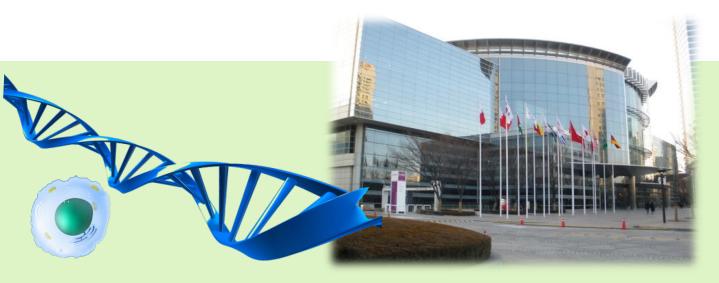




50 Years

International Society for Forensic Genetics 1968 - 2018







About this brochure:

We hope you will enjoy this brief foray into the past when Forensic Genetics was still Haemogenetics and even international meetings almost would have been in German. We know that our society and its working groups have fostered many friendships, lasting collaborations, and scientific progress. Our joint efforts have advanced the field and answered questions about identity, paternity, and ancestry for civil and criminal justice stake-holders. May the coming years continue to bring scientists together and improve all areas of forensic genetics.

Thank you to all brochure contributors, especially to our members sharing their personal memories.

On behalf of the current ISFG Board Mechthild Prinz NYC May 2019



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Cover photos – Top: University of Mainz, site of the 1968 meeting creating the ISFG Bottom: CoEx Conference Center, Seoul, Korea, site of the 26th International Congress in 2017.



Wolfgang Mayr

A brief History of the ISFG and its Name

The ISFG was founded in Mainz, Germany, in 1968 by a group of dedicated blood group serologists under the name "Gesellschaft für forensische Blutgruppenkunde e.V." (Society for Forensic Haemogenetics) to address the growing knowledge on inherited polymorphic markers in human blood and their use in kinship testing as well as in forensic stain analysis. The foundation assembly took place on June 24, 1968 at the Medical Faculty of the Johannes Gutenberg University Mainz. The founding members were: A. Arndt-Hanser, B. von Boros, B. Gumbel, K. Hummel, H. Leithoff, K. Luff, F. Petersohn, L. Wolff and W. Zimmermann. The Society was registered at the Amtsgericht (District Court) Mainz on July 31, 1968 under number 1006. The first Executive Board consisted of W. Zimmermann (President), L. Wolf (Vice President), K. Hummel (Treasurer) and A. Arndt-Hanser (Secretary).

Gesellschaft für Forensische Society for Forensic Blutgruppenkunde e.V. Haemogenetics

The first four congresses of the Society took place in Lübeck, Freiburg, Mainz and Trier in the years 1969 – 1972 (1 congress per year). Due to the growing interest of the international community in this field, the 5th Congress was organized in Amsterdam by J.J. van Loghem and P. Engelfriet; this internationalization implied an increased use of the English language in the life of the Society which until then was only German-speaking.



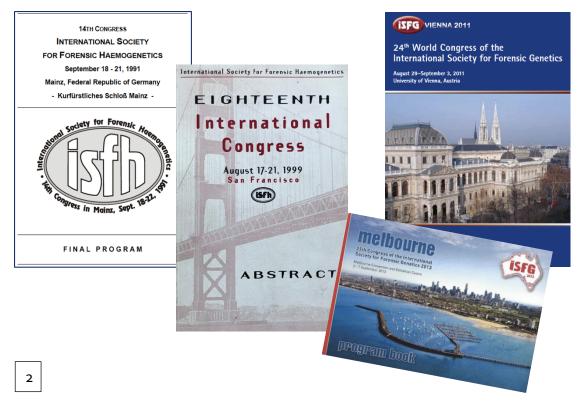
INTERNATIONAL SOCIETY FOR FORENSIC HAEMOGENETICS Internationale Gesellschaft für Forensische Hämogenetik e.V.

The name of the Society was changed to "Internationale Gesellschaft für Forensische Haemogenetik e.V." (International Society for Forensic Haemogenetics ISFH) during the General Assembly in Vienna on August 27, 1987,



INTERNATIONAL SOCIETY FOR FORENSIC GENETICS Internationale Gesellschaft für Forensische Genetik e.V. http://www.isfg.org

This name was soon obsolete again. The work of the Society's members covers forensic genetics in general (and not only forensic haemogenetics), therefore the name of the ISFH was changed one more time, now during the General Assembly in San Francisco on August 19, 1999 to its final version "Internationale Gesellschaft für Forensische Genetik e.V.", which is the legally registered name, but of course we now are the International Society for Forensic Genetics ISFG.







John Butler

As of 2018, ISFG has 11 active working groups. Eight of these groups are languagebased (German, English, French, Italian, Spanish & Portuguese, Chinese, Korean, and Polish). In a large international organization like the ISFG, language-based groups provide an effective means to exchange ideas and information and to discuss issues that may exist on a national or regional level. More information on each group can be found on the website: <u>https://www.isfg.org/Working+Groups</u>.

Several of the groups have developed quality control and proficiency testing exercises. The other three ISFG working groups – the Canine DNA Profiling Group (CaDNAP), European DNA Profiling Group (EDNAP), and the DNA Commission – serve to support research and harmonize developments in the field. Chairs of the working groups also assist the ISFG Board Member Representative of the Working Groups (currently John Butler, National Institute of Standards and Technology, USA) in evaluating short-term fellowship applications



The German-Speaking Working Group is currently chaired by Uta-Dorothee Immel (Institute of Legal Medicine, Mainz, Germany) and organizes annual conferences in cooperation with the German for Parentage Society Testing (DGAB) on the topic of ancestry and relationship studies along with forensic genetics.

