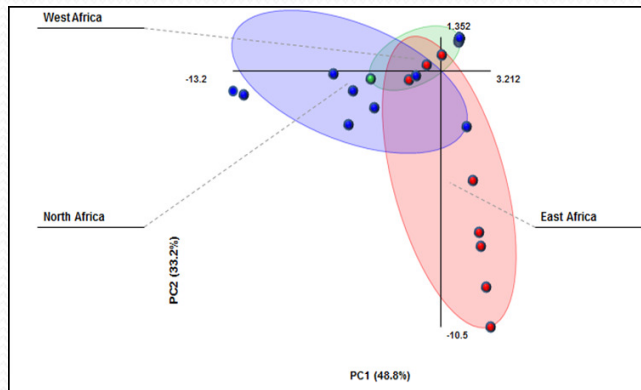
East Africa



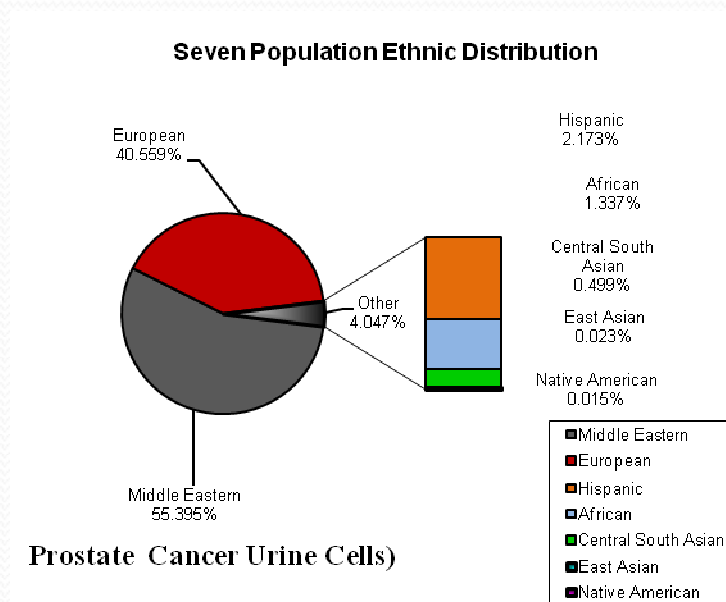
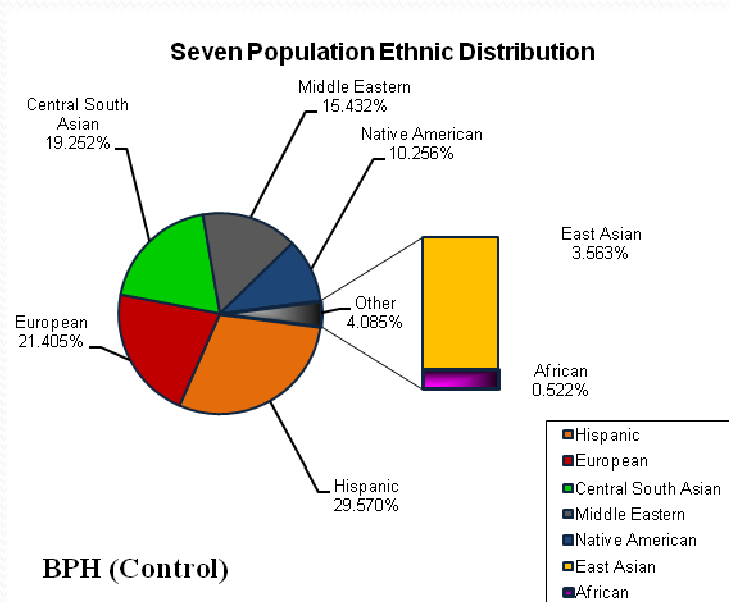
North Africa



