

International Society of Genetic Genealogy (ISOGG)

The **International Society of Genetic Genealogy (ISOGG)** is an independent non-commercial nonprofit organization of genetic genealogists run by volunteers. It was founded by a group of surname DNA project administrators in 2005 to promote DNA testing for genealogy. It advocates the use of genetics in genealogical research, provides educational resources for genealogists interested in DNA testing, and facilitates networking among genetic genealogists. As of June 2013, it comprises over 8,000 members in 70 countries. As of July 2013, regional meetings are coordinated by 20 volunteer regional coordinators located in the United States, Australia, Brazil, Canada, England, Egypt, Ireland and Russia.

ISOGG hosts the ISOGG Wiki, a free online encyclopedia maintained by ISOGG members which contains a wide variety of educational resources and guidance for genetic genealogy consumers and DNA project administrators. The ISOGG Wiki contains ethical guidelines for DNA project administrators and ISOGG members perform peer reviews of DNA project websites of other members on request, following which the websites may display the ISOGG Peer Reviewed graphic.

Industry regulation and standards

In 2006 ISOGG co-founder and director Katherine Borges explained there was interest in testing as "people want to connect", and in 2010 she estimated one million people had taken DTC genetic genealogy tests since they became available in 2000.

In 2008 ISOGG supported the passing of the Genetic Information Nondiscrimination Act designed to prohibit the improper use of genetic information in health insurance and employment in the United States. In July 2010 Borges represented ISOGG at an FDA public meeting on oversight of laboratory developed tests, where she spoke against FDA regulation preventing consumer access to DTC testing:

The general view of ISOGG's members is that regulatory agencies should not stand between a consumer who wishes to collect data on their own genome, and labs that can provide that service. The genome of an individual consists fundamentally of information, and every individual in a free society has an absolute right to information about their own genome from a source of their choosing. Our membership base consists of many MDs, PhDs, and other specialists who are willing to volunteer their time to assist with the development of industry standards, good practices, and advisory panels. These concepts could be developed in collaboration with federal agencies like NIST and the FTC. And FDA's regulatory requirements for DTCs could be met with something as simple as full and adequate disclosures of the limitations of the tests by the testing companies. The result could be a happy medium to the benefit of consumers, the laboratories, the testing companies, the government and to taxpayers.

An article published in *Genetics in Medicine* in March 2012 provides an overview of the diverse array of tests and practices in the emerging DTC genetic genealogy industry. In the article, the authors highlight ISOGG's potential role in developing industry best practice guidelines and consumer guidance:

Based on these data and our previous research, we believe that specific, federal regulation of the DNA ancestry testing industry is not warranted or justifiable at this time ... While no ethical or industry standards have been published, some companies in the DNA ancestry industry are accredited by the American Association of Blood Banks and the Clinical Laboratory Improvement Amendments ... We call on the International Society of Genetic Genealogy (ISOGG) to take a leadership role in

- (i) articulating an ethical code to guide the practices of the industry it advocates and
- (ii) developing a consumer guide to provide prospective consumers of the DNA ancestry testing industry with a reliable means to compare products and companies for their varying consumer motivations and interests. Moreover, we reiterate the need for a roundtable discussion (as recommended by the American Society of Human Genetics white paper) to better engage the many

parties with diverse needs for and interests in DNA ancestry inference and testing. Such a roundtable will be successful in developing best practice guidelines for DNA ancestry testing only if all parties approach the project in good faith.

The increasing affordability and popularity of DTC genetic genealogy testing has also raised ethical concerns about genealogists testing the DNA of others without consent. According to Borges, "People who realize the potential of DNA will go to great lengths to get it." The ISOGG Wiki contains a selection of external resources on ethics for genetic genealogists.

Y-STR nomenclature

ISOGG promotes the adoption of voluntary industry Y-STR nomenclature standards developed by NIST and published in the *Journal of Genetic Genealogy* in 2008. Borges explains ISOGG's rationale as follows:

As DNA testing for genetic genealogy purposes has become common and more people put their DNA profiles into online ancestry databases, the need for a universal format became apparent. This is a big benefit to consumers. They will definitely find more matches because of this new standard. Currently, consumers are often unaware they have to convert their results for use on different databases or are intimidated enough by the process that they don't check a variety of databases. Also, some errors find their way into conversion tools. All these problems reduce chances of finding matches.

