ISFG ESWG 2016

INTERNATIONAL SOCIETY FOR FORENSIC GENETICS

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ENGLISH SPEAKING WORKING GROUP

BUDAPEST, HUNGARY
31.08.-03.09.2016
NOVOTEL BUDAPEST CENTRUM* * * *
www.eswg2016.com

DEAR COLLEAGUES!

The International Society for Forensic Genetics (ISFG) was wet up to promote scientific knowledge in the field of genetic markers analysed for forensic purposes. The ISFG was founded in 1968 and represents more than 1100 members from over 60 countries. Regular meetings are held at a regional and international level. Scientific recommendations on relevant forensic genetic issues are developed and published by expert commissions of the ISFG. The working groups are an important forum for the exchange of information, and are quite helpful for dealing with special problems at regional levels. furthermore, they have developed into platforms for quality control and proficiency testing exercises.

The English Speaking Working Group (ESWG) has the genetic and statistical analysis of familial relationships as its focus and is therefore the key meeting for all practitioners in this area. In addition to the scientific meeting there will be a workshop to discuss results of the relationship testing exercise and an opportunity to present and discuss interesting cases from the community.

The ESWG is delighted to celebrate the 2016 meeting in Budapest - a city famed for its cultural and culinary heritages.

We will provide an exciting meeting with cutting edge forensic genetics presentations hosted by acknowledged leaders. The meeting will also include an exhibition program with leading suppliers in the forensic genetics field. We are confident that the accompanying social programme will provide you the opportunity to network, meet old friends, and create new ones.

We would like to thank all those who have sponsored, supported and helped along the way to organise what we are sure will be a most memorable meeting.

We welcome all to Budapest, a truly great city, and look forward to a few days of learning, networking and celebrating.

PATRON

Dr. Judit Szekér
Network of Forensic Science Institutes, Budapest, Hungary

PRESIDENT OF THE MEETING

Dr. Pamjav Horolma

SCIENTIFIC COMMITTEE

Dr. Gunilla Holmlund
Dr. Denise Syndercombe
Dr. Pamjav Horolma
Dr. Andreas Tillmar

ORGANIZING COMMITTEE

President:
Dr. Pamjav Horolma

Members:
Dr. Gunilla Holmlund
Dr. Denise Syndercombe
Dr. Pamjav Horolma
Dr. Andreas Tillmar

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GENERAL INFORMATION

Registration:
Registration desk:
The registration desk is to be found on the ground level of the Novotel Budapest Centrum.

Registration desk opening hours:
Wednesday, 31 August, 2016: 15.00 – 20.00,
Thursday, 01 September, 2016: 08.00 – 19.00,
Friday, 02 September, 2016: 08.00 – 19.00.
All Congress materials will be available at the registration desk at the conference venue during the conference.

Registration fee for participants includes: *name badge, congress bag, access to all scientific programs of meeting, program and abstract book, concert on Wednesday afternoon, welcome reception on Wednesday evening, lunch on Thursday, Friday, banquet dinner on Friday, coffee and soft drink tickets.*

Registration fee for accompanying persons includes: *welcome reception on Wednesday evening, English speaking guided sight-seeing tour on Thursday.*

Slide Preview Room opening hours:
Thursday, 01 September, 2016: 08.30 – 18. 00,
Friday, 02, September, 2016: 08.30 – 18.00.

Name Badges:
All registered delegates and accompanying persons are required to wear a congress name badge when attending sessions and social events. Admission tickets for the additional programs, social events are handed over at the registration desk.

Catering:
Coffee Breaks:
Refreshments and coffee will be served by the foyer during breaks. Tickets are handed over at the registration.
Lunch:
Lunch will be served in the restaurant of the hotel during lunch breaks. Tickets are handed over at the registration.

Certification of Attendance:
The Certificate of Attendance will be available at the registration desk on the last day of the Meeting.

Insurance:
The organizers do not accept any responsibility for injuries/damages or losses sustained by persons or personal belongings during the congress. Participants are strongly advised to carry appropriate travelling and health insurance.

Car parking:
There is a closed parking by the Novotel Budapest Centrum, with direct way to hotel mezzanine floor. The parking fee is to be paid individually.

PROGRAM

WEDNESDAY
31ST OF AUGUST, 2016

19:00 **GET TOGETHER – OPENING COCKTAIL**

THURSDAY
1ST OF SEPTEMBER, 2016

09:00-09:30 **WELCOME**

09:00–09:15 **Welcome to Budapest**
Dr. Judit Szekér (patron)
Network of Forensic Science Institutes, Budapest, Hungary

09:15–09:30 **Introduction to program**
Dr. Horolma Pamjav

09:30-10:15 **INVITED SPEAKER**

New technologies an introduction
Prof. Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen

10:15-10:45 **COFFEE BREAK**

10:45-12:25 **STR SEQUENCE VARIATION**
Chairs: *Prof. Niels Morling, Dr. Denise Syndercombe Court*

10:45–11:05 **Sequencing of 58 STRs using the Illumina® ForenSeqTM workflow and analysis of the data with the STRinNGS v.1.1 software**
Christian Hussing, Christina Huber, Rajmonda Bytyci, Helle Smidt Mogensen, Claus Børsting, Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

11:05–11:25 **Sequence variations in the short tandem repeat SE33 discovered by next generation sequencing**
Eszter Rockenbauer, Line Møller, Claus Børsting, Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

11:25–11:45 **Assessing the usefulness of sequence-specific allele frequencies across different populations using Massively Parallel Sequencing**
Laurence Devesse[1], David Ballard[1], Nicola Oldroyd Clark[2], Denise Syndercombe Court[1]
[1]King's College London, London, United Kingdom
[2]Illumina, Chesterford, United Kingdom

