## Genetic Identity and Community

DTC Genetic Testing Workshop The National Academies' August 31 - September 1, 2009

# the power of DNA:

Discovering Lost and Hidden Relationships

How DNA analysis techniques are assisting in the great search for our ancestors

The Sorenson Molecular Genealogy Foundation (SMGF)

#### The Mission of the SMGF

 To change the way that we think about each other, and hopefully the way we act towards each other, by showing that we are really one great human family



James L. Sorenson

- By creating a comprehensive database of the families of the world, using correlated genealogical and genetic information
- And providing the tools needed to reconstruct genealogies using DNA
- Make the data available to the public

#### **SMGF Database**

#### DNA Samples

- 108,000 (informed consent)
- ~170 countries

#### Extended Pedigrees

- 90,828 (11 gens average)
- 7.8 m names (2.6 m unique)

#### mtDNA sequences

- 76,766 D-loop
- 315 Complete

#### Y haplotypes

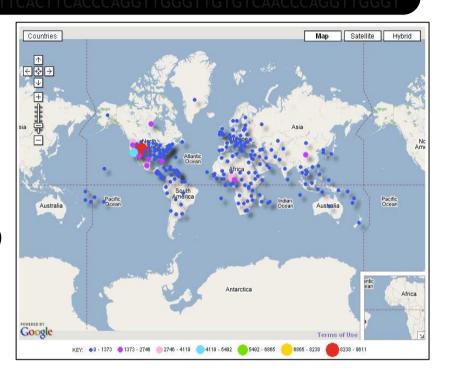
39,264 STR (complete at 37 loci)

#### X haplotypes

73,394 STR (complete at 13 loci, 3 linkage groups)

#### Autosomal genotypes

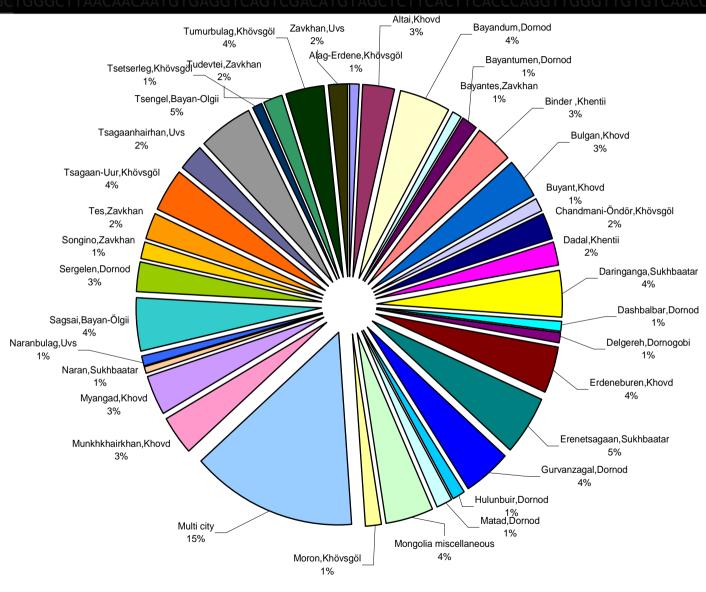
- 78,568 STR (68.5 average)
- 300 SNP (Affymetrics 6.0)