The **English-Speaking Working Group** is chaired by Andreas Tillmar (National Board of Forensic Medicine, Linköping, Sweden) and conducts a relationship proficiency testing exercise each year, which is now organized by the Norwegian Institute of Public Health. Annual meetings have been held since 1979, either as part of the ISFG Congress, or as a separate conference in alternate years.



The **French-Speaking Working Group** is led by Diane Séguin (Laboratoire de Sciences Judiciaires et de Médecine Légale, Montréal, Canada) and meets two to three days each year to discuss results of collaborative exercises, to receive training on relevant topics, and to exchange information about casework experience.



The **Italian-Speaking Working Group** or Ge.F.I. for Genetisti Forensi Italiani (<u>http://www.gefi-isfg.org/</u>) is led by Loredana Buscemi (Legal Medicine, Ancona, Italy). Ge.F.I. organizes annual meetings and regularly conducts collaborative exercises.



The **Spanish and Portuguese-Speaking Working Group** or GHEP-ISFG for Grupo de Habla Espanola y Portuguesa de la ISFG (<u>https://ghep-isfg.org/</u>) is led by Ulises Toscanini (PRICAI-Favaloro Foundation, Buenos Aires, Argentina). This group is among the most active in ISFG with annual meetings, proficiency tests, and interlaboratory comparison exercises. GHEP-ISFG has published 33 articles describing results of their collaborative exercises and other working commissions.



The **Chinese-Speaking Working Group** was founded in 2005 and is chaired by Yiping Hou (Institute of Forensic Medicine, Sichuan, China) and works to promote scientific knowledge and educational opportunities in China regarding forensic genetics.



The **Korean-Speaking Working Group** is chaired by Soong Deok Lee (Department of Forensic Medicine, Seoul National University, Seoul, Korea) and facilitates communication among Korean speaking scientists working in forensic genetics. This group began in 2011 and helped organize the ISFG 2017 Congress held in Seoul.



The **Polish-Speaking Working Group** which was established in 2017, is led by Wojciech Branicki (Jagiellonian University, Krakow, Poland) and seeks to promote standards, share best practices, improve quality assurance, and conduct collaborative research at national and international levels.



The **Canine DNA Profiling Group** (CaDNAP) has existed since 2003 as a collaborative research project between the Institute of Legal Medicine at the Medical University of Innsbruck in Austria and the German Federal Criminal Police, but only became an official ISFG working group in 2017. The purpose of the group is to harmonize canine DNA analysis (for further information see https://gerichtsmedizin.at/cadnap.html) although members of the group are actively working in other non-human animal species. The group is open to new participants.



The European DNA Profiling Group (EDNAP) began in 1988 as a European effort to harmonize forensic DNA technology and became an ISFG working group in 1991. Semi-annual meetings (https://www.isfg.org/EDNAP/Meetings) are organized by Niels Morling (Department of Forensic Medicine, Copenhagen, Denmark). On-going research is discussed, and collaborative exercises planned. Over 30 publications on various topics have resulted from collaborative exercises conducted over the past three decades.

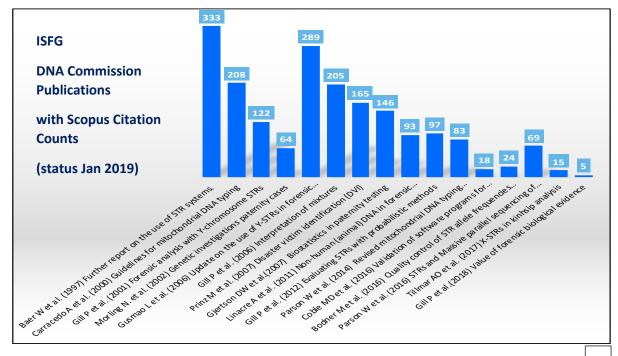


The **ISFG DNA Commission** was initiated in 1987 to discuss new developments and applications in forensic genetics and to make recommendations to assist methods and techniques and successfully move from research to operational implementation. The DNA Commission, which is comprised of subject matter experts along with the ISFG Executive Board, is led by Professor Peter Gill (Norwegian Institute of Public Health, Oslo, Norway) and over the past three decades has published 18 sets of recommendations on a variety of topics (https://www.isfg.org/Publications/DNA+Commission).

These ISFG DNA Commission recommenddations have provided important guidance on STR allele nomenclature, DNA mixture interpretation, disaster victim identifycation, use of non-human DNA in forensic investigations, validation of software programs performing biostatistical calculations, and interpretation of mitochondrial DNA, Y-STR, and X-STR results.



Commission Meeting 2005 in Porto, Portugal



*Three earlier publications did not have Scopus data.

