GOOD PRACTICE GUIDE FOR THE **USE OF FORENSIC GENETICS**

IN INVESTIGATIONS INTO HUMAN RIGHTS AND INTERNATIONAL HUMANITARIAN LAW VIOLATIONS

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PROLOGUE

On the occasion of the 63rd General Assembly of the United Nations, in September 2008, the Argentine President, Cristina Fernández de Kirchner, stressed the importance of creating DNA data banks as a mechanism "to account for human rights violations and identify victims".
In line with this presidential initiative, in March 2009 and October 2010, Argentina became the main co-sponsor of two resolutions within the context of the United Nations Human Rights Council (Resolution 10/26[1] and Resolution 15/5[2]), which were adopted by consensus and were co-sponsored by more than forty countries.
These resolutions had three essential goals: 1) recognizing the importance and promoting the use of forensic genetics as a significant tool of forensic sciences, in cases of serious violations of human rights and international humanitarian law; 2) promoting greater cooperation between states and organizations in the application of forensic genetics —in particular— and forensic sciences —in general— to the investigation of those violations; and 3) promoting the development of international standards to guide the application of forensic genetics.
In relation to the third goal, the resolutions adopted by the Council provided for the possibility of creating an internationally agreed-upon guide for application, with the relevant guarantees, in cases of serious human rights violations.
Therefore, Argentina, together with the International Committee of the Red Cross and a group of experts, decided to establish a Working Group for the creation of this Guide, which entailed a long drafting process and the gathering of contributions from various specialists.
It is not a coincidence that our country has decided to take the lead in an initiative such as this. The work and history of organizations such as Grandmothers of Plaza de Mayo and the Argentine Forensic Anthropology Team (EAAF) bear witness to the experience that we have gained and that we can now share with the international community.
The Guide was designed as a tool that will be available to the courts, legislators, government officials, human rights defenders and relatives of victims and its main goal is to guide the application of forensic genetics and the creation and management of DNA data banks within the framework of investigations relating to the identification of victims of violations of human rights and/or international humanitarian law.
There is no doubt that forensic genetics is currently an exceptional tool which, through this Guide, will be available to all States in order to comply with their obligations in cases of human rights violations, thus guaranteeing the rights to justice, truth and identity.

Héctor M. Timerman
Ministro de Relaciones Exteriores y Culto

1 The draft Resolution was co-sponsored by 45 countries: Argentina, Bolivia, Uruguay, Chile, United Kingdom, Brazil, Canada, Switzerland, Germany, Egypt, Italy, France, Ukraine, Saudi Arabia, Bosnia-Herzegovina, Slovakia, Bahrain, Japan, South Africa, Mexico, Republic of Korea, Croatia, Romania, Colombia, Luxemburg, Morocco, Peru, Austria, Spain, Montenegro, Cyprus, Denmark, Portugal, Poland, Finland, Costa Rica, Latvia, Ecuador, Belgium, Ireland, Dominican Republic, Guatemala, Serbia, Israel and the United States.

2 The draft was co-sponsored by 48 countries: Argentina, Bolivia, Uruguay, Chile, United Kingdom, Brazil, Switzerland, Germany, Egypt, Italy, France, Saudi Arabia, Bosnia-Herzegovina, Slovenia, South Africa, Mexico, Republic of Korea, Croatia, Japan, Colombia, Morocco, Peru, Austria, Spain, Hungary, Cyprus, Denmark, Portugal, Poland, Finland, Costa Rica, Latvia, Ecuador, Belgium, Ireland, Andorra, Azerbaijan, Armenia, Guatemala, Serbia, Greece, Netherlands, Paraguay, Panama, Macedonia, Palestine, Norway and the United States.

Argentina's experience in the long process of Memory, Truth and Justice that began with the restoration of democracy shows that collective construction is an indispensable stepping-stone to achieve the kind of moral reparation for victims and for society as a whole that only justice can provide.

The three concepts -Memory, Truth and Justice- become intertwined and reinforce each other when a nation overcomes genocide. Since crimes against humanity are not subject to any statute of limitations, Memory is the only reservoir that a nation has to keep the claim for justice alive, as well as to fight against the genocidal strategy of oblivion.