11:45–12:05 **Autosomal STR Variations Reveal Genetic Heterogeneity in the Mon-Khmer Speaking Group of Northern Thailand**
Dr. Antónia Völgyi[1], Jatupol Kampuansai[2], Wibhu Kutanan[3], Daoroong Kangwanpong[2], Horolma Pamjav[1]
[1]DNA Laboratory, Institute of Forensic Medicine, Network of Forensic Science Institutes, Budapest, Hungary
[2]Department of Biology, Faculty of Science, Chiang Mai University, Chiang Mai, Thailand
[3]Department of Biology, Faculty of Science, Khon Kaen University, Khon Kaen, Thailand

12:05–12:25 **Next generation sequencing and STR mutation rates – is sequence knowledge helpful?**
Federica Giangasparo, David Ballard, Laurence Devesse, Denise Syndercombe Court
King's College London, London, United Kingdom

12:25–13:30 LUNCH

13:30–16:50 POPULATION GENETICS
Chairs: *Dr. Horolma Pamjav, Ass. Prof. Gunilla Holmlund*

13:30–13:50 **Using relatives to find criminals in the Hungarian offender database**
Dr. Daniel Kling[1], Dr. Sándor Füredi[2]
[1]Norwegian Institute of Public Health, Division of Forensic Services, Oslo, Norway
[2]Department of Genetics, Hungarian Institute for Forensic Sciences, Budapest, Hungary

13:50–14:10 **Statistical and population genetics issues of two Hungarian datasets from the aspect of DNA evidence interpretation**
Dr. Zoltán Szabolcsi, Zsuzsa Farkas, Andrea Borbély, Gusztáv Bárány, Dániel Varga, Attila Heinrich, Antónia Völgyi, Horolma Pamjav
DNA Lab, Institute of Forensic Medicine, Network of Forensic Science Institutes, Budapest, Hungary

14:10–14:30 **Forensic and Population Genetic Investigations on the Turkish Cypriots from Cyprus**
Dr. Cemal Gurkan
Turkish Cypriot DNA Laboratory, Nicosia, Cyprus

14:30–14:50 **Allele frequency distribution for 15 STR loci among 7 islands in Indonesia and the genetic relationship in the world**

Prof. Toshimichi Yamamoto[1], Hajime Asai[1], Yuuji Hiroshige[1], Takashi Yoshimoto[1], Aya Takada[2], Masaaki Hara[2], Akira Ishii[1], Tuntas Dhanardhono[3]
[1]Department of Legal Medicine and Bioethics, Nagoya University, Nagoya, Japan
[2]Department of Forensic Medicine, Saitama Medical University, Iruma-gun, Japan
[3]Forensic and Legal Medicine, Diponegoro University, Semarang, Indonesia

14:50–15:20 COFFEE BREAK

15:20–15:40 **Analysis of next generation sequencing data of 11 STRs in 208 Somalis using the STRinNGS v1.0 software**

Eszter Rockenbauer, Anders Buchard, Susanne Lunøe Friis, Johanna Manninen, Claus Børsting, Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

15:40–16:00 **Evaluation of the DNA identification efforts for the victims and body parts from the terrorist attacks at Zaventem Airport and the Brussels subway station (Belgium)**

Prof. Ronny Decorte[1,2], Wim Van De Voorde[3,2], Nancy Vanderheyden[1], Joke Wuestenbergs[3], Wim Develter[3], Wouter Van Den Bogaert[3], Bram Bekaert[3,2]
[1]UZ Leuven, Department of Forensic Medicine, Laboratory of Forensic Genetics, Leuven, Belgium
[2]KU Leuven, Department of Imaging & Pathology, Leuven, Belgium
[3]UZ Leuven, Department of Forensic Medicine, Leuven, Belgium

16:00–16:20 **Forensic DNA Phenotyping: prediction of complex appearance traits with autosomal SNP markers**

Gusztáv Bárány, Ágnes Erős, Andrea Windbrechtinger, Zsuzsanna Farkas, Attila Heinrich, Antónia Völgyi, Horolma Pamjav
DNA Lab, Institute of Forensic Medicine, Network of Forensic Science Institutes, Budapest, Hungary

16:20–16:50 **Promega: Innovation, Science & Quality**

Stefan Kutranov
Promega

19:00- INVITED SPEAKERS' DINNER

FRIDAY
2ND OF SEPTEMBER, 2016

09:00–11:40 **MARKERS IN RELATIONSHIP TESTING**
Chairs: *Dr. Daniel Kling, Ass. Prof. Andreas Tillmar*

09:00–09:20 **Validation of the HID-Ion Ampliseq™ Identity Panel for relationship testing and reflections on the first 10 months of experience with MPS in real case**
Dr. Anders Buchard, Marie-Louise Kampmann, Claus Børsting, Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

09:20–09:40 **Increasing the power in paternity and relationship testing utilizing massive parallel sequencing for the analysis of a large SNP panel**
Dr. Andreas Tillmar, Ida Grandell
Department of Forensic Genetics and Forensic Toxicology, National Board of Forensic Medicine, Linköping, Sweden

09:40–10:00 **Kinship testing with (many) more markers**
Maarten Kruijver
VU University, Amsterdam, The Netherlands

10:00–10:20 **Effect of genetic inconsistencies on duo parentage testing using COrDIS Plus DNA marker system**
Dr. Vladislav Zavarin, Yevgeniy Krassotkin, Svetlana Vinogradova, Viktoria Smirnova, Andrei Semikhodskii
LLC Medical Genomics, Tver, Russian Federation

10:20–10:50 **COFFEE BREAK**

10:50–11:10 **Assessment of the ForenSeq NGS typing system in relationship casework**
Dr. David Ballard, Immy Riethorst, Laurence Devesse, Denise Syndercombe Court
King's College London, United Kingdom

11:10–11:20 **Short Case Presentation: Non-exclusion paternity case presenting with 3 genetic inconsistencies solved by massively parallel sequencing**
Laurence Devesse
King's College London, United Kingdom