Citizen science

ISOGG members such as Leo Little, Roberta Estes, Rebekah Canada and Bonnie Schrack have been involved in important citizen science discoveries regarding human phylogeny and ethnic origins. The broader ISOGG membership participates in and supports the Genographic Project, a genetic anthropology study that uses crowd sourcing to facilitate new discoveries about human genetic history, and other genetic databases where broader and larger databases aid the identification of participants' ancestral origins.

Y chromosome phylogenetic tree

Since 2006 ISOGG has hosted the regularly updated online ISOGG Y-chromosome phylogenetic tree. ISOGG aims to keep the tree as up-to-date as possible, incorporating new SNPs which are being discovered frequently. The ISOGG tree has been described by academics as using the accepted nomenclature for human Y-chromosome DNA haplogroups and subclades in that it follows the Y Chromosome Consortium nomenclature as described in Karafet et al. 2008, and being "one of the most up-to-date, if not completely academically verified, phylogenetic trees of Y chromosome haplogroups". The ISOGG tree is widely cited in peer reviewed academic literature.

The first phylogenetic chart to unify nomenclature was published in 2002 by the Y Chromosome Consortium (YCC). The 2003 Y-DNA phylogenetic chart appeared in Mark A. Jobling and Chris Tyler-Smith, *The Human Y Chromosome: An Evolutionary Marker Comes of Age* Nature Reviews|Genetics, Figure 5. In 2005 Family Tree DNA created the 2005 Y-Chromosome Phylogenetic Tree. An ISOGG group was formed in November 2005 to create a web-based document using Richard Kenyon's style of an indented list which could be updated to keep pace with the rapid developments in the field.

Current ISOGG members who work with the tree are: Coordinator: Alice Fairhurst. Design team: Tanmoy Bhattacharya, Katherine Hope Borges, Michael Hebert, Tom Hutchison, Richard Kenyon, Kriss Malachowski, Doug McDonald. Content experts: Abdulaziz Ali, Whit Athey, Ray H. Banks, Nigel Bond, Aaron R. Brown, David Dowell, Neal Downing, Charles Fueston, Phil Goff, Vladimir Gurianov, Gareth Henson, Greg Hockings, Robert Hughes, Tim Janzen, Mark Jost, James Kane, Kathleen Kerwin, Atanas Kumbarov, Zdenko Markovic, Eugene Matyushonok, Bob May, Lawrence Mayka, Christopher McCown, Charles Moore, Stephen Parrish, Marja Pirttivaara, Bonnie Schrack, David Stedman, Aaron Salles Torres, Steve Trangsrud, Leonard Trujillo, Ann Turner, Vladimir Volkov,

Michael W. Walsh. Content experts liaison with experts from various DNA labs to determine what information is needed to amend the tree.

The Y Haplogroup classification of the male Y-chromosome is currently used to estimate the population group of the paternal line. The haplogroups are identified by the letters, A through T. Haplogroups are subdivided into one or more levels, called subclades, and thus forming a tree. The Y-chromosome haplogroup is determined by performing a sequence of SNP tests.

Each line lists a haplogroup or subclade in boldface, then one or more SNPs follow on the same line. For a sample to belong to a particular subclade, it must test positive for any one of the SNPs appearing on the line, providing positive results were obtained for its haplogroup and any intervening subclades.

SNPs development indicated by beginning letters:

A = Thomas Krahn, MSc (Dipl.-Ing.), YSEQ.net, Houston, Texas

AD = Dr. Mohammed Al Sharija, Ministry of Education (Kuwait)

AF = Fernando Mendez, Ph.D., University of Arizona, Tucson, Arizona

BY = Big Y, Family Tree DNA, Houston, Texas

CTS = Chris Tyler-Smith, Ph.D., The Wellcome Trust Sanger Institute, Hinxton, England

DC = Dál Cais, an Irish group believed to be descended from Cas, b. CE 347, related to SNP R-L266; Dennis Wright

DF = anonymous researcher using publicly available full-genome-sequence data, including 1000 Genomes Project data; named in honor of the DNA-Forums.org genetic genealogy community

F = Li Jin, Ph.D., Fudan University, Shanghai, China

F* = Chuan-Chao Wang, Hui Li, Fudan University, Shanghai, China (Beginning letter F; second letter Haplogroup, i.e. FI is Fudan Haplogroup I)

FGC = Full Genomes Corp. of Virginia and Maryland

G = Verónica Gomes, IPATIMUP Instituto de Patologia e Imunologia Molecular da Universidade do Porto (Institute of Molecular Pathology and Immunology of the University of Porto)

