University: The University of Hong Kong
Unit of Assessment (UoA): 11 - Mathematics & Statistics

Title of case study:
Statistical assessment and user-friendly software for complex paternity and kinship testing and DNA mixture analysis in legal contexts

(1) Summary of the impact

DNA testing is the most powerful tool in parentage and kinship determinations, and for mixed stain analysis in criminal investigations. Novel statistical methods and user-friendly software developed by researchers at the University of Hong Kong (HKU) have provided a simple means of assessing the weight of DNA evidence. The software has been used by forensic and paternity laboratories worldwide in evaluating thousands of cases, with very positive feedback on its analysis of complex kinship and DNA mixtures. A particularly striking example of the impact of HKU's tool was its success in identifying three badly burnt bodies in the Hot-Air Balloon Burst Tragedy in Egypt, the investigation of which was concluded in the Hong Kong Coroner's Court in 2016.

(2) Underpinning research

DNA evidence has emerged as the most important forensic tool in assessing the culpability or innocence of a suspect in a criminal investigation. But statistical evaluations of the weight of DNA evidence, as required by the court, can be very challenging. Professor WK Fung's research in this area began 20 years ago, when he helped the Hong Kong Government Laboratory to assess the DNA evidence in a case where a couple, whose newborn girl was found missing in a hospital, claimed parentage of an abandoned baby girl found on the street.

After the Court of Final Appeal's rulings of a right of abode case in 1999, the HKSAR Government estimated that the number of Mainland people who were eligible for the right of abode in Hong Kong was 1.67 million. In 2001, Fung was invited to brief the Hong Kong Legislative Council (similar to the Parliament in UK) on "DNA Analysis in Parentage Testing" (see minutes of the Legislative Council http://www.saasweb.hku.hk/EasyDNA/LEGCO.pdf). At that time, the Council was deliberating on a bill that would give Mainland-born children of Hong Kong parent(s) right of abode. DNA testing was needed to establish familial relationships between the children and their claimed parent(s). Fung's work helped to address problems such as unavailable parental samples, or close-relative impersonation. This fueled further research into the hitherto little-studied area of statistical analysis of biological relationships. However, statistical evaluations of kinship among ten or more people, in a complex pedigree of three generations or more, are extremely complicated. Based on conditioning arguments and pedigree analysis, taking possible mutations into account, Fung developed the general statistical methodology on complex kinship assessment [1, 2]. This work resulted in the development of a suite of user-friendly computer software [1]. The research findings were presented as a Keynote Speech in the 17th Triennial Meeting of the International Association of Forensic Sciences, 2005, a key target audience of practitioners.

In criminal investigations, DNA-carrying materials, such as blood, semen, saliva and skin are often encountered in admixtures, with multiple contributors. Therefore, in a rape case, the victim's DNA, from her vaginal secretions, is mixed with that from the semen left by the culprit; or, in volume crimes like theft, the culprit's DNA left on doorknobs is often mixed with that from the inhabitants. In concert with the international forensic science community, Fung has made considerable contributions in researching this topic. Based on combinatorics and the theory of population subdivision, Fung has

worked out a globally applicable method to evaluate DNA results from admixtures, accounting for population sub-structure [3] and contributors of different ethnicities [4]. Fung solved the DNA mixture problem in the OJ Simpson case where the defendant was an African-American, the victim was a Caucasian and other unknown contributors could have been Hispanics or of other ethnicities [4].

Using a set of DNA results to trawl for a possible match/suspect in a DNA database, although commonplace, has prompted arguments as to what statistically valid evidential weight should be accorded such a match. Fung proposed a novel approach in resolving the well-known "DNA database search controversy" in the DNA mixture context [5]. The methods and software that were developed [3, 4] provide a conservative estimate of the culpability of suspects if they are identified through a database search.

Fung has been a Professor at the university since 1988. His research team includes Chung, a PhD student (2005-2009), and a lecturer since 2005; and Hu, a visiting research associate (2000-2003), a PhD student (2003-2007), and then assistant professor (2007-2009).