11:20–11:40 **X chromosome investigation to resolve the genetic relationship in family members separated by several generations**
Dr. Balázs Egyed[1], Tibor Varkonyi[2], Akos Kertesz[3], Flora Fodor[4]
[1]Synlab GenoID DNA Laboratory, Budapest, Hungary
[2]Hungarian Society for Family History Research, Budapest, Hungary
[3]Synlab GenoID DNA Laboratory, Budapest, Hungary
[4]Synlab GenoID Molecular Diagnostic Laboratory, Budapest, Hungary

11:40-12:20 LUNCH

12:20-14:00 ASSESMENT OF RESULTS
Chairs: *Prof. Ronny Decorte, Prof. Klaas Slooten*

12:20–12:40 **Identifying common donors in DNA mixtures**
Prof. Dr. Klaas Slooten
Netherlands Forensic Institute, The Hague, The Netherlands

12:40–13:10 **Summarizing the results from the ESWG 2016 paper challenge**
Dr. Daniel Kling
Norwegian Institute of Public Health, Division of Forensic Services, Oslo, Norway

13:10–13:30 **Quality Assurance (QA) of the Biostatistical Workflow in Forensic Genetic Casework**
Ass. Prof. Andreas Tillmar
Department of Forensic Genetics and Forensic Toxicology, National Board of Forensic Medicine, Linköping, Sweden

13:30–14:00 **New ChrX- and SNP-Assays for use in kinship analysis**
Carole Peel[1], A. Prochnow[1], M. Bussmann[1], A. Tillmar[2], M. Scherer[1]
[1]QIAGEN GmbH, Hilden, Deutschland
[2]Department of Forensic Genetics and Forensic Toxicology, Linköping, Schweden

14:00-14:30 COFFEE BREAK

14:30 ESWG MEETING

19:00 BANQUET DINNER IN DOMONYVÖLGY

SATURDAY
3RD OF SEPTEMBER, 2016

07:00-10:00 BREAKFAST

New technologies - an introduction

Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Denmark

The most important new technology within forensic genetics today is Massively Parallel Sequencing (MPS). The technology is capable of sequencing the majority of the key forensic genetic systems as well as new markers that may be useful in advanced forensic case work and that will give us important new information. Thus, MPS offers a large amount of information concerning identity, ancestry, physical traits, etc., from minute amounts of DNA investigated in one or a few reactions. However, there are currently several challenges associated with MPS in forensic genetics, including certain technical aspects, transparent and efficient IT tools, nomenclature of STR sequences, etc. The latest recommendations on nomenclature of sequenced STRs and considerations on data analysis will be presented. The challenges concerning implementation of MPS based investigations will be discussed. An example of the implementation of a SNP based ISO 17025 accredited relationship testing system based on MPS will be presented. Other emerging MPS based investigations including e.g. phenotypical traits and ancestry informative markers will be discussed.

Sequencing of 58 STRs using the Illumina® ForenSeqTM workflow and analysis of the data with the STRinNGS v.1.1 software

Christian Hussing, Christina Huber, Rajmonda Bytyci, Helle Smidt Mogensen, Claus Børsting, Niels Morling

Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

The newly released ForenSeqTM workflow (Illumina®) provides simultaneous typing of 58 STRs of interest to forensic genetics, including 27 autosomal, 7 X-, and 24 Y-STRs, with massively parallel sequencing (MPS). The kit was developed for use with the MiSeqFGx instrument and the ForenSeq Universal Analysis (FUA) software to make up a complete forensic workflow based on the Illumina® sequencing technology.

The concordance of the results obtained with the MPS method and PCR-CE as well as the ability to type low template samples were tested. More than 50 % and 92 % of the STRs were correctly typed with ≥31.3 pg DNA and ≥250 pg DNA, respectively. Moreover, mixtures were typed in ratios from 1:1 to 1:1,000. The flagging system of the FUA software proved efficient in identifying allele imbalances and autosomal STR loci with >2 alleles.

A new version of the bioinformatic pipeline STRinNGS [1] was developed in-house for analysis of the ForenSeqTM assay. STRinNGS provided three features that were not accessible with the FUA software: 1) analysis of the STR flanking regions, 2) automatic naming of the SNP-STR alleles, and 3) detailed coverage information of all unique sequences from all samples in a run. Substitution variations and insertion/deletions were found in the flanks of 38 and 6 STR markers, respectively.

Locus and allele balances of the ForenSeqTM kit were clarified using STRinNGS. The large allele imbalances in D22S1045 and the very low coverage in DXS10103, DYS390, DYS460, and DYS522 resulted in unreliable genotype calls. High stutter ratios were seen for several loci and interfered with the genotyping of some of the Y-STRs.

Friis SL, Buchard A, Rockenbauer E, Borsting C and Morling N: Introduction of the Python script STRinNGS for analysis of STR regions in FASTQ or BAM files and expansion of the Danish STR sequence database to 11 STRs. Forensic Sci Int Genet. 2016; 21: 68-75.

Sequence variations in the short tandem repeat SE33 discovered by next generation sequencing

Eszter Rockenbauer, Line Møller, Claus Børsting, Niels Morling
Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

SE33 (ACTBP2) is a complex and highly variable short tandem repeat system consisting of four nucleotide repeat units (AAAG) interrupted by di-nucleotide repeat units (AA and AG). Owing to the wide span of allele length variation in the SE33 locus (alleles with lengths corresponding to 3-39 four-repeat units have been reported), SE33 was selected as one of the core loci in the European Standard Set of STRs for forensic genetic investigations, and is included in many commercial kits. Next generation sequencing (NGS) offers the possibility to exploit the true sequence variation of complex and compound STR systems. SE33 is however, not yet included in any commercially available NGS kit. There may be several reasons for this. Firstly, the SE33 alleles can be up to 343 bp long, and most NGS assays rarely produce reads longer than 300 bp. Secondly, the sequence of SE33 contains several poly-A stretches (up to five A's in a row) that might reduce the quality of the sequencing output. Here, we present NGS sequencing of the SE33 locus in 203 samples using the GS Junior instrument (Roche), a platform capable of producing sequence reads longer than 400 bp.