ISFG Scientific Prize Winners



2017 Manfred Kayser for Y-chromosome and Phenotype Marker Research

2015 Thomas J. Parsons for DVI Research



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2013 Peter Gill for pioneering forensic genetics

2009 Antonio Salas for mt DNA and GHEP work

2007 Reinhard Szibor for X-Chromosome Research

2005 Walther Parson for mt DNA and EMPOP

2003 John Butler – STRs and Capillary Electr.

1999 Lutz Roewer – Y Chromosome & Y-HRD

1997 Antti Sajantila – work on PCR markers



1997 Colin Kimpton - the UK National Database

1989 Manfred Hochmeister for DNA Extraction

1987 Wolfram Dahr for Iso Electric Focusing



A short History of our Journal: Forensic Science International: Genetics

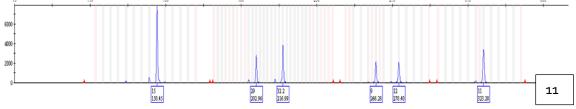
Angel Carracedo

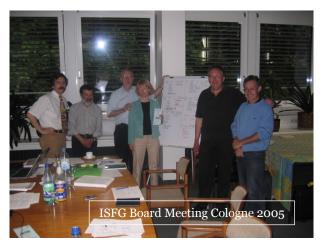
Prelude

For many years several members of the ISFG board were actively involved in editorial work related to forensic science. Since 1980 Dr. Patrick Lincoln had been the associate editor of Forensic Science International (FSI), a post taken over by Prof. Angel Carracedo in 1995. Prof. Bernd Brinkmann was editor-in-chief- of the International Journal of Legal Medicine (IJLM).

The plan to create a journal specifically devoted to Forensic Genetics started around the beginning of the century after some brainstorming within the ISFG board. By then, we had changed the society's name from ISFH to ISFG and it was clear that Forensic Genetics was a reasonably mature field, which was generating sufficient high quality content to support a dedicated journal. The field was continuously evolving, far from stabilizing, and major advances in molecular biology allowed for DNA profiles to be obtained from ever smaller quantities of biological material. Applications of forensic DNA technology were expanding outside the traditional areas, e.g. non-human DNA typing or forensic molecular pathology were fields with progressively more active research, something which also applied to mixture interpretation and statistics and interpretation. There were no signs the field would be slowing down.

As a consequence of this expansion, the topic of forensic genetics accounted for the largest proportion of submissions to general forensic journals, in 2002 for 46% of submissions to FSI and 56% to IJLM. This skewed submission pattern caused complaints from FSI readers working in other forensic disciplines.





The proposal

During the ISFG board meeting in Cologne, Germany in April 2005 we decided to make a proposal to Elsevier and to Prof. Pekka Saukko, (by then editor of FSI), to set up a new journal called Forensic Science International: Genetics, as a daughter journal of FSI. This would capitalise on the rapid growth and interest in this field.

The proposal also included a possible affiliation of the ISFG with FSI: Genetics and free journal access to the ISFG General Assembly (16th September, Azores). This followed a model, where IJLM was successfully affiliated to the International Academy of Legal Medicine.

In June, 2005, Prof. Saukko visited Santiago de Compostela, Spain, to discuss the idea of establishing, over the next 2-10 years, a strong family of FSI-branded journals to extend the relevance of FSI to smaller user groups. FSI: Genetics was meant to be the first step toward this goal, leading to a rebalancing of FSI content between the various forensic disciplines, thus maintaining its relevance to all specialty fields.

In September 2005, Dr. Lisa Colledge from the Netherland's headquarter of Elsevier submitted a proposal for Elsevier to launch a Forensic Science International daughter journal: FSI: Genetics, as the first of a serial of journals (including FSI: Toxicology), with Professor Saukko as overall Chairman, working closely with the daughter journal's Editor-in-Chief and Associate Editors to ensure that the journals moved in a



similar direction with no damaging competition. The proposal also nominated Prof. Carracedo as editor-in-chief for FSI:Genetics while continuing to serve as associate editor for forensic genetics in FSI. The idea was to retain forensic molecular pathology and anthropology in FSI while rerouting all other applications

of forensic DNA typing in criminal justice to FSI: Genetics.

Dr. Colledge's proposal, which was immediately accepted by the Elsevier board, envisioned that FSI: Genetics will develop a very close affiliation with the ISFG under the following conditions:

- 1. FSI: Genetics is designated as the official journal of the ISFG and this will be clearly indicated on the journal cover and any promotional material
- 2. FSI: Genetics cannot affiliate with any other society unless the ISFG agrees.
- 3. ISFG will be consulted on all invitations to and removals from the Editorial Board
- 4. The ISFG is allocated space in the journal to promote their own activities.
- 5. The publisher will meet at least once annually with the ISFG Board to report on the status of FSI: Genetics (in addition to other meeting to be held as necessary with the editor-in-chief and associate editors).
- 6. ISFG will make reasonable efforts to promote and to encourage submissions to the journal, while Elsevier makes reasonable efforts to promote the ISFG.
- 7. All ISFG members will receive a free Member Subscription to the journal.



The proposal was accepted by the ISFG General Assembly in Ponta Delgada, Azores' during the 21st Congress in 2005. Elsevier accepted simultaneously and an agreement was signed in 2006.



Launching the new journal

The launch of the new journal took place in March 2007. Prof. Carracedo (University of Santiago de Compostela, Spain) serving as Editor-in-Chief, and two associate editors: Prof. Peter Schneider (University of Cologne, Germany) and Dr. John Butler (Gaithersburg, Maryland, USA). In 2012, Prof. Adrian Linacre (University of Adelaide, South Australia) and Prof. Leonor Gusmão (University of Rio de Janeiro, Brazil) were added as additional associate editors to aid work with non-human DNA and population genetics articles, respectively. In 2016 Prof. Walther Parson (University of Innsbruck) was added to the team of associate editors due to the spectacular increase in submissions after the journal had surpassed other forensic journals regarding impact factor.