(3)   References to the research

1. **Fung, W.K.** (2003). User-friendly programs for easy calculations in paternity testing and kinship determinations. *Forensic Science International*, **136**: 22-34.
2. **Fung, W.K.**, Carracedo, A. and **Hu, Y.Q.** (2003). Testing for kinship in a subdivided population. *Forensic Science International*, **135**: 105-109.
3. **Fung, W.K.** and **Hu, Y.Q.** (2000). Interpreting forensic DNA mixtures: allowing for uncertainty in population substructure and dependence. *Journal of Royal Statistical Society, A,* **163**: 241-254.
4. **Fung, W.K.** and **Hu, Y.Q.** (2001). The evaluation of mixed stains from different ethnic origin: general result and common cases. *International Journal of Legal Medicine*, **115:** 48-53.
5. **Chung, Y.K.**, **Hu, Y.Q.** and **Fung, W.K.** (2010). Evaluation of DNA mixtures from database search. *Biometrics*, **66**: 233-238.

(4)   Details of the impact

Determinations of kinship in cases such as incest, multiple relatives and missing persons can be very complicated. Fung derived the optimal methodology for such testing. In order to substantially reduce the great challenges that forensic scientists and chemists faced in analyzing complex kinship problems, computer software comprising four modules was developed. The software expedites analysis in a wide spectrum of kinship testing scenarios (e.g., standard trio, motherless, motherless-with-relatives-of-the-mother-typed, missing person case with relatives typed, incest cases, inheritance dispute cases). Due to its user-friendly interface based on pedigree diagrams and its efficient data processing in returning a result within just one minute, the software was named "EasyDNA_In_1_Minute". The software can resolve various complex familial relationships.

Besides parentage/kinship testing, DNA profiling is widely applied in crime investigations, such as DNA mixture analysis. To credibly ascribe individual DNA characteristics found in an admixture of DNA-contributing materials in a test sample to possible contributors with statistical validity (and the evidential weight that this provides) is a complicated process, and prone to mistakes if due attention has not been exercised (e.g., the OJ Simpson trial, https://www.latimes.com/archives/la-xpm-1995-06-24-mn-16688-story.html). Fung and his colleagues provided the appropriate methodology and developed the computer software "EasyDNA_Mixture" which can handle complex mixture cases involving genetic relatedness, missing suspects and structured populations. This mixture software and

the "EasyDNA_In_1_Minute" software on complex kinship determinations were originally commercially available, but, together with two other items of software on population data analysis and simple paternity testing, they have been on open source since 2008 (http://www.saasweb.hku.hk/EasyDNA). They have attracted over 1,600 downloads since 1 October, 2013 [A], from practitioners in private paternity testing and government forensic laboratories all over the world (e.g., Austria, Australia, Argentina, Brazil, Canada, China, France, Italy, Korea, Malaysia, Mexico, Russia, Singapore, South Africa, Switzerland, UK, USA, Zimbabwe).

The software has analyzed thousands of cases worldwide in the assessment period. The significant impact of the EasyDNA programs is strongly demonstrated by the Hot-Air Balloon Burst Tragedy, which occurred in Luxor, Egypt, in 2013, with the investigation being concluded at a Coroner's Court held in Hong Kong in February, 2016 [B]. As indicated in the letter from the Hong Kong Government Laboratory: *"Three (out of nine Hong Kong people who died) of the bodies were badly burnt"*. *"Hence, DNA matching was the only means to verify their identity"*. *"Complicated tree relationship maps had to be built"*. *"As some of the family tree diagrams had become complex and involved various non-standard, kinship scenarios with the unidentified deceased, …, the EasyDNA program designed for missing person verification had been extremely useful and had offered tremendous help in this situation"* [C].