## Collection in Mongolia



#### Sample Collection Locations



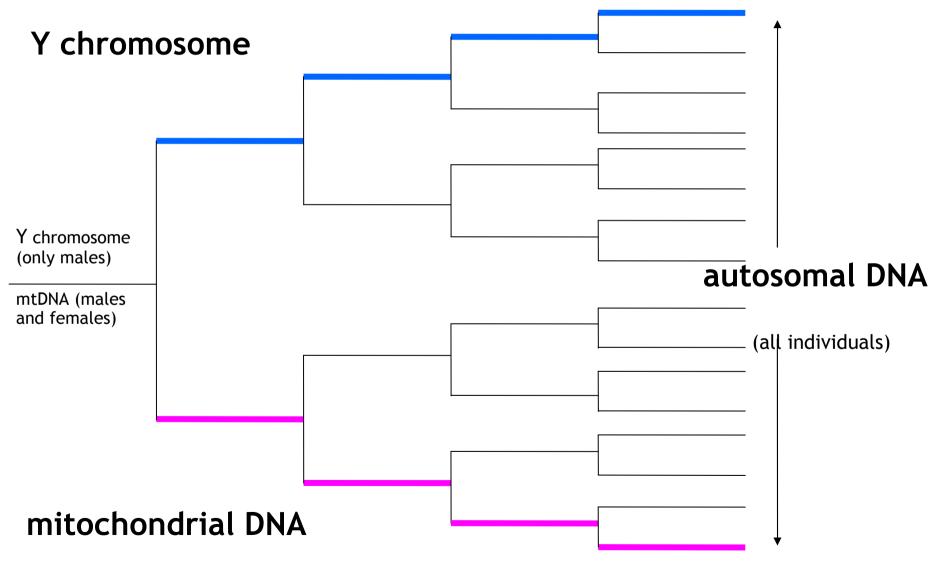
#### Informed Consent

- Description of Study
- Procedures
- Risks
  - Linking genetic results to you
  - Loss of confidentiality
- Benefits
- Payments
- Voluntary Participation/Withdrawal
- Confidentiality
- Use of data
- Funding

#### Issues

- Privacy vs. self-awareness
- Who owns genetic (and genealogical or any other 'personal' data)?
  - Secondary participants
- Discrimination and effectiveness of GINA
- Social Networks based on DTC genetic testing results
  - mt and Y 'haplogroups'
  - Ethnicity