IMS-JST = Institute of Medical Science-Japan Science and Technology Agency

K = Youngmin JeongAhn, grad student; Education: Seoul National University and the University of Arizona

KHS = Functional Genomics Research Center, Korea Research Institute of Bioscience and Biotechnology

KL = Key Laboratory of Contemporary Anthropology, School of Life Sciences and Institutes of Biomedical Sciences, Fudan University, Shanghai, China

KMS = Segdul Kodzhakov; Albert Katchiev; Anatole Klyosov; Astrid Krahn; Thomas Krahn; Bulat Muratov; Chris Morley; Ramil Suyunov; Vadim Sozinov; Pavel Shvarev; SF "National clans DNA project"; EHP "Suyun" Ph.D. of Technical Science; Prof. Elsa Khusnutdinova, Sc.D. of Biological Sciences, Laboratory of Molecular Human Genetics, Institute of Biochemistry and Genetics, Ufa Research Centre, Russian Academy of Sciences

L = Thomas Krahn, MSc (Dipl.-Ing.) of Family Tree DNA's Genomics Research Center; snps named in honor of the late Leo Little

M = Peter Underhill, Ph.D. of Stanford University

MC = Christopher McCown, University of Florida; Thomas Krahn, MSc (Dipl.-Ing.), YSEQ.net, Houston, Texas

N = The Laboratory of Bioinformatics, Institute of Biophysics, Chinese Academy of Sciences, Beijing

NWT = Northwest Territory, Theodore G. Schurr, Ph.D., Laboratory of Molecular Anthropology, University of Pennsylvania, Philadelphia, PA

P = Michael Hammer, Ph.D. of University of Arizona

Page, PAGES or PS = David C. Page, Whitehead Institute for Biomedical Research

PF = Paolo Francalacci, Ph.D., Università di Sassari, Sassari, Italy

PK = Biomedical and Genetic Engineering Laboratories, Islamabad, Pakistan

PR = Primate (gorilla and chimpanzee), Thomas Krahn's WTTY

S = James F. Wilson, D.Phil. at Edinburgh University

SA = South America, Theodore G. Schurr, Ph.D., Laboratory of Molecular Anthropology, University of Pennsylvania, Philadelphia, PA

SK = Mark Stoneking, Ph.D., Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

SUR = Southern Ural; SF "National clans DNA project"; B.A. Muratov; EHP "Suyun" Ph.D. of Technical Sciences; Ramil Suyunov; Prof. E.K. Khusnutdinova, Sc.D. of Biological Sciences, Laboratory of Molecular Human Genetics, Institute of Biochemistry and Genetics, Ufa Research Centre Russian Academy of Sciences; Alexander Zolotarev; Igor Rozhanskii; Bayazit Yunusbaev, Institute of Biochemistry and Genetics, Ufa Research Centre, Russian Academy of Sciences

TSC = Gudmundur A. Thorisson and Lincoln D. Stein, The SNP Consortium, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

U = Lynn M. Sims, University of Central Florida; Dennis Garvey, Ph.D. Gonzaga University; and Jack Ballantyne, Ph.D., University of Central Florida

V = Rosaria Scozzari and Fulvio Cruciani, Dipartimento di Biologia e Biotechnologie "Charles Darwin", Sapienza Università di Roma, Rome, Italy.

VL = Vladimir Volkov, Tomsk University, Russia

Y = Y Full Team using data from the 1000 Genomes Project

YP = SNPs identified by citizen scientists from genetic tests, then submitted to the Y Full team for verification.

YSC = Thomas Krahn, MSc (Dipl.-Ing.) of Family Tree DNA's Genomics Research Center

Z = Gregory Magoon, Ph.D., Richard Rocca, Vince Tilroe, David F. Reynolds, Bonnie Schrack, Peter M. Op den Velde Boots, Ray H. Banks, Roman Sychev, Victor Mas, Steve Fix, Christian Rottensteiner, Alexander R. Williamson, Ph.D. and an anonymous individual, independent researchers of publicly available whole genome sequence datasets, and Thomas Krahn, MSc (Dipl.-Ing.), with support from the genetic genealogy community.

ZP = Peter M. Op den Velde Boots, David Stedman using Big Y and other NGS sources.

ZS = Gregory Magoon, Ph.D., Aaron Salles Torres from samples from the 1000 Genome Project.