Memory is the history of a people. It is its resistance to official history. In the case of Argentina, Memory was the fight against the story told during the military dictatorship and many years of democracy, according to which the disappeared were "walking around in Europe" and our country underwent "a war in defence of Western and Christian values". Memory is the testimony of the victims and their relatives, who, for decades, suffered from slander and abandonment. This is why Memory, in essence, belongs to the people: because it represents resistance to hegemonic violence and because it has a strong component of oral transmission. Finally, Memory is an approach to the past; it is a political interpretation of and position on a historical moment. When that interpretation is based upon physical evidence, the genocidal discourse becomes nothing but a huge alibi to avoid prosecution and guarantee impunity. It is at this point that Memory and Truth come together.

In view of the pact of silence and the complicity shown by the sectors that benefited from the military dictatorship, the work of forensic genetics becomes fundamental. When the Argentine Forensic Anthropology Team (EAAF) looks for, finds and identifies the bodies of persons who disappeared during the dictatorship, it not only returns the mortal remains to the relatives, but it also gradually answers the question that has kept, and still keeps, the determined Argentine people awake, a question that those responsible for the disappearances still refuse to answer: Where are the disappeared? These findings also show the modus operandi of the machinery of crime of the State as well as a web of silence and complicity, since —up until now— most of the remains identified have been found buried in unmarked common graves at municipal cemeteries, among other forms of concealment of bodies.

Since forced disappearance is an offence that persists over time, the identification of disappeared persons closes a circle that, together with testimonies and documents, constitutes the plain Truth about the genocide. Thus, forensic genetics becomes a link between Memory and Justice.

In this respect, we should highlight the contribution made by science to the search for and the restitution of the identity of children appropriated under State terrorism. In 1984, the Grandmothers of Plaza de Mayo, embracing Memory and in a constant search for Truth, managed to persuade scientists from the United States to agree to use genetic blood tests in order to determine the parentage of a child in the absence of his parents. The results of those tests, which make it possible to determine the biological parentage of children with 99.99% accuracy, were called "grandparenthood index". Thus, in 1987, the Argentine National DNA Data Bank was created with a view to storing genetic information so as to determine the relationship between victims of the dictatorship and their relatives.

Still, we must not forget that this has not always been the case, but is the result of a long struggle to instil democratic values and bring down the wall of impunity, thanks to which we can now talk about Justice in Argentina. The Mothers and Grandmothers of Plaza de Mayo played an exemplary role in that struggle and resistance. Likewise, the former President Néstor Kirchner supported that conviction to fight by making political decisions to face and pull down the wall of impunity. Once impunity began to crumble, it was possible to think about equal rights. Within the framework of the reconstruction of our country, the trials for crimes against humanity began to repair the damage caused by impunity; that is, the breach of the social pact of respect for and equality before the law.

Furthermore, the trials for crimes against humanity uncover the Truth and legitimize Memory, thus feeding the virtuous cycle of reparation. After a public oral trial —where the evidence consists of testimonies, documents, expert opinions, and statements by historians—, the judgment not only condemns or acquits the accused, but also expresses the values of the democratic State and conveys an educational message that reinforces and legitimizes Memory, thus making it a core element of History.

In order to go further along this path, ever since she took office as President of Argentina, Cristina Fernández de Kirchner has implemented public policies to restore the rights that were taken away from the people in previous decades. One of the policies on which emphasis should be placed here is once more to stress the key role of science in contributing to human development and to the sovereignty of the country. In this regard, some recent milestones have been reached: the creation of the Ministry of Science and Technology and the repatriation of more than a thousand scientists who had gone to other countries during the social crisis that hit the country in 2001-2002. This series of reparations have strengthened and improved democracy, and the expansion of rights has allowed us to recover the ideals of social justice.

The stress on the historical path of Memory, Truth and Justice is aimed at raising awareness of the need for a commitment by civil society and the State, because although we have been able to counter decades of impunity and make progress in recent years, we cannot allow any steps backwards with respect to the achievements made. In this regard, theoretical contributions, scientific participation, respect for human rights, and the constant expansion and strengthening of rights are fundamental in order to continue to build every day an increasingly democratic, inclusive and egalitarian culture.

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Secretary of Human Rights
Ministry of Justice and Human Rights of Argentina

PREFACE

The tragedy of the missing (the term “missing” is used to represent individuals, both dead and alive, whose fate/whereabouts is unknown to their families/communities, as a result of repressive dictatorial regimes, armed conflict, other situations of violence and catastrophes) can also have a severe psycho-social impact on the families of the missing and their communities and may constitute a barrier to the peace-building efforts following from conflict. In many cases the circumstances around the missing persons may amount to serious violations of international humanitarian law and human rights law.