## Genetic Systems and Genealogy

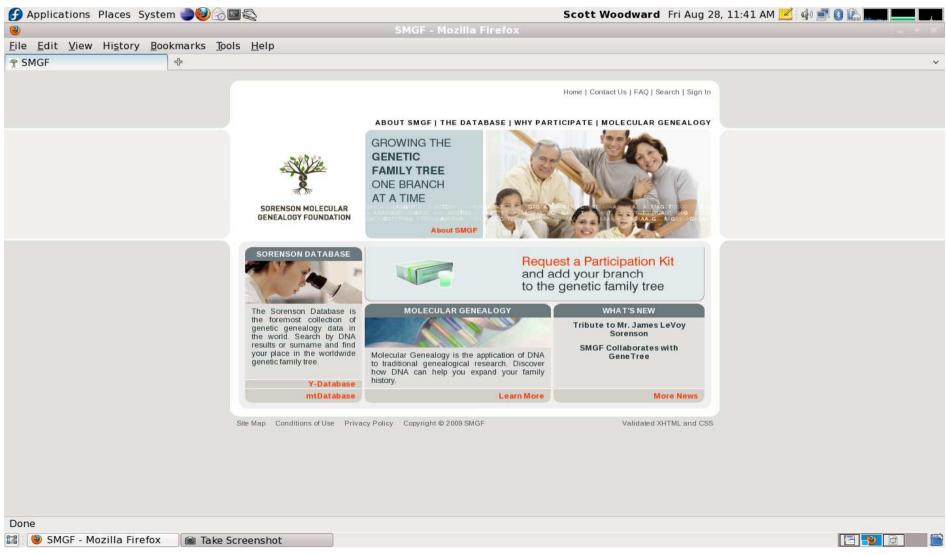


## Who (What) am !?

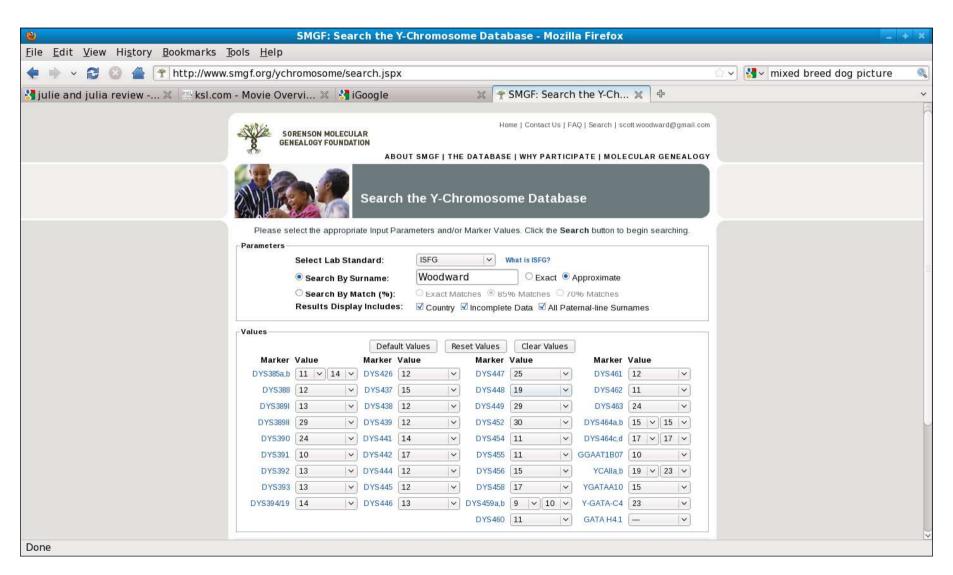
- 2009
  - Utah, USA 100%
- 1956
  - Virginia, USA 100%
- 1933
  - Utah, USA 100%
- 1900
  - Utah, USA 75%
  - Leigh, Lancashire, England 25%
- 1800
  - USA 37.5%
  - England 25%
  - Denmark 12.5%
  - Sweden 12.5%
  - Switzerland 12.5%