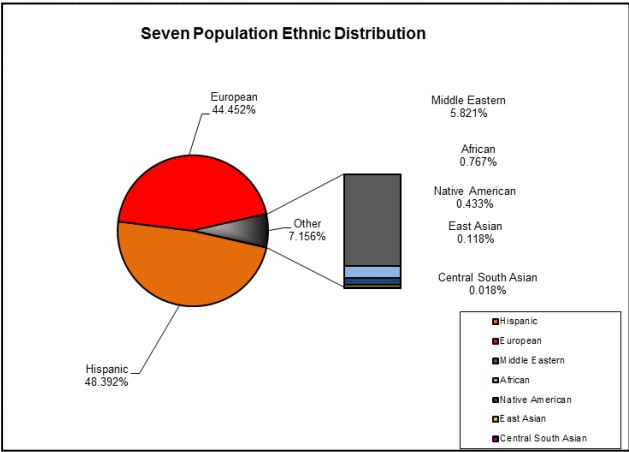
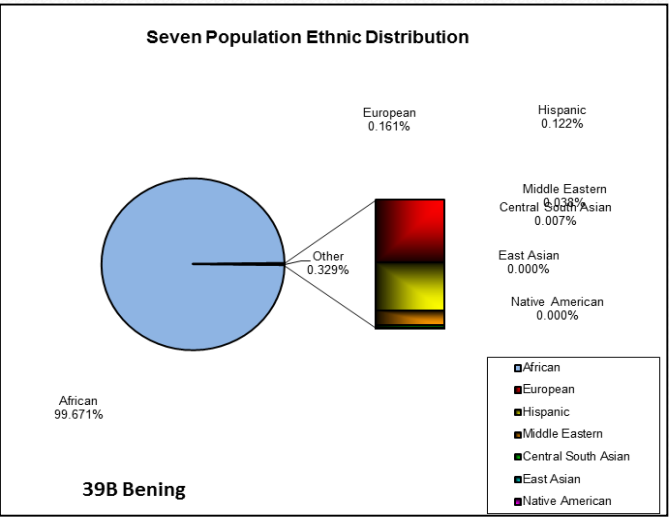
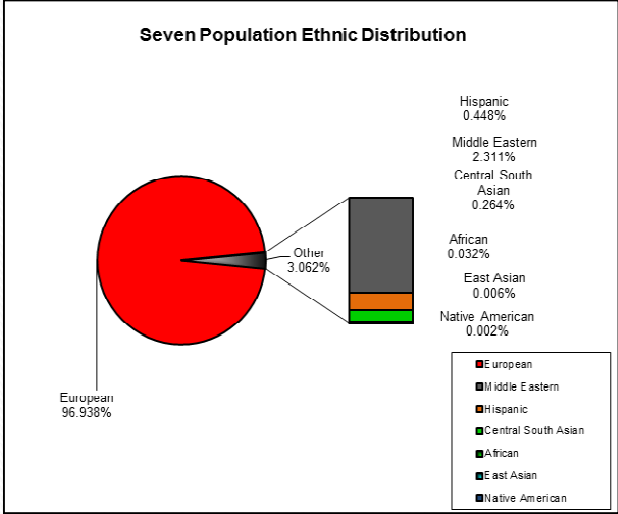
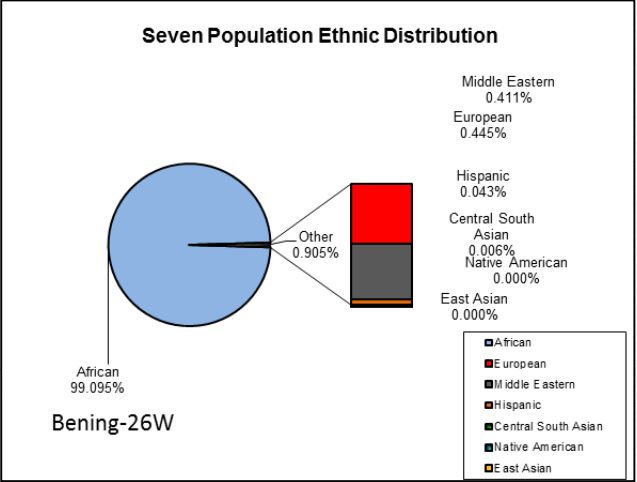
Analysis of self-claimed Africans from different geographical locations



Ethnic distribution Comparison between BPH (control sample) and Prostate Cancer Urine cells of an Individual



Prostate cancer vs control samples. Disparity in ethnic genetic background!





Acknowledgment

Kevin Ray Condel, MS

Quality Assurance Manager and Supervisor for
The DNA Unit at Wyoming State Crime Laboratory

Lizmery S Ferguson, MS

Forensic DNA Analyst DRL



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