



Editorial: Role of Y Chromosome in Molecular Anthropology, Forensics, and Genetic Genealogy

Sibte Hadi¹, Jun Yao^{2*} and Atif Adnan^{2*}

¹Department of Forensic Sciences, College of Criminal Justice, Naif Arab University for Security Sciences, Riyadh, Kingdom of Saudi Arabia, ²Department of Forensic Genetics, School of Forensic Medicine, China Medical University, Shenyang, China

Keywords: Y chromosomal STRs, archaeogenetic, rapidly mutating Y STRs, snps, forensic genetics

Editorial on the Research Topic

Role of Y Chromosome in Molecular Anthropology, Forensics, and Genetic Genealogy

The Human Y chromosome has proven to be a potent tool for studying human genetic anthropology due to its haploid state and the presence of a wide variety of markers (Quintana-Murci and Fellous, 2001). The accumulation of mutations has led to the separation of the Y lineages, which have been extensively studied (Quintana-Murci and Fellous, 2001; Knight et al., 2003; Tambets et al., 2004; Yang et al., 2014; Jobling and Tyler-Smith, 2017; Kivisild, 2017). Various markers have been utilised to study populations and lineages over different time scales. This makes the Y chromosome more powerful than mtDNA, which does not harbour the array of markers that the Y chromosome contains (Jobling and Tyler-Smith, 1995). Y chromosome microsatellites (Y-STRs) and single nucleotide polymorphisms (SNPs) have been extensively used in forensic applications and population studies. However, Alu insertion YAP and duplications/deletions have also been useful in population studies (Hammar et al., 1981; Hammer, 1994).

STRs have been extensively employed in forensic analysis and kinship testing and various commercial multiplexes are available. Specifically, Y STRs are used in the analysis of samples in sexual offense casework and various mixture analyses. Two mega multiplexes containing several Y-STRs are Powerplex Y 23 by Promega (Thompson et al., 2013) and Y Filer Plus by Thermo Fisher Scientific (Gopinath et al., 2016). Both kits are employed by forensic laboratories for casework and are used for kinship testing. Among the Y-STRs, a unique variety is Rapidly Mutating (RM) Y STRs, which have a niche application in separating paternally related individuals (Ballantyne et al., 2010; Ballantyne et al., 2012; Adnan et al., 2016; Neuhuber et al., 2022). Similarly, slowly mutating Y STRs (Baeta et al., 2018) might prove complementary to SNP markers in evolutionary studies, though the number of such markers needs to be increased for precise inferences.

Y SNPs like sY81 and SRY were studied initially, followed by the detection of a vast array of such markers (Underhill et al., 2000; Hinds et al., 2005; Repping et al., 2006; Karafet et al., 2008). The Y consortium published the Y chromosome phylogenetic tree topology based on 243 SNP markers (Consortium, 2002). A further 351 markers were used to develop a better-resolved tree (Karafet et al., 2008).

In the current era of rapid data generation traditional and more recently Massive Parallel Sequencing (MPS) platforms have been used, which together with the development of extremely powerful software have started to provide us with new markers and lineages. Hallast et al. (Hallast and Jobling, 2017) reported 13,621 SNPs after resequencing 3.7 Mb of the Male Specific Region of Y Chromosome (MSY). However, many thousands have been detected by others as well using Massive Parallel Sequencing techniques, increasing the potential of the Y chromosome as an evolutionary and

OPEN ACCESS

Edited and reviewed by:

Gyaneshwer Chaubey,
Banaras Hindu University, India

*Correspondence:

Atif Adnan
mirzaatifadnan@gmail.com
Jun Yao
yaojun198717@163.com

Specialty section:

This article was submitted to
Evolutionary and Population Genetics,
a section of the journal
Frontiers in Genetics

Received: 27 January 2022

Accepted: 09 May 2022

Published: 09 June 2022

Citation:

Hadi S, Yao J and Adnan A (2022)
Editorial: Role of Y Chromosome in
Molecular Anthropology, Forensics,
and Genetic Genealogy.
Front. Genet. 13:863455.
doi: 10.3389/fgene.2022.863455

human identification tool (Francalacci et al., 2013; Poznik et al., 2013; Wei et al., 2013; Scozzari et al., 2014). These developments are helping to determine new directions in human population migrations and evolution.

This Research Topic on the “Role of the Y Chromosome in Molecular Anthropology, Forensics, and Genetic Genealogy” aims to generate a review of the role of various Y markers, bringing to light current developments and applications within the field.