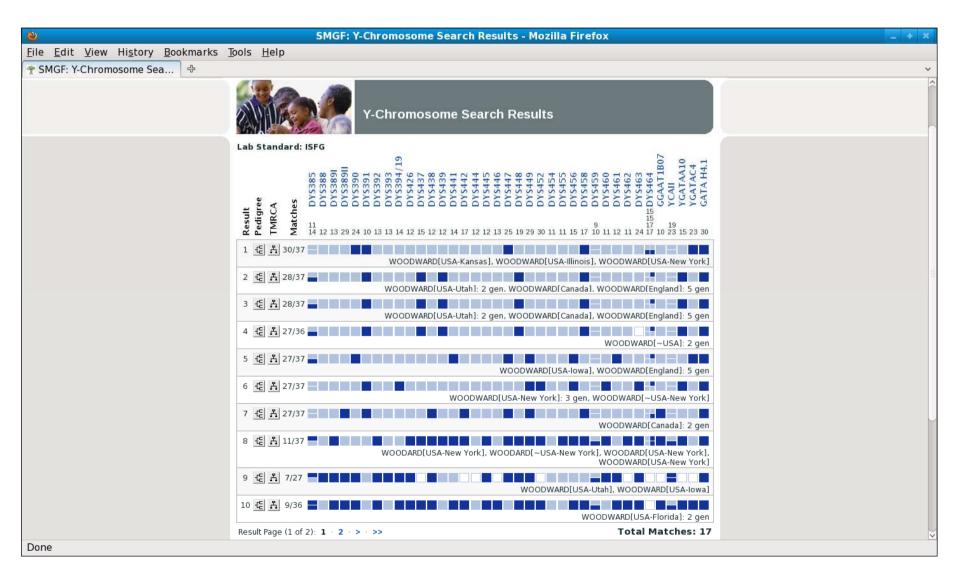
## www.SMGF.org



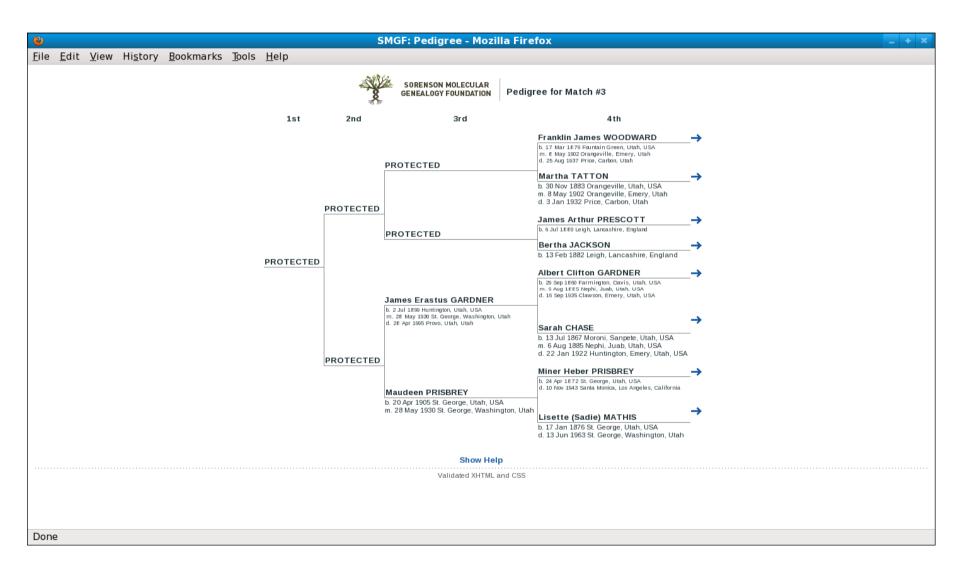
#### Y Surname search



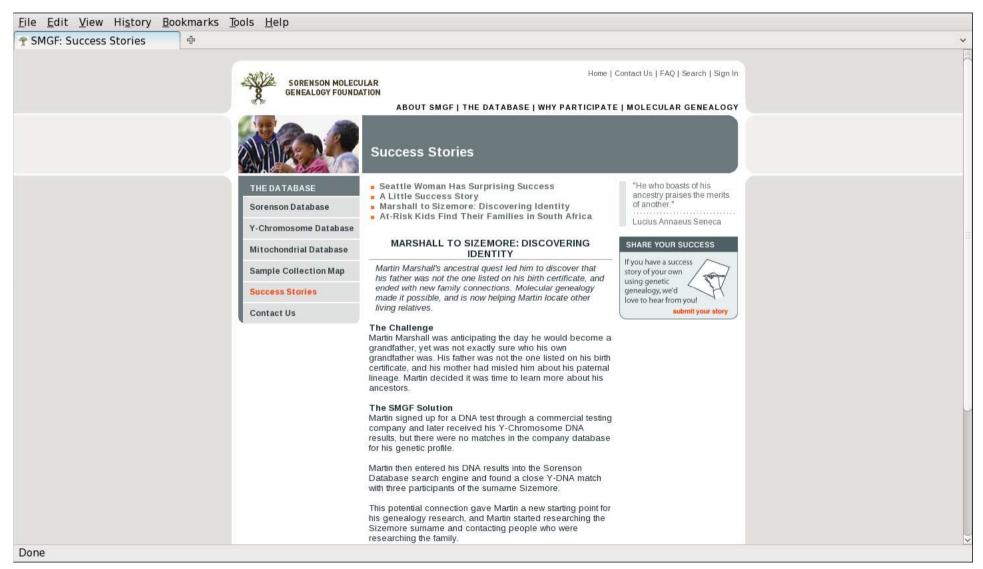
#### Y Search Result



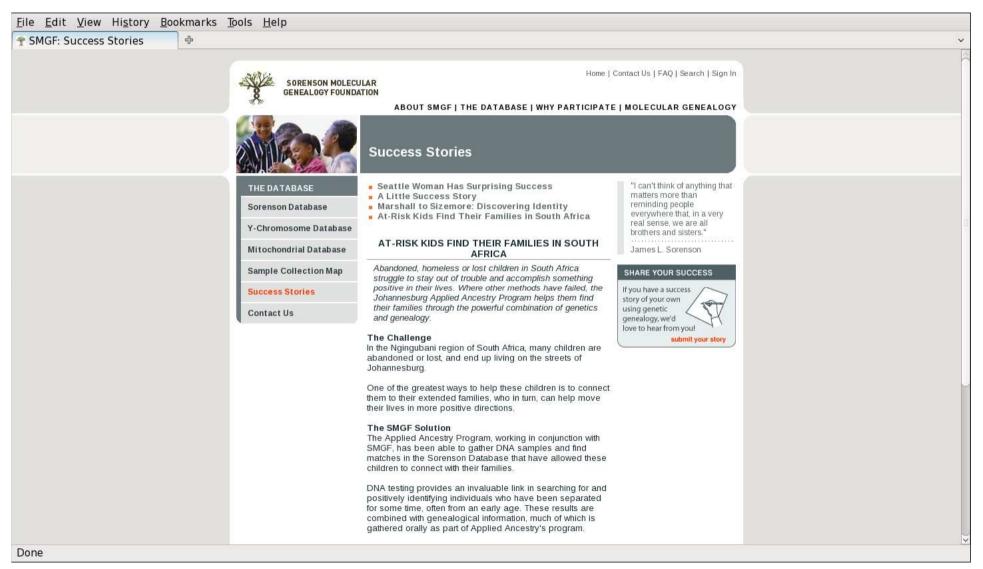
#### Pedigree Result



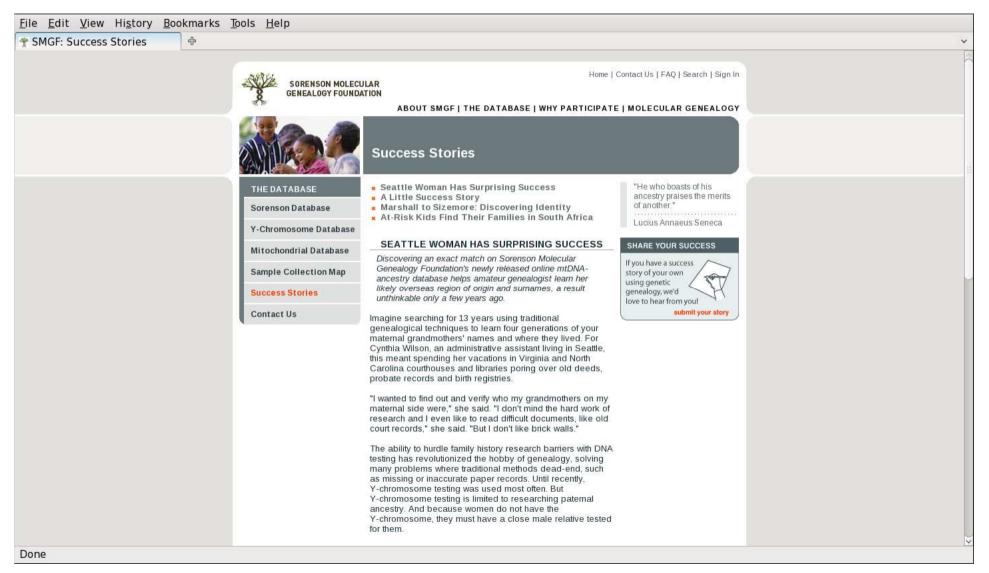