The journal

The number of submissions to the journal has been increasing around 10% annually with a total of 487 submissions in 2018 and a rejection rate of 60%. China is leading in number of submissions followed by United States and Australia, although all European countries together account for a second place in continents after Asia and Europe is still leading in number of accepted papers. Articles are available on-line within a few weeks of acceptance and are now typically printed within six months after passing the peer-review process.

Impact factors (IF), which are calculated each year by Thompson Reuters/ Clarivate Analytics from the number of citations to recently published

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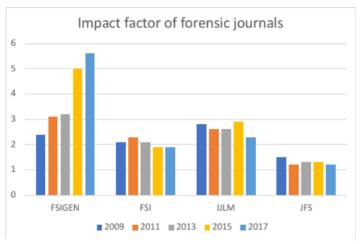
articles in a journal, are a measure of status amongst journals and a major factor for authors when deciding where to submit their work. The first couple of years prior to having an IF were a critical time for the journal. We needed to maintain high quality standards but, in many countries, an IF was needed for scientists to claim an article as part of their publication record. Here we would like to thank our many friends and colleagues in the ISFG who were contributing to the journal despite this fact.

Soon, in 2011, the journal was ranked first among all the forensic journals (Science Citation Index of Legal Medicine) and we never left this position. Furthermore, the difference with competing journals is exponentially increasing (see Figure 1). Our latest Impact Factor is 5.64, making FSI Genetics ranked #1 out of 16 journals in

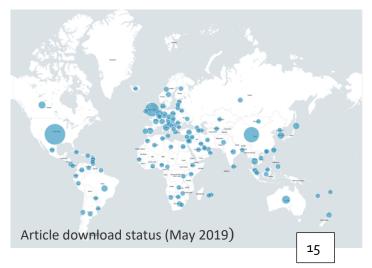
the "Medical, Legal" category.

The success of the journal is a combination of factors including the vitality of the field, the editorial policy (under the guidance and advise of an editorial board of the highest quality and reputation), the enthusiasm of the editors, the tireless work of its many peer reviewers, the efficiency of the publisher and its excellent team (now led by Alexander J. Smith, Elsevier, Oxford, UK) and the commitment of the ISFG board and members to maintain this journal in the highest standards of quality and excellence.

Last but not least, we would like to use this opportunity to thank everybody involved with the journal, but especially our peer reviewers!



Impact factor since inauguration



Blood wanted



Susanne Hirtz, from the Institute of Forensic Investigations and Kinship testing in Oldenburg, Germany, first joined the field of haemogenetics in 1971 by accepting a position at the Institute of Legal Medicine at the Georg August University in Göttingen, Germany. She started attending ISFG meeting shortly thereafter, with her first meeting 1972 in Trier, Germany. When she joined the society in 1978, she probably was the first female members of what was then the "Gesellschaft für Forensische Blutgruppenkunde". She remembers training in HLA typing with Wolfgang Mayr in Vienna, who liked her rare HLA type, was eager to obtain a reference sample, and with his typical Viennese morbid sense of humor suggested "to bleed her dry". This did not scare her away, she and her co-workers continued to attend ISFG meetings.



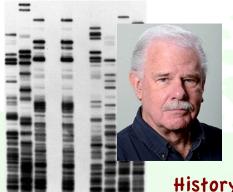


Years of service

Wolfgang Mayr, Department of Blood Group Serology and Transfusion Medicine at the Medical University of Vienna, Austria attended hist first meeting in 1977 in Hamburg and for the next 36 years did not miss a single meeting until scheduling conflicts prevented him from attending in 2015 and 2017. This must surely be the record. For 29 years he served as the secretary on the ISFG executive board and, needless to say, formed many important connections and friendships along the way. With his dedication to service it is no surprise that his favorite ISFG meeting is the 2011 meeting in Vienna, where he was the conference president and which was organized by his own research group. He had also served as the conference president for the 12th ISFG congress in Vienna 1987.

Over all these years his priority was the science. "The ISFG always tried to include the current trends in the scientific sessions. Genotyping by NGS, and definition of physical characteristics by DNA are the most exciting forensic genetics trends right now.

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George Sensabaugh from the School of Public Health, University of California in Berkeley, CA, USA has many ISFG memories. His first meeting was in 1979 in London, where he was happy to reconnect with friends from the Metropolitan Police Laboratory, who he met during his post doc in London 1971/72. George truly witnessed ISFG and science history and remembers it

History was made

I have many fun ISFG (and ISFH) memories, mostly little things like surprise encounters, great meals with old friends and new acquaintances, etc. But for me, the most memorable meeting was the 11th ISFH Congress in Copenhagen in early August 1985. This meeting stands out for three reasons:

- Up to that point in time, the official language of ISFH was German. Accommodations were made for non-German speakers but the German speakers delivered their papers in German. It was my understanding that Klaus Henningsen, the Congress President, proposed English as the official language of the Congress and that all papers were to be delivered in English. The ISFH had German origins and I think it fair to say that the Germans considered the Society to be theirs. The discussion of the proposal at the business meeting was quite heated; that the Germans were very upset was obvious, even though I, a non-German speaker, didn't really know what they were saying. English carried the day and I believe since that meeting virtually all presentations have been delivered in English. Arguably this was the point at which the ISFH transitioned from a German society to an international society.
- This meeting marked the first presentation of papers describing the forensic use of DNA polymorphism (*RFLP* analysis) in the examination of dried blood stains and semen. Note that this was 4 months before the Gill, Jeffreys, & Werrett Nature paper in December 1985. I had prior knowledge of the two *RFLP* papers presented at this meeting; they had been accepted by the Journal of Forensic Sciences and the editor of JFS had asked me to write an editorial lead for the March 1986 issue in which the papers were to appear. Although the Nature paper opened the flood gates on forensic DNA analysis, these two ISFH papers were the first presentations on the topic to the forensic genetics community. Both Peter Gill and Dave Werrett were at the Copenhagen meeting and were no doubt stimulated to pursue their research.
- The weather was wonderful that week in Copenhagen and the English and American attendees were treated to nude sunbathing in the parks, a new experience for them. Walter Bär observed that nude bodies were like enzymes: much polymorphism. (This comment will be obscure to readers entering the field after the polymorphic enzyme era.)





Angel Carracedo, from the Institute of Forensic Science, Medical Faculty, University of Santiago de Compostela, Spain started to attend ISFG meetings in 1983, with the meeting in Munich, Germany. Back then he was excited to meet Sebastian Weidinger from the University of Munich and Vince Pascali from the University of Rome, fellow researchers of alpha-1-antitrypsin polymorphisms. He became active in DNA research and applications, served as the ISFG president from 1994 to 1998, and is the editor of Forensic Science International Genetics.

ISFG Meetings can be romantic

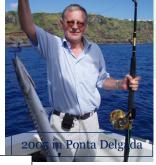
"I especially have good memories of the 1985 meeting in Copenhagen (my first trip with Montse, then my girlfriend, now my wife) for obvious reasons although the trip was a disaster from the very beginning (we have planned to go by car with a group of friends and we had an accident in León- central Spain) so we decided to continue by train with Interrail with stops in Paris, Amsterdam ... with the guitars. I don't remember the meeting at all (well, it was my second talk at a ISFG meeting) but of course I remember every single moment with Montse."

Seaside Congress

Christian Doutremepuich, Laboratoire Hemato-logie Medico-Legale, Bordeaux, France joined ISFG activities at his first meeting in Vienna 1987. Later in 2003 he served as the organizer and congress president for the 2003 meeting in Arcachon, France. This congress remains an ISFG member favorite and is also his favorite meeting: *"We had an interesting meeting in a beautiful town."*



All the Congresses were great



Niels Morling, Department of Forensic Genetics, University of Copenhagen, became active in the society in 1989 and for many years served as the ISFG treasurer, president and vice president. He remembers: "*My first ISFG congress in 1989 in New Orleans was very important for me, because I changed to forensic genetics in August 1989. Thus, I learned a lot and met our colleagues from all over the world. I am sure that many colleagues have had the same experience.*"

Mathematics and Bears



Charles Brenner, Forensic Mathematician from Berkeley, USA , became an active ISFG contributor with his first meeting in Mainz in 1991. He then attended 14 consecutive meetings. He very much enjoyed meeting Alec Jeffreys and was able to spend most of 1999 in the Jeffreys' laboratory in Leicester, UK. His least favorite encounter was with several bears after the social event of the 2008 ESWG meeting in Sinaia, Romania; he recalls a narrow escape.

Asked about the most exciting trends in forensic genetics right now, he replies: *I can only speak to mathematics as it is the only area in which I'm more than a bystander. The theory of dealing with mixtures is now on a firm footing. There are useful but not definitive advances toward a satisfactory population genetic solution to dealing with Y haplotype and mtDNA forensic evidence. However, I believe that in some respects we are now bumping into inherent limitations in the mathematics of evaluating evidence.*

It's the people that make the difference

Susi Pelotti, Department of Medical and Surgical Sciences, University of Bologna, Italy also attended her first meeting in Mainz in 1991. She was happy to encounter several of the authors of groundbreaking DNA identification papers, like Alec Jeffreys, Peter Gill, Angel Carracedo, and Bernd Brinkmann. But the first person she met in Mainz was Peter Schneider, who as the part of the organizing committee, was welcoming the participants. *"Peter Schneider became a reference point for me and my group. Not only for scientific issues, but also through the longtime friendship with him. He was guest of the Ge.F.I. meeting in Bologna in 2004, talking about the need of lab accreditation, a urgent problem also nowadays in Italy. He was always present for the Italian working group and welcomed students in his lab for research projects."*

Susi's favorite fun ISFG memory is from an English Speaking Working Group (ESWG) meeting in 2006 in Helsinki Finland. They had organized an outing that turned out to be a *"very funny boat trip ...it was not so simple for participants to be rowing in the right direction!"*



MEMORIES

Science and Opera



Yiping Hou, Institute of Forensic Medicine, Sichuan University, Chengdu, PR China was a Humboldt Fellow in Germany at the time and thus able to attend the meeting in Lido di Venezia, Italy in 1993. He shares his favorite meeting memories with John Butler. His favorite meeting was the one in 2005, Ponta Delgada, Azores. He enjoyed the island scenery, but more importantly this was the congress, where the general assembly approved the creation of the Chinese Speaking Working Group. The decision to launch FSI:Genetics was also made here. His second favorite meeting location is Vienna, the center of classical music. As an avid opera fan, Yiping fondly remembers a visit to the famous Vienna opera house during the 2011 congress.