The reality is that in many cases the individuals have been killed, and only through identification of the remains can closure be made possible for their families/communities. However, in some situations the missing person(s) may be alive and their identification is required for restoring family links. This situation most commonly impacts on young children separated from their parents; examples of this are well documented in Argentina where several children of individuals that had been killed and disappeared, were illegally adopted, and only through forensic investigations, including genetic testing, could their identities and family links be re-established.

Forensic genetics is playing an increasingly important role in identification of the missing persons. However, in many scenarios problems can be faced when attempting to implement identification programs. These problems can be legal, including issues around data protection and consent; and technical, including, the recovery of remains, extraction of DNA, collection of reference samples, the matching of the DNA profiles and quality control and assurance. The challenges faced in the search for the missing, alive or dead, are exacerbated by the scale of the identification programs and often the multiplicity of participating agencies and countries.

Since the early 1980s, Argentina has pioneered the use of forensic sciences, including forensic genetics, for the search of the missing. In response to problems repeatedly faced by countries and agencies involved in the identification of the missing, the Ministry of Foreign Affairs and Worship (MREC) of Argentina, proposed that the UN support general recommendations on best practices in the use of forensic genetics to establish the identity of the missing, dead or alive: this initiative was adopted in Resolutions A/HRC/RES/10/26 and A/HRC/RES/15/5 of the UN Human Rights Council (2009 and 2010 respectively).

The Resolutions welcomed the use of forensic genetics and encouraged states to consider using forensic genetics to contribute to the identification of remains and identify persons separated from their families. One aspect of the Resolutions was a request to the UN Office of the High Commissioner of Human Rights to consider drafting a manual on best practices for the use of forensic genetics for human rights and humanitarian investigations.

With the support of the International Committee of the Red Cross (ICRC), the MREC has coordinated experts, both legal and forensic, within Argentina (including the Argentine Forensic Anthropology Team (EAAF) and the Grandmothers of Plaza Mayo), Latin America, Spain and Portugal; and then worldwide, to draft a set of guidelines which aim at helping fill a gap in international best practices to guide the use of forensic genetics in such sensitive investigations.

Some important standards and guidelines that deal with forensic genetics in general and the identification of human remains in particular are already published[3]. However, issues experienced when dealing with the missing from situations involving abuses and violations of international humanitarian and human rights law are not fully covered by existing guidelines, in particular all applicable universal ethical and legal considerations.

These guidelines will be made available and their use promoted to national agencies, regional intergovernmental organisations, such as the MERCOSUR in South America, and the UN itself. They capture the experience of several agencies that have extensive experience of undertaking such sensitive investigations, particularly from South and Central America, where the problem of the missing is widespread. It is hoped that the guidelines will provide a valuable reference for forensic identification programs framed under international human rights and international humanitarian law provisions to help resolve the tragic legacy of armed conflicts and of grave violations of human rights and humanitarian law.

3 International Committee of the Red Cross (ICRC), Missing People, DNA Analysis and Identification of Human Remains: A Guide to Best Practice in Armed Conflict and Other Situations of Armed Violence, 2nd ed., ICRC, Geneva 2009. American Association of Blood Banks (AABB), Guidelines for Mass Fatality DNA Identification Operations, AABB, Bethesda, MD, 2010.
NNNational Institute of Justice (NIJ), Lessons Learned from 9/11: DNA Identification in Mass Fatality Incidents, NIJ, Washington, DC, 2006. M. Prinz, A. Carracedo, W.R. Mayr, N. Morling, T.J. Parsons, A. Sajantila, et al., DNA Commission of International SAociety for Forensic Genetics (ISFG): recommendations regarding the role of forensic genetics for disaster victim identification (DVI), Forensic Sci. Int. Genet. 1 (2007) 3-12.