## Discovering Identity



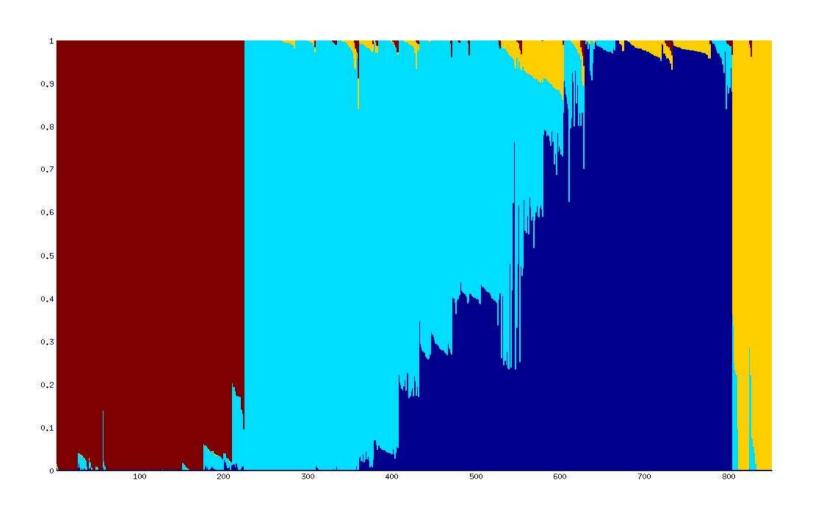
## Rediscovering Family



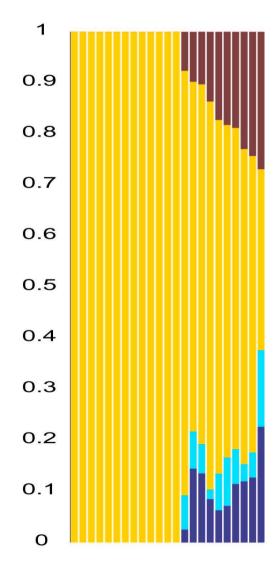
## Discovering Community



## Autosomal admixture

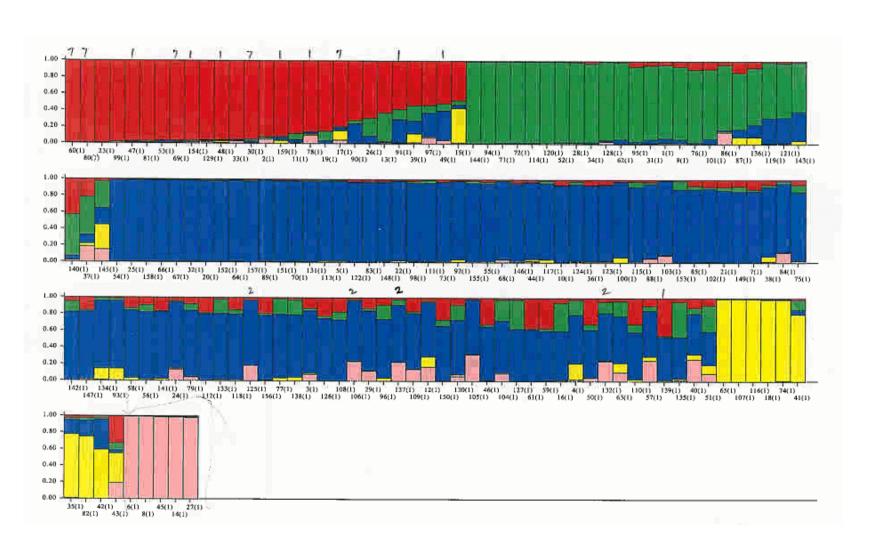


## A Native American Group

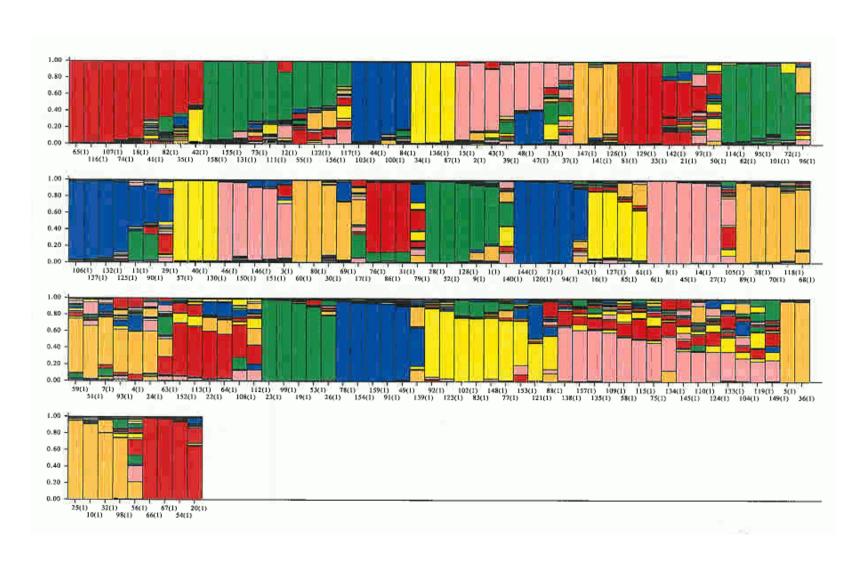


- Samples from Bolivia
- Affymetrix 6.0 data

#### Filomena k=5



#### Filomena k=26



## What does "Race" mean?



#### Collaborative Efforts



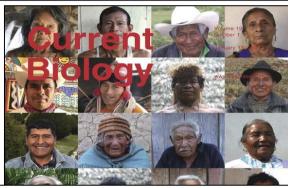
The Phylogeny of the Four Pan-American MtDNA Haplogroups: Implications for Evolutionary and Disease Studies