Spanish Portuguese Speaking WG and Quality Control

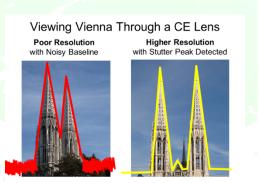
Daniel Corach, of the Servicio de Huellas Digitales Genéticas (SHDG) at the University of Buenos Aires, Argentina, also counts the 1993 congress in Lido di Venezia, Italy, as his first meeting. He remembers: *"I was introduced to the ISFH by Dr. Mariam Martinez de Pancorbo, during the International Conference of DNA Fingerprinting held in Belo Horizonte, Brazil, a year before. Marian introduced me to Angel Carracedo,*



who became the president of the Society that year and organized the 1995 Congress in Santiago de Compostela, Spain. During the Venice meeting I got in contact with some of the founders of the Spanish and Portuguese speaking group, where I joined and started to participate in the Collaborative Exercises of Quality Control (Proficiency Testing). "Daniel's laboratory is still participating in the yearly proficiency testing scheme and he credits the Grupo Hablo Espanol y Portuguese (GHEP) for inspiring the formation of the Argentinian Society of Forensic Genetics in 2000. He lauds the ISFG meetings for the continuing education and training opportunities, including coverage of updated analytical processes and technological innovations, that are important for scientific progress and compliance to IS017025 quality assurance standards in working laboratories.

Meeting colleagues worldwide

John Butler from the National Institute of Standards in Gaithersburg, MD, USA attended his first ISFG meeting in 1999 in San Francisco. He fondly remembers meeting there e.g. Gillian Tully (UK, FSS) and Chantal Frégeau (Canada, RCMP) whose research articles he had read and work he admired. He always enjoys the opportunity to meet researchers from around the world and likes the





ISFG meetings for the extensive program with hundreds of posters and excellent oral presentations. John, which one was your favorite ISFG meeting? "ISFG 2005 in Ponta Delgada: The Azores were an exotic location and we enjoyed an island tour following the meeting. I gave the scientific prize lecture, so it will always be meaningful to me for this reason."

Here is another meeting memory: "At the 2011 ISFG meeting in Vienna, I used a photo of two church steeples to illustrate resolution of STR peaks for a CE workshop. I had snapped the photos from different angles the previous day and decided at the last minute to bring a local flavor to the workshop presentation."



Truly International

Sally Ann Harbison of the Environmental Science Research Institute in Auckland, New Zealand counts 2003 in Arcachon as her first meeting, where Peter Gill introduced her to many other colleagues. Sally Ann considers Titia Sijen and colleagues at the Netherlands Forensic Institute important ISFG connections. She states: "ISFG is where you hear about new developments that may or may not make it into the operational laboratories. As I do both research and casework, I appreciate finding out about the new developments so I can incorporate them, or not, back in the lab. I like how ISFG has become more diverse in attendees, speakers and locations because it is now truly an international society." And 2013 was great: "Having ISFG in Melbourne was a highlight for me. Being able to showcase our part of the world, the local wildlife and amazing dancing prowess after the dinner was a highlight. As well as fun, it really opened up and promoted forensic research to the local forensic community which has grown amazingly ever since."





Young scientists get a chance

Hwan Young Lee, now at the Department of Forensic Medicine, Seoul University, Republic of Korea, fondly remembers Arcachon in 2003 as her first meeting. She and her colleagues practiced how to pronounce "Arcachon" with an Air France attendant on their flight from Paris. Thomas Parsons was the first non-Korean scientist to talk to her at the meeting. He and others became esteemed colleagues and inspired her to participate in every ISFG meeting since. She continued to meet future collaborators at ISFG meetings, like Jana Naue in Krakow in 2005. She likes that ISFG meetings promote new research: "*The ISFG was always quick to introduce new technologies, such as applying mass spectrometry and NGS to genotyping, employing mRNA or methylation for body fluid ID, etc. and gives presentation opportunities to anyone who has a brilliant idea. In my personal opinion, providing equal opportunities for oral presentations to prestigious scholars and promising young scientists is really good."*

Beatriz Martinez, from the University of Cartagena, Colombia attended her first ISFG meeting in 2005, in Ponta Delgada on the Azores. For her the meeting "*was the beginning of good collabo-rative relationships between us and groups from Spain and Portugal, especially with Antonio Amorim, Leonor Gusmao, Angel Carracedo, and Mariam de Pancorbo.* "Scientifically Beatriz is excited about the potential of DNA databases and DNA phenotyping. Picking a favorite meeting is difficult, each meeting has its own "*special charm*", but Vienna in 2011 is worth mentioning for its cultural events – a film and a gastronomic festival. ISFG meetings are social events and create fun memories with friends and .colleagues.

Collaborations are key







The history of the haploid marker databases YHRD and EMPOP – a brief sketch

Walther Parson

For many forensic geneticists and colleagues from related scientific fields, the biannual "Haploid Marker Meeting" has become a tradition. Since the 2004 Berlin meeting, Y-chromosomal and mitochondrial (mt)DNA related topics have been discussed together in the same forum (Table 1); despite, or maybe because, they are tackling opposite inheritance schemes and address different forensic casework scenarios. These workshops have been very valuable to the community and typically bring more than 200 scientists from around 50 countries together to discuss recent scientific developments.