Male and female humans have contributed unequally to geographic expansion due to biological and behavioural differences. Dispersive genetic forces act quickly on uniparental sequences, which leads to changes in sequences, with one population branching off from the other. Mutations at the AZFc region or DYS448, which are regional specific or to some extent ethnic group-specific, are examples (Adnan et al., 2018; Adnan et al., 2021). These haplogroups were previously predicted using SNPs, which was a conventional method, but now these haplogroups can be successfully predicted from Y-STRs using different software packages like Nevgen and Whit Athey’s Haplogroup Predictor Tool (Athey, 2006). These software packages render quite precise information, which is supported by SNPs typing results. This might be due to STRs being more polymorphic as compared to SNPs (He et al., 2019). Studies on Y-STRs are still ongoing with several new applications. Ravasini et al. have identified four phylogenetically related samples with a null allele at DYS448 and a tetrallelic pattern at DYF387S1, two Y-STRs located in the AZFc. Through MPS analysis, they found that the unusual Y-STR pattern may be due to a 1.6 Mb deletion arising concurrently or after a 3.5 Mb duplication event. Jordamovic et al., predicted the Y haplogroup using Whit Athey’s Haplogroup Predictor, based on haplotype data on Powerplex Y 23 -STR markers. They found that Athey’s Haplogroup Predictor offers more accurate results and a higher probability of detecting rare haplogroups as compared to SNPs results. Luo et al. have characterized the genetic structure and forensic parameters of the She and Hakka ethnic groups from

Guangdong province of China. They ascertained that Guangdong Hakka has a close relationship with Southern Han, and the genetic pool of Guangdong Hakka was influenced by closely located Han populations. The predominant haplogroups of the Guangdong She group were O2-M122 and O2a2a1a2-M7, while Guangdong She clustered with other Tibeto-Burman language-speaking populations.

Bini et al. studied the relationships between old (16–18th century remains) and modern male populations of a population isolate from Roccapelago, which is a small village located in the Northern Central Apennines in Italy. In total, 14 modern samples and 25 ancient mummies were genotyped. They utilized several techniques to ascertain relationships between the old and modern-day populations from this area. Y haplogroup predictor tool (Athey, 2006) and Nevgen software (Cetkovic Gentula, 2015) were used to infer Y haplogroups from Y-STR haplotypes, which showed a close relationship between old and modern samples. Network analysis showed relationships between the two sets of samples.

The combined employment of various forensic genetics and archaeogenetic techniques demonstrates the value of a multidisciplinary approach, which is perhaps the way forwards for human Y chromosome analysis. Indeed, the Y chromosome continues to march on.

AUTHOR CONTRIBUTIONS

SH and AA drafted it, and SH, JY, and AA proofread it. Before submitting it, all authors reviewed it.

ACKNOWLEDGMENTS

To all the brilliant *Frontiers* team whose invaluable assistance at each step made the topic successful.