Assessing the usefulness of sequence-specific allele frequencies across different populations using Massively Parallel Sequencing

Laurence Devesse[1], David Ballard[1], Nicola Oldroyd Clark[2], Denise Syndercombe Court[1]
[1]King's College London, London, United Kingdom
[2]Illumina, Chesterford, United Kingdom

The transition in forensic genetics from capillary electrophoresis (CE) based approaches to massively parallel sequencing (MPS) provides both benefits and challenges, in addition to presenting an opportunity to review the currently used suite of genetic markers and assess how we utilise them.

Autosomal, X and Y STRs can now be analysed simultaneously and combined with identity-informative SNPs to maximize information recovery from degraded or low-template samples and phenotypic-informative SNPs can benefit no-suspect, no-hit cases. The number of discrete alleles distinguishable for autosomal STRs can also be increased due to the ability to detect intra-allelic variations within the repeat regions. Utilising markers that are already in common use ensures that sequencing-based methods generate data which aligns with STR and SNP calls generated using current methodologies, and affords compatibility with existing databases.

In this work, a series of samples from three different population groups (Caucasian, Chinese and West-African) have been genotyped using the Illumina ForenSeq™ DNA Signature Prep Kit and MiSeq® FGx System in order to investigate population variability at a sequence-level. This work demonstrates instances where sequence information can improve the level of discrimination at a targeted marker. We also identify and describe several markers where the level of discrimination of traditionally used autosomal STRs is significantly increased within the population groups studied, and how differences in sequence-specific population allele frequencies may be useful when identifying a sample of unknown origin.

Autosomal STR Variations Reveal Genetic Heterogeneity in the Mon-Khmer Speaking Group of Northern Thailand

Antónia Völgyi[1], Jatupol Kampuansai[2], Wibhu Kutanan[3], Daoroong Kangwanpong[2], Horolma Pamjav[1]

[1]DNA Laboratory, Institute of Forensic Medicine, Network of Forensic Science Institutes, Budapest, Hungary

[2]Department of Biology, Faculty of Science, Chiang Mai University, Chiang Mai, Thailand

[3]Department of Biology, Faculty of Science, Khon Kaen University, Khon Kaen, Thailand

Since the prehistorical period Mon-Khmer speaking people were recognized as indigenous ethnic groups living in northern Thailand. After the Tai colonization in the thirteenth century A.D., the Mon-Khmer inhabitants were fragmented, some were expelled to rural area while some were integrated to the mainstream of Tai society. Autosomal STR variations revealed that the present-day Mon-Khmer people could be genetically divided into two clusters, which appeared in consistency with the level of historical contact with the Tai majority ethnics. The cluster consisted of the Khamu, Lua, Paluang, Htin, who live in the remote area and had little historical contact with the Tai, appeared to maintain the Mon-Khmer ancestral genetic trace and genetically diverged from the Tai. The cluster comprised of the Mon and Lawa had exclusively close relationship with the Tai during the establishment of the prosperous Lan Na kingdom. A fraction of the Tai genetic component investigated in the Mon and some Lawa populations reflected the evidence of genetic admixture. However, some Lawa, who live in the mountainous area of Mae Hong Son province exhibited a unique gene pool, which might have been shaped by the founder effect during their historical fragmentation. The raising of genetic assimilation of the hill-tribe Karen into the Mon-Khmer and the Tai gene pools indicated that different language, culture, and geographical distance have lost their power to be a barrier of inter-ethnic marriages in the present days.

Next generation sequencing and STR mutation rates – is sequence knowledge helpful?

Federica Giangasparo, David Ballard, Laurence Devesse, Denise Syndercombe Court
King's College London, London, United Kingdom

With the advent of next generation sequencing (NGS) technologies, it is now possible to routinely discern the exact sequence of an STR allele. It is known that STR alleles mutate at a specific frequency between parents and children (in the general order of 1 mutation per 1000 transmissions for most forensically relevant STRs); additionally, research has discovered many different factors that can affect this rate and hence dictate the weight given to the possibility of a mutation when assessing the related state of two or more individuals.

There are multiple compound or complex STRs in common use within the forensic community that consist of a non-homogenous repeat region, such as D12S391 which is in essence two different STRs joined together, one with an AGAT repeat motif and one with an AGAC repeat motif. We have taken a set of confirmed parent-child mutations in such compound or complex STRs and analysed the precise nature of the mutation based on the change in allele sequence – from knowledge about how such STRs mutate, it is then possible to begin using more relevant mutation rates in kinship analysis depending on the specific sequence change. This has applications both to normal parent-child relationship tests and to assessments of more distant relationships with STR markers on the X or Y chromosome.

Using relatives to find criminals in the Hungarian offender database

Daniel Kling[1], Sándor Füredi[2]

[1]Norwegian Institute of Public Health, Division of Forensic Services, Oslo, Norway

[2]Department of Genetics, Hungarian Institute for Forensic Sciences, Budapest, Hungary

The Hungarian offender database contains more than 130,000 reference and trace profiles. The latter may be mixtures, i.e. profiles with more than one contributor. The profiles are typed on different STR marker kits, with the total number of markers ranging from 13 to 15. Several of the trace profiles are connected to high profile cases, e.g. murder, with no matches among the reference persons.

In crime cases where the trace profiles have yielded no successful leads, familial searching may instead be addressed to find the perpetrator. Familial searching promotes the idea that the relative of a person/trace in the database is the donor to a profile found at a crime scene. The presentation will provide a brief description of the concept, with some mathematical points, and its successful use in the Hungarian offender database resulting in five high profile cases obtaining new investigative leads.