Alessandro Achilli<sup>1,2</sup>, Ugo A. Perego<sup>1,3</sup>, Claudio M. Bravi<sup>4</sup>, Michael D. Coble<sup>5</sup>, Qing-Peng Kong<sup>6</sup>, Scott R. Woodward<sup>3</sup>, Antonio Salas<sup>8</sup>, Antonio Torroni<sup>1\*</sup>, Hans-Jürgen Bandelt<sup>9</sup>

> Y-chromosome Short Tandem Repeat Intermediate Variant Alleles DYS392.2, DYS449.2, and DYS385.2 Delineate New Phylogenetic Substructure in Human Y-chromosome Haplogroup Tree CM

H. Ritchie<sup>1</sup>, Alice A. Lin<sup>2</sup>, Robert H. Hughes<sup>1</sup>, Scott R. Woodward<sup>1</sup>, Peter A. Underhill<sup>2</sup>





#### **Distinctive Paleo-Indian Migration Routes from Beringia Marked** by Two Rare mtDNA Haplogroups

Ugo A. Perego, 1,2,9 Alessandro Achilli, 1,3,9 Norman Angerhofer, 2 Matteo Accetturo, 1 Maria Pala, 1 Anna Olivieri, 1 Baharak Hooshiar Kashani, 1 Kathleen H. Ritchie,2 Rosaria Scozzari,4 Qing-Peng Kong,5,6 Natalie M. Myres,2 Antonio Salas,7 Ornella Semino,1 Hans-Jürgen Bandelt,8 Scott R. Woodward,2 and Antonio Torroni1.5

European Journal of Human Genetics (2008), 1 – 11
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Y-chromosomal evidence of the cultural diffusion of agriculture in southeast Europe

Vincenza Battaglia<sup>1</sup>, Simona Fornarino<sup>1,12</sup>, Nadia Al-Zahery<sup>1</sup>, Anna Olivieri<sup>1</sup>, Maria Pala<sup>1</sup>, Natalie M Myres<sup>2</sup>, Roy J King<sup>3</sup>, Siiri Rootsi<sup>4</sup>, Damir Marjanovic<sup>5,6</sup>, Dragan Primorac<sup>7,8</sup>, Rifat Hadziselimovic<sup>5</sup>, Stojko Vidovic<sup>9</sup>, Katia Drobnic<sup>10</sup>, Naser Durmishi<sup>11</sup>, Antonio Torroni<sup>1</sup>, A Silvana Santachiara-Benerecetti<sup>1</sup>, Peter A Underhill<sup>3</sup> and Ornella Semino\*.

Y-chromosome Short Tandem Repeat DYS458.2 Non-consensus Alleles Occur Independently in Both Binary Haplogroups J1-M267 and R1b3-M405



Natalie M. Myres<sup>1</sup>, Jayne E. Ekins<sup>1</sup>, Alice A. Lin<sup>2</sup>, L. Luca Cavalli-Sforza<sup>2</sup>, Scott R. Woodward<sup>1</sup>, Peter A. Underhill<sup>2</sup>

The American Journal of Human Genetics 84, 814–821, June 12, 2009

#### REPORT

Mitochondrial Haplogroup U5b3: A Distant Echo of the Epipaleolithic in Italy and the Legacy of the Early Sardinians

Maria Pala, Alessandro Achilli, Anna Olivieri, Baharak Hooshiar Kashani, Ugo A. Perego, 1,3 Daria Sanna,4 Ene Metspalu,5 Kristiina Tambets,5 Erika Tamm,5 Matteo Accetturo,1 Valeria Carossa,1 Hovirag Lancioni,<sup>2</sup> Fausto Panara,<sup>2</sup> Bettina Zimmermann,<sup>6</sup> Gabriela Huber,<sup>6</sup> Nadia Al-Zahery,<sup>1,7</sup> Francesca Brisighelli,8 Scott R. Woodward,3 Paolo Francalacci,4 Walther Parson,6 Antonio Salas,8 Doron M. Behar,<sup>9</sup> Richard Villems,<sup>5</sup> Ornella Semino,<sup>1</sup> Hans-Jürgen Bandelt,<sup>10</sup> and Antonio Torroni<sup>1,\*</sup>