REFERENCES

- Adnan, A., Rakha, A., Kasim, K., Noor, A., Nazir, S., Hadi, S., et al. (2018). Genetic Characterization of Y-Chromosomal STRs in Hazara Ethnic Group of Pakistan and Confirmation of DYS448 Null Allele. *Int. J. Leg. Med.* 133, 789–793. doi:10.1007/s00414-018-1962-x
- Adnan, A., Rakha, A., Nazir, S., Alghafri, R., Hassan, Q., Wang, C.-C., et al. (2021). Forensic Features and Genetic Legacy of the Baloch Population of Pakistan and the Hazara Population across Durand Line Revealed by Y-Chromosomal STRs. *Int. J. Leg. Med.* 135, 1777–1784. doi:10.1007/s00414-021-02591-2
- Adnan, A., Ralf, A., Rakha, A., Kousouri, N., and Kayser, M. (2016). Improving Empirical Evidence on Differentiating Closely Related Men with RM Y-STRs: A Comprehensive Pedigree Study from Pakistan. *Forensic Sci. Int. Genet.* 25, 45–51. doi:10.1016/j.fsigen.2016.07.005
- Athey, T. (2006). Haplogroup Prediction from Y-STR Values Using a Bayesian Allele-Frequency Approach. *J. Genet. Geneal.* 2, 34–39.
- Baeta, M., Núñez, C., Villaescusa, P., Ortueta, U., Ibarbia, N., Herrera, R. J., et al. (2018). Assessment of a Subset of Slowly Mutating Y-STRs for Forensic and Evolutionary Studies. *Forensic Sci. Int. Genet.* 34, e7–e12. doi:10.1016/j.fsigen.2018.03.008
- Ballantyne, K. N., Goedbloed, M., Fang, R., Schaap, O., Lao, O., Wollstein, A., et al. (2010). Mutability of Y-Chromosomal Microsatellites: Rates, Characteristics, Molecular Bases, and Forensic Implications. *Am. J. Hum. Genet.* 87, 341–353. doi:10.1016/j.ajhg.2010.08.006
- Ballantyne, K. N., Keerl, V., Wollstein, A., Choi, Y., Zuniga, S. B., Ralf, A., et al. (2012). A New Future of Forensic Y-Chromosome Analysis: Rapidly Mutating Y-STRs for Differentiating Male Relatives and Paternal Lineages. *Forensic Sci. Int. Genet.* 6, 208–218. doi:10.1016/j.fsigen.2011.04.017
- Cetkovic Gentula, M. (2015). Y-DNA Haplogroup Predictor—NevGen. Available at: www.nevgen.org (Accessed January 27, 2022).
- Consortium, T. Y. C. (2002). A Nomenclature System for the Tree of Human Y-Chromosomal Binary Haplogroups. *Genome Res.* 12, 339–348. doi:10.1101/gr.217602
- Francalacci, P., Morelli, L., Angius, A., Berutti, R., Reinier, F., Atzeni, R., et al. (2013). Low-Pass DNA Sequencing of 1200 Sardinians Reconstructs European Y-Chromosome Phylogeny. *Science* 341, 565–569. doi:10.1126/science.1237947
- Gopinath, S., Zhong, C., Nguyen, V., Ge, J., Lagacé, R. E., Short, M. L., et al. (2016). Developmental Validation of the Yfiler® Plus PCR Amplification Kit: An Enhanced Y-STR Multiplex for Casework and Database Applications. *Forensic Sci. Int. Genet.* 24, 164–175. doi:10.1016/j.fsigen.2016.07.006
- Hallast, P., and Jobling, M. A. (2017). The Y Chromosomes of the Great Apes. *Hum. Genet.* 136, 511–528. doi:10.1007/s00439-017-1769-8