Statistical and population genetics issues of two Hungarian datasets from the aspect of DNA evidence interpretation

Zoltán Szabolcsi, Zsuzsa Farkas, Andrea Borbély, Gusztáv Bárány, Dániel Varga, Attila Heinrich, Antónia Völgyi, Horolma Pamjav

DNA Lab, Institute of Forensic Medicine, Network of Forensic Science Institutes, Budapest, Hungary

When the DNA profile from a crime-scene matches that of a suspect, the weight of DNA evidence depends on the unbiased estimation of the match probability of the profiles. For this reason, it is required to establish and expand the databases that reflect the actual allele frequencies in the population applied. 21,473 complete DNA profiles from Databank samples were used to establish the allele frequency database to represent the population of Hungarian suspects. We used fifteen STR loci (PowerPlex ESI16) including five, new ESS loci. We examined the effects of the routinely used reference population data consisted of unrelated samples and the Databank data on the statistical interpretation of the DNA evidence. Due to the low level of inbreeding effect and the high number of samples, the new dataset provides unbiased and precise estimates of LR for statistical interpretation of forensic casework and allows us to use lower allele frequencies.

Forensic and Population Genetic Investigations on the Turkish Cypriots from Cyprus

Cemal Gurkan
Turkish Cypriot DNA Laboratory, Nicosia, Cyprus

Turkish Cypriot DNA Laboratory (TCDL) operates under the Turkish Cypriot Member Office of the Committee on Missing Persons in Cyprus (CMP) [1]. TCDL was set up in 2005 to contribute to the UN-led CMP Project on "Exhumation, Identification and Return of Remains of Missing Persons". To this end, a family reference sample (FRS) bank was created first, which currently comprises around 1,250 samples corresponding to the relatives of the 493 Turkish Cypriot (TC) missing persons (MiPs) from the 1963/64 and 1974 era. Since 2012, TCDL assumed full responsibility for the DNA typing of all TC FRSs, and so far issued over 500 17-loci autosomal STR profiles, which were used for familal DNA searching/matching with the profiles of skeletal remains obtained at international laboratories such as ICMP and Bode Technology. Furthermore, a DNA biobank currently comprising around 650 samples from the general Turkish Cypriot population was also created for research purposes, such as for the compilation of population genetic data requisite for statistical evaluations of MiP identifications. Results from such in house population studies based on autosomal STR (15-loci) and Y-chromosomal STR (17-loci) analyses were recently published [2,3], as well as those from larger international collaborations, such as on ancestry-informative (AI) single nucleotide polymorphisms (SNPs) and Insertion-deletion polymorphisms (InDels) analyses [4,5]. Further efforts to introduce additional DNA typing methods at TCDL are also in progress.

[1] Gurkan C, Demirdov D and Sevay H: Population genetics of Turkish Cypriots from Cyprus: forensic and anthropological implications. Forensic Sci. Int. Genet. Suppl. Ser. 2015; 5: e384-e386.

[2] Gurkan C, Demirdov D, Yamaci R and Sevay H: Population genetic data for 15 autosomal STR markers in Turkish Cypriots from Cyprus. Forensic Sci. Int. Genet. . 2015; 14: e1-e3.

[3] Terali K, Zorlu T, Bulbul O and Gurkan C: Population genetics of 17 Y-STR markers in Turkish Cypriots from Cyprus. Forensic Sci Int Genet. 2014; 10: e1-3.

[4] Bulbul O, Duvenci A, Zorlu T, Gurkan C, Santos C, Philllips C, Lareu M and Filoglu G: Studies of East European populations with a 46-plex ancestry-informative indel set Forensic Sci Int Genet Suppl Ser. 2015; 5: e16-e18.

[5] Pakstis AJ, Haigh E, Cherni L, ElGaaied AB, Barton A, Evsanaa B, Togtokh A, Brissenden J, Roscoe J, Bulbul O and others: 52 additional reference population samples for the 55 AISNP panel. Forensic Sci Int Genet. 2015; 19: 269-71.

Allele frequency distribution for 15 STR loci among 7 islands in Indonesia and the genetic relationship in the world

Toshimichi Yamamoto[1], Hajime Asai[1], Yuuji Hiroshige[1], Takashi Yoshimoto[1], Aya Takada[2], Masaaki Hara[2], Akira Ishii[1], Tuntas Dhanardhono[3]

[1]Department of Legal Medicine and Bioethics, Nagoya University, Nagoya, Japan
[2]Department of Forensic Medicine, Saitama Medical University, Iruma-gun, Japan
[3]Forensic and Legal Medicine, Diponegoro University, Semarang, Indonesia

Indonesia consists of more than 13 thousands of islands with more than 250 million people, is located geographically, historically and genetically important regions which connects between Asia and Oceania. In the present study, 409 DNA samples collected from 7 islands (Sumatra, Java, Bali, Kalimantan, Sulawesi, NTT: Nusa Tenggara Timor, and Papua) in Indonesia were genotyped for 15 autosomal STRs using an Identifiler plus kit, and the allele frequencies at each locus for each island and the pooled were calculated by counting. The tests for Hardy–Weinberg equilibrium (HWE) based on an exact test were performed, and the deviations from HWE were not observed at all loci in all of the populations except at D2S1338 in the NTT population after applying Bonferroni correction. The forensic statistic values such as the expected heterozygosities (expHz) were also calculated, the expHz for D2 or FGA were the highest in each population. The statistical pairwise difference Fst and Structure analysis revealed that Papuans show the most significant different from other populations.

Genetic structure analysis from totally 53 populations, were performed by mixing with the published genotype data from 46 worldwide populations. As a result, the elements patterns for the 7 Indonesian islands were similar each other, but very different from those of the otherworld wide populations. We also constructed a phylogenetic tree (Fst-NJ) from total 144 populations by adding the previously published allele frequency data from 92 worldwide populations, then 7 Indonesian populations located closed to the South-east Asian populations. Furthermore, the tree constructed by mixing with 8 island in Indonesia published just recently indicated that all of the 15 Indonesian made a cluster and the populations from each same island distributed in the same or close to cluster. Interestingly, the our Papua population distributed in the same cluster as the other Papua population with a longer branch, and our Papua, Sulawesi and NTT in the eastern area of Indonesia made a cluster with the other Papua, Sumba and Flores population reported previously. Accordingly, these suggested that even though allele frequency data only for 15 STRs could obtain a somewhat correct topology of a phylogenetic tree.