The paternity and kinship software is not only useful for civil cases, but also powerful for forensic crime investigations. Dr Otteson of GenQuest DNA Laboratory, USA, commented that *"we completed approximately 200 forensic cases using this software"; "This software was very useful in completing about 20 incest cases and about 30 missing persons"*[D] − both types of cases that can be very complicated and difficult to analyze. A sample of the favourable comments received from users (from Austria, Hong Kong, Russian Academy of Sciences, UK, USA) of the software include: *"We completed approximately 10,000 relationship case"* [D]. *"This software is extremely well-developed based on beautiful statistical theories. The EasyDNA_In_1_Minute can easily handle extremely complicated kinship determinations involving more than 10 persons"* [E] (the majority of other software can only handle simple (≤ 3 people) kinship testing). *"uses it to deal with up to 100 cases per year." "it runs excellently without any problems"* [F]. *"deal with about 100 cases per year." "It is extremely helpful … including various complex cases". "The results would be obtained instantaneously – just in seconds!!". "It is the most user-friendly software in the field"* [G]. *"used routinely for our criminal case work". "capable of generating reliable results for court trials". "All case reporting officers are required to apply the software as a standard practice". "In 2017, … used for reporting over 70 cases"* [C]. Comments received from users in other countries such as Czech Republic, France and Germany are not included for brevity.

Fung provided expert witness advice to the Legal Aid Department in Hong Kong in an inheritance dispute in 2013, involving complex kinship determinations among people with a complicated family tree of four generations or more [H]. The trials were held in the High Court in November, 2013 [H].

The impact of Fung's research has considerable reach internationally. In March 2014, he was invited to hold a paid two-day workshop titled "Advanced Kinship Statistics" for 37 Government forensic scientists in Singapore, receiving this feedback: *"Prof Fung is a recognized leader and was most highly recommended to us by Prof Bruce Budowle, Director, University of North Texas Center for Human Identification, an authority and pioneer in forensic genetics". "Truly a great teacher, he explained difficult concepts in layman's terms and used numerous easy-to-understand examples to demonstrate complex statistical calculations. This was greatly appreciated by our staff". "Prof Fung also demonstrated the analytical solutions for six paper challenges of complex kinship testing obtained from CAP Paternity Proficiency Test using the simple-to-use software (EasyDNA) that he created". "The workshop conducted by Prof Fung was extremely relevant and impactful … to address the complex challenges they face in the statistical treatment of difficult kinship cases"* [I]. Fung is

often consulted by forensic scientists in China, Hong Kong, Macau and Singapore, and directly by officers of the Hong Kong Police Force.

(5)   Sources to corroborate the impact

A.   [October 2018] Numbers of downloads of the EasyDNA, Simple Paternity Testing and Population Data Analysis software.
http://www.saasweb.hku.hk/EasyDNA/Download.pdf

B.   [February 2016] A news item on the inquest of the Coroner's Court on the deaths of 9 Hongkongers in the Luxor Hot Air Balloon Tragedy.
http://www.saasweb.hku.hk/EasyDNA/HotAirBalloon.pdf

C.   [January 2019] A letter from Christina Li, Senior Chemist, Forensic Science Division, Hong Kong Government Laboratory.
http://www.saasweb.hku.hk/EasyDNA/HKGL.pdf

D.   [November 2018] A letter from Dr Elmer Otteson, GenQuest DNA Laboratory, USA.
http://www.saasweb.hku.hk/EasyDNA/Otteson.pdf

E.   [May 2019] A letter from Dr Igor Kornienko, Chief Researcher of Southern Scientific Centre, Russian Academy of Sciences, Russia.
http://www.saasweb.hku.hk/EasyDNA/Kornienko.pdf

F.   [October 2018] A letter from Professor Dr Erwin Petek, Deputy Institute Director, Vice Dean of Studies, Diagnostics and Research Institute of Human Genetics, Medical University of Graz, Austria**.
http://www.saasweb.hku.hk/EasyDNA/Petek.pdf

G.   [November 2018] A letter from Dr S. Hadi, Reader in Forensic Genetics and Medicine, School of Forensic and Applied Sciences, University of Central Lancashire, UK**.
http://www.saasweb.hku.hk/EasyDNA/Hadi.pdf

H.   [November 2013] A High Court case on an inheritance dispute between Tsang Ka Fai (Plaintiff) and Tsang Wan Kiu (1st Defendant) and Wong Kin Chau (2nd Defendant).
http://www.saasweb.hku.hk/EasyDNA/InheritanceDisputeExpertOpinion.pdf

I.   [June 2019] A letter from Dr Christopher Syn, Director, Biology Division, Health Sciences Authority, Singapore.
http://www.saasweb.hku.hk/EasyDNA/Syn.pdf

** These institutions provide consultancy services to individuals, companies, lawyers and courts.