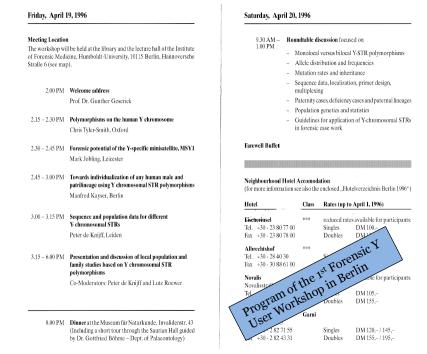
Forensic Y-chromosomal and mtDNA analyses are both formed around centrally curated databases, the Y-Chromosomal Haplotype Reference Database (YHRD, <u>https://yhrd.org/</u>) and the EDNAP mtDNA Population Database (EMPOP,

https://empop.online). Both are scientifically endorsed by the ISFG as freely available web-based resources; they serve as a source of carefully collected and curated genetic variation; and they both assist quality control procedures needed to maintain the high data quality that is required in forensic genetics. Yet, the two databases have different histories.



It was sunny and mild on Friday April 19th, 1996, when I navigated myself by nondigital means (a paper map) through Berlin to find Hannoversche Strasse 6, the address of the former Institute of Legal Medicine.

Here, the first Y-Chromosome User Workshop was supposed to take place. I did not even reach the gate of the building, when a friendly looking young man - a typical scientist by appearance, waved from the open entrance door and welcomed me warmly. This was my first personal contact with Lutz Roewer and little did I know that I had just met for the first time, one of my dearest colleagues, on my 30th birthday.



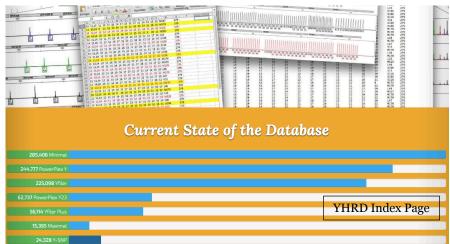
The workshop started with introductory population genetic lectures on various Ychromosomal marker classes and led into the presentation and later discussion of a defined set of nine Y-chromosomal STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a and DYS385b). These nine loci became known as the 'Minimal Y-Chromosomal STR Haplotype' and when the YHRD launched, formed its fundamental core set [1].

[1] Roewer, L., et al., Online reference database of European Y-chromosomal short tandem repeat (STR) haplotypes. Forensic Sci Int, 2001. **118**(2-3): p. 106-13.

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The history of YHRD

The first database release in 1999 was populated with 3,825 minimal haplotypes from 41 European populations to serve the forensic community with



a resource for probability estimates. Additional Y-STR loci were identified and added to the analytical panels to increase the information content for forensic purposes.

Today, on its 61st release, the YHRD presents more than 285,000 searchable minimal haplotypes in more than 1,300 populations from 135 countries, and various sets of extended haplotypes (up to 29 loci), and provides the most comprehensive and relevant resource of Y-chromosomal variation for forensic purposes.

In addition to the fundamental task of data collection that soon extended all over the globe to include worldwide populations, the Berlin team, particularly Lutz Roewer and Sascha Willuweit, carefully monitored the demands of forensic practitioners. In response to these needs they developed a variety of tools that are now offered within YHRD, including Mixture Analysis and Kinship Analyses that make the daily work with Y-STR haplotypes an easier task [2]. YHRD has also included the development and provision of the Discrete Laplace (DL) method; acting as a frequency estimator that takes the phylogeny of the haplotypes into consideration and thus provides a more realistic estimate of the probability of a given haplotype. The DL method has found broad acceptance in the forensic community and represents a state-of-the-art method to convey Y-STR evidence. It is based on YHRD haplotypes and presents a good example of the importance of well curated, centrally held data.

[2] Willuweit, S., L. Roewer, and Y.C.U.G. International Forensic, *Y chromosome haplotype* reference database (*YHRD*): update. Forensic Sci Int Genet, 2007. **1**(2): p. 83-7.

The history of EMPOP

In the late 1990s, individual laboratories and research groups hosted their own local mtDNA databases. These were not directly accessible by outside users, nor were the various laboratories who were pioneering forensic mtDNA tests at the time, actively communicating with each other. Therefore, it came as no surprise that the nomenclature systems used for each database were quite different. This initiated discussions at the 18th ISFH Congress in San Francisco 1999 concluding that the forensic community required a centrally curated, internet accesible mtDNA population database.

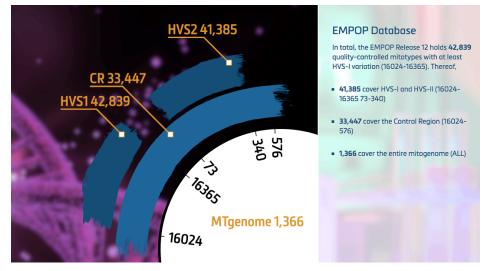
	5.	Suggestic	n for an EDNAI	P mtDNA database	e	Walther Parson			
	A dı	raft descriptio	n of the project i	s attached at Annex	7				
	Geri maii	many. The p ntained by El	VA databases are already in existence; one from the FBI and the other from The proposal is that there should be an open database on the Internet that is by EDNAP. The acronym EMPOP was suggested for 'The EDNAP mitochondria lation Database Project'.						
	to en is ir	nsure that it i	s a sensible resul	which has been set up in Innsbruck, uses software that can check input data a sensible result. Statistical data on major ethnic types and sub-affiliations oftware also records who has accessed the database and what has been					
inc	0								
STADA	AP *	STADNAP Min	ites - 6 May 2000 -	Dublin	Doc: MinST	ADNAP0052.doc		5	

The concept of EMPOP was proposed at the EDNAP meeting in Dublin in 2000 (see above) and solidified in the following years. Soon after starting the collection of mitotype data it became apparent that the literature across a range of scientific disciplines was not a reliable source of mtDNA variation data, as it was fraught with error. The EDNAP group performed a collaborative blind test that revealed a 10% error rate in the reported results, the majority of which could be attributed to clerical errors. These results triggered the development and dissemination of: i) elaborate laboratory methods to support the generation of high-quality mitotypes; and ii) software to control the quality of collected mtDNA sequences using phylogenetic methods. In 2006, the first version of EMPOP was launched based on its first data release of 5,173 quality-controlled sequences [3].