Analysis of next generation sequencing data of 11 STRs in 208 Somalis using the STRinNGS v1.0 software

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Next generation sequencing technologies (NGS) are well suited for STR sequencing and will provide alternatives to CE-based STR typing in forensic genetics in the near future. Presently, three test assays have been presented for the Ion PGM platform (ThermoFisher Scientific): the HID-Ion STR 10-plex Panel, the Early Access 24plex STR kit, and the Early Access Ion AmpliSeq GlobalFiler Mixture ID panel. One commercially available assay known as the ForenSeq DNA Signature Prep Kit has been launched for the MiSeq FGx platform (Illumina). One of the biggest challenges with NGS has been to design well-functioning, automated, and flexible software solutions to properly screen and analyze the huge amount of data produced by these technologies. We have recently published the first version of our software solution, STRinNGS v1.0, which finds the relevant STR region in the NGS output files, analyse the STR sequence and the flanking regions, names the alleles, and generates a report with the identified alleles and a bar plot of the STR profile. Here, we present results from the analysis of NGS data of 11 STRs in 208 Somalis using STRinNGS v1.0.

Evaluation of the DNA identification efforts for the victims and body parts from the terrorist attacks at Zaventem Airport and the Brussels subway station (Belgium)

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On March 22, 2016, a bomb exploded around 8 AM in the departure hall of Zaventem Airport (Brussels, Belgium) followed immediately by a second bomb explosion. While rescue teams were helping the wounded victims and evacuating people from the airport, a third bomb exploded at 9:11 AM in the subway station Maalbeek, close to the European parliament in the center of Brussels. In total, 32 victims died in the explosions or later in hospital, three out of four terrorists died immediately, and over 200 victims were transferred to different hospitals. The identification process of the deceased victims was concentrated at the Military Hospital in Brussels for the victims of the subway station and at the University Hospitals in Leuven for the victims of Zaventem Airport. The last two victims of the two terrorist attacks were formally identified (ante-mortem and post-mortem information, DNA analysis) on March 29th. On March 27th and March 31st, a second DNA identification process started with the analysis of body parts (larger than 5 cm) that had been collected at both locations. In total 177 DNA profiles for Maalbeek and 286 DNA profiles for Zaventem have been established from the body parts, reference samples from the deceased victims and from the three terrorists. On April 25th, all body parts were identified except for 12 fragments that could not be associated with a deceased victim or one of the terrorists of Zaventem Airport. While the DNA identification process was finished within a month, some recommendations from this case can be formulated for future DNA identifications.

Forensic DNA Phenotyping: prediction of complex appearance traits with autosomal SNP markers

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Forensic DNA Phenotyping refers to the prediction of appearance traits of unknown sample donors, directly from biological materials found at the scene. Currently, group-specific pigmentation traits are already predictable from DNA, while several other externally visible characteristics are under genetic investigation.

The aim of this study to develop complex phenotype prediction model involving hair, iris and skin colour, and with the usage of 21 autosomal SNP markers. We have investigated 597 Hungarian, 69 African (Ghanaian) and 100 Mongolian volunteers. The iris colours were defined based on the laboratory and the Martin-Schultz scale, hair colour with Fischer–Saller scale, and skin colour with Fitzpatrick scale. Furthermore, we defined in a two scale base the dark and light complex phenotypes.

Based on the G-test p values (genotypes) the phenotype specific SNPs were selected for each trait. The prediction models were built with multinomial logistic regression (R-Gui version 2.15.3, nnet package). All models were verified with receiver operating characteristic curve (ROC) and evaluated based on AUC values (ROC area under curve).

Finally, 14 SNP markers were selected located in 9 different genes. Four different prediction models were built with different SNP sets: first to predict iris colour, second to predict hair colour, the third to predict skin colour, and the fourth that can predict dark and light complex phenotypes. The model works in user-friendly Microsoft Excel surface.

The applications lead to description of unknown persons from crime scene DNA samples with reasonably high accuracies delivering information to police investigations and missing person cases.

Promega: Innovation, Service & Quality

Stefan Kutranov
Promega

Promega continues to support the Forensic DNA community by providing innovative tools, unrivalled service and support and the highest quality products possible. The Spectrum CE System and the new range of 8-dye STR chemistries will provide new opportunities to obtain high levels of information from the most difficult samples while at the same time enabling highly efficient and flexible workflow possibilities for all Forensic DNA laboratories. To further assist in the creation of optimised workflows, Promega's team of automation experts are able to implement and optimise all of our chemistries, from extraction through to CE setup, on all of the common large-platform automated liquid handlers. Combined with our involvement in the creation of the new 'Forensic Grade' ISO standard (ISO 18385), through our Manufacturing and Quality Assurance experts, and our commitment to providing the highest quality products, Promega is able to provide full, optimised and forensically safe solutions to support all Forensic DNA laboratories.

Validation of the HID-Ion Ampliseq™ Identity Panel for relationship testing and reflections on the first 10 months of experience with MPS in real case

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The HID-Ion Ampliseq™ Identity Panel (ThermoFisher Scientific) is a massively parallel sequencing (MPS) assay in which 90 autosomal and 34 Y-chromosome SNPs are typed simultaneously using the Ion PGM™ system (ThermoFisher Scientific). The SNPs in the panel is a subsample of human identification SNPs originally included in the SNPforID and Kenn Kidd panels. Furthermore, 34 upper-clade Y-chromosome SNPs are typed. We typed 100 Danes (50 males and 50 females) in duplicates using the HID-Ion Ampliseq™ Identity Panel. The samples were previously typed with an ISO17025 accredited SNaPshot based version of the SNPforID assay allowing for a direct comparison of allele calls. The dataset was produced in order to validate the assay for relationship testing according to the ISO17025 standard. An in-house script was developed in order to make the analysis of the data easy and to allow for minimum acceptance criteria for each SNP. In-house scripts were developed to automate comparisons of duplicate typings and export of data to the final report written in word. In general, the kit performed well. However, the heterozygote balances of two autosomal SNPs, rs7520386 and rs576261, were not satisfactory and these SNPs are not reported. An overview of the experiences with the assay in an ISO17025 accredited laboratory will be presented.