- Hammar, L., Månsson, S., Rohr, T., Chester, M. A., Ginsburg, V., Lundblad, A., et al. (1981). Lewis Phenotype of Erythrocytes and Leb-Active Glycolipid in Serum of Pregnant Women. *Vox Sang.* 40, 27–33. doi:10.1111/j.1423-0410.1981.tb00665.x
- Hammer, M. F. (1994). A Recent Insertion of an Alu Element on the Y Chromosome Is a Useful Marker for Human Population Studies. *Mol. Biol. Evol.* 11 (5), 749–761. doi:10.1093/oxfordjournals.molbev.a040155
- He, G., Adnan, A., Rakha, A., Yeh, H.-Y., Wang, M., Zou, X., et al. (2019). A Comprehensive Exploration of the Genetic Legacy and Forensic Features of Afghanistan and Pakistan Mongolian-descent Hazara. *Forensic Sci. Int. Genet.* 42, e1–e12. doi:10.1016/j.fsigen.2019.06.018
- Hinds, D. A., Stuve, L. L., Nilsen, G. B., Halperin, E., Eskin, E., Ballinger, D. G., et al. (2005). Whole-Genome Patterns of Common DNA Variation in Three Human Populations. *Science* 307, 1072–1079. doi:10.1126/science.1105436
- Jobling, M. A., and Tyler-Smith, C. (1995). Fathers and Sons: the Y Chromosome and Human Evolution. *Trends Genet.* 11, 449–456. doi:10.1016/S0168-9525(00)89144-1
- Jobling, M. A., and Tyler-Smith, C. (2017). Human Y-Chromosome Variation in the Genome-Sequencing Era. *Nat. Rev. Genet.* 18, 485–497. doi:10.1038/nrg.2017.36
- Karafet, T. M., Mendez, F. L., Meilerman, M. B., Underhill, P. A., Zegura, S. L., and Hammer, M. F. (2008). New Binary Polymorphisms Reshape and Increase Resolution of the Human Y Chromosomal Haplogroup Tree. *Genome Res.* 18, 830–838. doi:10.1101/gr.7172008
- Kivisild, T. (2017). The Study of Human Y Chromosome Variation through Ancient DNA. *Hum. Genet.* 136, 529–546. doi:10.1007/s00439-017-1773-z
- Knight, A., Underhill, P. A., Mortensen, H. M., Zhivotovsky, L. A., Lin, A. A., Henn, B. M., et al. (2003). African Y Chromosome and mtDNA Divergence Provides Insight into the History of Click Languages. *Curr. Biol.* 13, 464–473. doi:10.1016/S0960-9822(03)00130-1
- Neuhuber, F., Dunkelmann, B., Griefßner, I., Helm, K., Kayser, M., and Ralf, A. (2022). Improving the Differentiation of Closely Related Males by RMplex Analysis of 30 Y-STRs with High Mutation Rates. *Forensic Sci. Int. Genet.* 58, 102682. doi:10.1016/j.fsigen.2022.102682
- Poznik, G. D., Henn, B. M., Yee, M.-C., Sliwerska, E., Euskirchen, G. M., Lin, A. A., et al. (2013). Sequencing Y Chromosomes Resolves Discrepancy in Time to Common Ancestor of Males versus Females. *Science* 341, 562–565. doi:10.1126/science.1237619
- Quintana-Murci, L., and Fellous, M. (2001). The Human Y Chromosome: The Biological Role of a “Functional Wasteland”. *J. Biomed. Biotechnol.* 1, 18–24. doi:10.1155/S1110724301000080
- Repping, S., van Daalen, S. K. M., Brown, L. G., Korver, C. M., Lange, J., Marszalek, J. D., et al. (2006). High Mutation Rates Have Driven Extensive Structural Polymorphism Among Human Y Chromosomes. *Nat. Genet.* 38, 463–467. doi:10.1038/ng1754
- Scozzari, R., Massaia, A., Trombetta, B., Bellusci, G., Myres, N. M., Novelletto, A., et al. (2014). An Unbiased Resource of Novel SNP Markers Provides a New Chronology for the Human Y Chromosome and Reveals a Deep Phylogenetic Structure in Africa. *Genome Res.* 24, 535–544. doi:10.1101/gr.160788.113
- Tambets, K., Rootsi, S., Kivisild, T., Help, H., Serk, P., Loogväli, E.-L., et al. (2004). The Western and Eastern Roots of the Saami—The Story of Genetic “Outliers” Told by Mitochondrial DNA and Y Chromosomes. *Am. J. Hum. Genet.* 74, 661–682. doi:10.1086/383203
- Thompson, J. M., Ewing, M. M., Frank, W. E., Pogemiller, J. J., Nolde, C. A., Koehler, D. J., et al. (2013). Developmental Validation of the PowerPlex® Y23 System: A Single Multiplex Y-STR Analysis System for Casework and Database Samples. *Forensic Sci. Int. Genet.* 7, 240–250. doi:10.1016/j.fsigen.2012.10.013
- Underhill, P. A., Shen, P., Lin, A. A., Jin, L., Passarino, G., Yang, W. H., et al. (2000). Y Chromosome Sequence Variation and the History of Human Populations. *Nat. Genet.* 26, 358–361. doi:10.1038/81685
- Wei, W., Ayub, Q., Chen, Y., McCarthy, S., Hou, Y., Carbone, I., et al. (2013). A Calibrated Human Y-Chromosomal Phylogeny Based on Resequencing. *Genome Res.* 23, 388–395. doi:10.1101/gr.143198.112
- Yang, X., Wang, M., and Li, S. (2014). The Evolution of Human Y Chromosome. *Yi Chuan* 36, 849–856. doi:10.3724/SP.J.1005.2014.0849

Conflict of Interest: The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Publisher’s Note: All claims expressed in this article are solely those of the authors and do not necessarily represent those of their affiliated organizations, or those of the publisher, the editors and the reviewers. Any product that may be evaluated in this article, or claim that may be made by its manufacturer, is not guaranteed or endorsed by the publisher.

Copyright © 2022 Hadi, Yao and Adnan. This is an open-access article distributed under the terms of the Creative Commons Attribution License (CC BY). The use, distribution or reproduction in other forums is permitted, provided the original author(s) and the copyright owner(s) are credited and that the original publication in this journal is cited, in accordance with accepted academic practice. No use, distribution or reproduction is permitted which does not comply with these terms.