^[3] Parson, W. and A. Dur, *EMPOP--a forensic mtDNA database*. Forensic Sci Int Genet, 2007. **1**(2): p. 88-92.

In 2010, *Forensic Science International Genetics* and *International Journal of Legal Medicine* started to require authors to perform a mandatory quality check of mtDNA population data before submission of the manuscript for review (a system also established for Y-STR data submitted to YHRD). In practice, the vast majority of datasets had issues that were corrected in conversation with the authors, which significantly improved the quality of published datasets in these forensic journals.

MtDNA sequences are usually reported relative to the human reference sequence (rCRS), which - depending on the alignment method applied - can result in different nomenclature.



This is an inherent characteristic of all reference-coded haplotypes and genotypes and can be problematic in the forensic context, as it may lead in criminal casework to false exclusions from undetected matches in DVI database searches and to a general overestimation of the evidence due to biased match probabilities. To harmonize mtDNA nomenclature, the phylogenetic alignment system was proposed in 2008 and generally accepted by the forensic community in 2014. In 2019, EMPOP 4 was released including a new query engine (SAM 2) that: i) performs unaligned mitotype searches in order to provide unbiased probability estimates; and ii) outputs the phylogenetic alignment of EMPOP query mitotypes, thus providing the basis for a unique mtDNA nomenclature in both forensic genetics and other fields of genetics [4].

^[4] Huber, N., W. Parson, and A. Dur, *Next generation database search algorithm for forensic mitogenome analyses*. Forensic Sci Int Genet, 2018. **37**: p. 204-214.

Most of the developments described above for YHRD and EMPOP were triggered by input from colleagues during the Haploid Marker Workshops and advanced in these forums The idea for YHRD started with the first Y-User Meeting. Chromosome Between 2006 and 2014, the thematical scope of the workshops extended to include Xwas Chromosomal and pharmacogenetic/medical genetic



topics, which was reflected by the broader title "DNA in Forensics".



The history of this forum and the research focus of the audience have however led to the reduction of the scope to mainly cover Y- and mtDNA-related topics that form the core of this group. The workshops were often inspired by mottos that inform the participants about the focus of the meeting and invite related

contributions, e.g. "Exploring the Phylogenies" (2012), "Is NGS now GS in Forensics" (2014), "Inferring Ancestry from DNA" (2018). Right now, preparations for the next Haploid Markers meeting in 2020 have already started to continue this success story.



Overview of Y-Chromosome User Workshops and EMPOP Meetings (combined in 2004).

Thank you to everybody supporting the meeting.

Year	Meeting,	Meeting Organizer		
1996	Y-ChromosomeUser Workshop, Berlin	Dept. Forensic Genetics, Institute of Legal Medicine and Forensic Sciences, Charité, Berlin (Germany)		
2000	Y-Chromosome User Workshop, Berlin	Dept. Forensic Genetics, Institute of Legal Medicine and Forensic Sciences, Charité, Berlin (Germany)		
2002	Y-Chromosome User Workshop, Porto	IPATIMUP, Porto (Portugal)		
2004	Y-Chromosome User Workshop and EMPOP Meeting, Berlin	Dept. Forensic Genetics, Institute of Legal Medicine and Forensic Sciences, Charité, Berlin (Germany)		
2006	DNA in Forensics, Innsbruck	Institute of Legal Medicine, Medical University of Innsbruck (Austria)		
2008	DNA in Forensics, Ancona	Institute of Legal Medicine, Medical University of Ancona (Italy)		
2010	DNA in Forensics, Berlin	Dept. Forensic Genetics, Institute of Legal Medicine and Forensic Sciences, Charité, Berlin (Germany)		
2012	DNA in Forensics, Innsbruck	Institute of Legal Medicine, Medical University of Innsbruck, (Austria)		
2014	DNA in Forensics, Brussels	Belgian Institute for Forensic Science and Criminology, Brussels (Belgium)		
2016	Haploid Markers, Berlin	Dept. Forensic Genetics, Institute of Legal Medicine and Forensic Sciences, Charité, Berlin (Germany)		
2018	Haploid Markers, Bydgoszcz	Department of Forensic Medicine, Nicolaus Copernicus University, Bydgoszcz (Poland)		



Save the date:





Don't forget to explore the ISFG website:

Available to all - free copies of all DNA Commission publications and articles in conference proceedings (1985 – present). Also look for Working Group updates and forensic software resources.

The "members only" section offers a link to your free copy of Forensic Science International Genetics, the capability to search for other ISFG members, and under the education tab, access to workshop and plenary lectures from previous conferences.

50th Anniversary Brochure distributed at the



The 28th Congress of the International Society for Forensic Genetics

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