Increasing the power in paternity and relationship testing utilizing massive parallel sequencing for the analysis of a large SNP panel

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Within forensic genetics, there is still a need for supplementary DNA marker typing in order to increase the power to solve cases for both identity testing and complex relationship issues. We have designed a customized GeneRead DNASeq SNP panel (Qiagen) including 140 autosomal forensically relevant identity SNPs, using massive parallel sequencing (MPS) for the analysis. We have validated and implemented the SNP panel to be used as supplementary markers for inconclusive paternity cases and for complex relationship cases in routine casework. The validation showed that the analysis of the SNP panel is robust, reproducible, and has a high accuracy. Validation of methods based on MPS will include the investigation of other validation parameters compared to when using standard analysis with capillary electrophoresis. For example quality measurements and thresholds were established for parameters such as marker coverage and allelic balance. Biostatistical aspects of using data from the SNP panel were also studied and validated. This included the establishment of allele frequencies, studies of the effects and the handling of linkage and linkage disequilibrium on a case-by-case basis. Moreover, we show, by simulations and with data from routine case examples, that the usage of this large SNP panel will be helpful for solving paternity and relationship cases which earlier have shown to be ambiguous when using information from 29 STR markers. In summary, we have designed, validated and implemented a method based on MPS technology which have been shown to increase the power in paternity and relationship testing.

Kinship testing with (many) more markers

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The most commonly used multiplexes have limited power of discrimination beyond standard cases such as paternity testing. The upcoming generation of multiplexes, and the emerging NGS kits, will include more loci, reducing false positive rates and increasing statistical power. We study the extent to which kinship testing and familial database searching will benefit from expanding the locus set. We find that the power of discrimination for first-degree relatives quickly becomes so large that database searches will become possible without the need to eliminate false positives. On the other hand, discriminating between more distant relatives and unrelated persons remains difficult, unless thousands of markers are used. We make use of a novel R-package implementing a version of the Lander-Green algorithm incorporating both mutations and linkage.

Effect of genetic inconsistencies on duo parentage testing using COrDIS Plus DNA marker system

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Following ISFG 2007 recommendations [1] when genetic inconsistencies between DNA profiles of the child and an alleged parent are observed the decision as to parentage exclusion should be based on comparing the obtained CPI value for the case with the threshold CPI value, at which parentage is considered not excluded, adopted by the laboratory and not on the number of inconsistent loci observed between the two DNA profiles. It has been shown that when 3 out of 13 CODIS STR markers are inconsistent the CPI value would be in the region of 1/4,600 [2]. The purpose of the present study was evaluation of the effect of small number of genetic inconsistencies on duo parentage cases (both paternity and maternity) using COrDIS Plus STR marker system. COrDIS Plus (GORDIZ Ltd, Russia) marker system contains 19 autosomal STR loci (D3S1358, TH01, D12S391, D1S1656, D10S1248, D22S1045, D2S441, D7S820, D13S317, FGA, TPOX, D18S51, D16S539, D8S1179, CSF1PO, D5S818, VWA, D21S11, SE33) and Amelogenin as gender marker. The experimental sample consisted of 3,129 duo parentage tests (both paternity and maternity) performed in our laboratory. No genetic inconsistencies between the child and alleged parent were observed for 2,446 (78.17%) cases but in 74 cases (2.36%) up to 4 genetic inconsistencies were detected. A single inconsistence was found to be present in 51 cases (in 8 of these cases a partial profile was obtained for one or both participants) with the minimum CPI=241. In 3 cases we observed inconsistencies at 2 STR loci (minimum CPI=0.0711). Inconsistencies at 3 loci were found to be present in 5 cases (in 3 of these cases a partial profile was obtained for one or both participants) with the value of maximum CPI=1/27,395. For 15 cases we found inconsistencies for 4 STR loci (in 5 of these cases a partial profile was obtained for one or both participants) with the maximum CPI=1/1,758,652. Overall, if the threshold value of CPI=1/1,000 is adopted results for all the cases with 3 and 4 genetic inconsistencies will be in favour of exclusion of paternity/maternity in question.

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Assessment of the ForenSeq NGS typing system in relationship casework

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The ForenSeq DNA Signature Prep Kit (Illumina) amplifies 27 autosomal STRs along with 94 identity SNPs. Analysed using a next generation sequencing (NGS) methodology, the combination of numerous markers along with the provision of precise allele sequence information provides an opportunity to improve the calculated likelihood ratio in identification and complex relationship scenarios.

In identification situations where DNA may be degraded, it is likely that amplification will be more successful with the SNP markers than STRs, and here we have assessed how the 94 SNP markers within this kit perform, and how useful they are in parent-child duos to either correctly exclude or include paternity/maternity in the absence of STR results. We have additionally quantified the benefit to half-sibling relationship investigations that an NGS approach, encompassing the extra markers and knowledge of STR allele sequence, has over standard capillary electrophoresis STR typing.

Short Case Presentation: Non-exclusion paternity case presenting with 3 genetic inconsistencies solved by massively parallel sequencing

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We describe a paternity case where the presence of 3 genetic inconsistencies had previously led two laboratories to exclude paternity in favour of an avuncular relationship. On addition of extra CE based STR systems, no further inconsistencies were discovered, leading us to analyse these samples using the MiSeq FGx™ based ForenSeq system in order to clarify the relationship. A total of 129 additional markers were tested, revealing no further inconsistencies and providing a LR in support of paternity.

X chromosome investigation to resolve the genetic relationship in family members separated by several generations

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In genetic relationship analyses – for example in family tree investigation – usually Y chromosome or mitochondrial DNA examinations are carried out to prove the genetic heritage along the questioned family members on both paternal and maternal lineages, mainly if there is no obviouse chance to confirm the kinship by analysing autosome markers. Seldom, especially if family members are missing or in cases where the paternal or maternal lineage is interrupted the X chromosome analysis can only help to resolve the questioned relations.

Here we present the X chromosome analyses of thirteen living members of a questioned family tree dating back the founder father to the 19th century whose mother is questioned. Because of the interrupted paternal and maternal lineages in the examined family only X chromosome investigation was supposed to be informative to resolve the questioned connection between the two branches of the family tree. Among the recent donors the closest presumed relatives are nine generations apart from each other's at least, and the most farest pairs are in twelve generations distance. Altogether twelve X chromosome STR markers have been examined on each recent donors by means of the Argus X-12 QS system (Qiagen) and the resulted haplotypes have been biostatistically evaluated and reported on FamLinkX software [1]. The casework proves that in case of missing donors or of interrupted lineage markers X chromosome analyses can be informative to prove or resolve questioned genetic relationship along several generations as well.

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Identifying common donors in DNA mixtures

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Mixed DNA profiles are often compared to reference profiles in order to obtain evidence on the identity of the donors, but they are not always compared to other mixtures.

We explain how to compare DNA mixtures in such a way as to obtain statistical evidence indicating whether they may have a contributor in common. The mixtures may have allelic dropout.

This information can be useful to establish links between cases.

We report on the comparison of the mixtures in the Dutch DNA database carried out in this way.

Summarizing the results from the ESWG 2016 paper challenge

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The English Speaking Working Group (ESWG) annually aims to provide members with a proficiency test relating to the use of statistical methods when inferring relationships based on DNA data. The current study will provide a summary of the data for 2016 as well as suggested solutions. In addition, any difficulties or deviating results will be discussed.

The paper challenge of 2016 consists of a regular paternity case as well as a questionnaire, summarizing the laboratory practice. In addition, the paper challenge contains a more complex relationship case with inbreeding. The latter will be discussed and a brief evaluation of the level of difficulty will be assessed.

Quality Assurance (QA) of the Biostatistical Workflow in Forensic Genetic Casework

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Quality assurance (QA) is a key element in an accredited forensic laboratory. Quality involves several components such as validation of methods, instruments, and software; documented maintenance; secured chain of custody; documented operating procedures; and proven competence of the staff. The general goal is to produce test reports of forensic genetic investigations with legal certainty. DNA is a powerful tool in forensic analysis for linking a suspect to a crime scene, resolving biological relationships, and identifying disaster victims. Traditionally, DNA investigations can be divided into two parts: (1) the establishment of DNA profiles; and, (2) the evaluation of the evidential weight of these DNA profiles, given some hypotheses about the true circumstances. There are well-documented standards for quality assurance of the first part, but for the second part, guidelines have not yet fully been established within the community. The evaluation of the weight of evidence, using a biostatistical workflow, includes several components such as different computational methods, reference data, secured transfer of case data, and expert opinions made by reporting officers. Sophisticated biostatistical computation models are implemented in dedicated software packages making up the basis of the biostatistical workflow. To assure the quality of this workflow requires not only validation of software packages per se, but also validation of population reference databases and reference parameter settings and it embraces qualification demands for the authorization of competence of expert practitioners. We have designed, validated and implemented a biostatistical workflow for use in paternity and relationship testing and for missing person identification. The workflow contains four different software packages and population reference data for more than 50 autosomal, X-chromosomal, and Y-chromosomal DNA markers from different populations. Parameters for the validation of this biostatistical workflow have been established and will be discussed as well as other QA aspects. Moreover, competence requirements for the staff specific for each software and type of casework have been implemented. The requirements also include maintenance of competence through annual exercises.

New ChrX- and SNP-Assays for use in kinship analysis

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The standard practice of forensic kinship evaluation uses unlinked autosomal and Y-chromosomal markers routinely. However, X-chromosome markers have gained recognition as a powerful tool to complement the information provided by autosomes, particularly in solving complex deficiency cases, but are still not validated by all paternity laboratories. We will present the new Investigator Argus X-12 QS Kit and will focus on its features and benefits compared to the former product configuration. The kit makes use of the recently developed Fast Reaction Mix 2.0, which allows shorter PCR cycling protocols, the ability for direct amplification and a higher inhibitor tolerance. Furthermore, a Quality Sensor has been added as an internal control to indicate if the PCR reaction has performed properly. D21S11 was included as an autosomal alignment marker. By comparing the genotype of the alignment maker between X-chromosomal and autosomal STR analysis done within a case, potential mix up of samples can be detected.

In addition, there's no doubt that the future of human identification will be heavily influenced by next-generation sequencing (NGS). QIAGEN is already working closely to help early adopters realize the practical benefits of NGS in their routine testing workflows. These benefits include the ability to analyze a much larger number of markers in a single test, thereby increasing discrimination power and the statistical significance of the results obtained. This requirement for increased discrimination is increasingly demanded in complex paternity cases, for example, in cases involving siblings and other closely related individuals, where the standard short tandem repeat (STR) test cannot always provide sufficient statistical values to identify the true relationships with adequate confidence. Our new SNP ID enrichment panel QIAseq Investigator SNP ID, along with the library prep kits and software, enabled users of MiSeq and Ion Torrent platforms the development of a streamlined and focused workflow for the generation and analysis of SNP data in complex paternity cases.

We will show internal and external data of the development and validation of both technologies and demonstrate the advantages for their use in paternity and kinship testing.

INDEX