INTRODUCTION

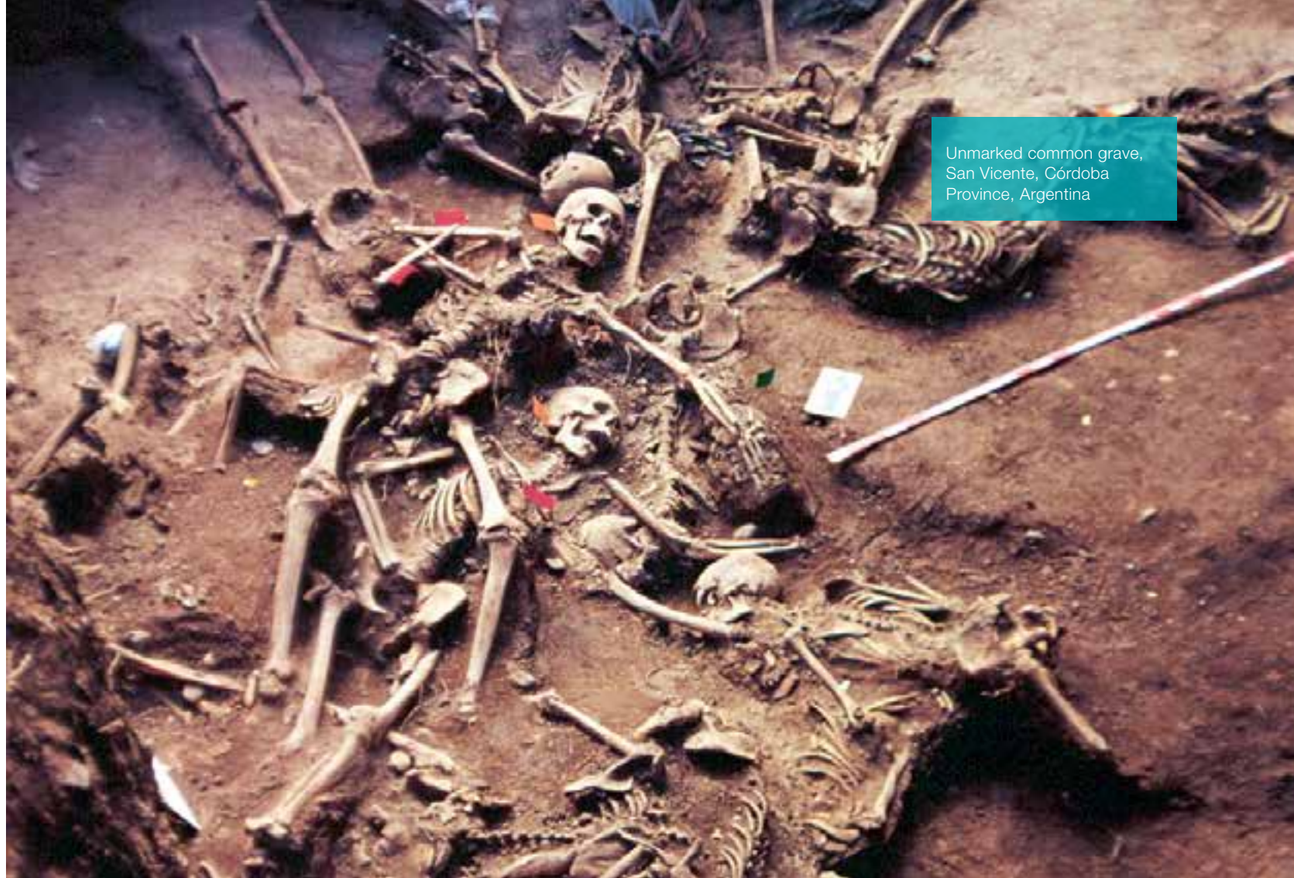
A. Use of forensic genetics as a tool for identifying victims of serious human rights and international humanitarian law violations

1.
Societies emerging from a period of serious human rights and international humanitarian law violations face major challenges in the investigation of the crimes committed, particularly when they include enforced disappearances. Forensic genetics, together with other scientific disciplines – medicine, odontology, anthropology and fingerprinting – can make an effective contribution to establishing the truth about what happened and bringing the perpetrators to justice, by identifying human remains and living persons. Each individual, except for monozygotic twins, has a unique DNA (deoxyribonucleic acid) profile, which is identical in all the body's tissues. In the case of human remains, the samples collected as part of the investigation are analysed to obtain the DNA profile of the remains which is then compared against the profile obtained from biological samples that originate from belongings of the missing person or from biological relatives, thus identifying the victims. Forensic genetics can contribute to the identification of victims, even in large-scale numbers.

2.
Forensic genetics has also been used to identify and restore the identity of living persons in cases involving the abduction of children whose parents have been subjected to forced disappearance, including babies born in captivity, and the suppression of identity during detention through the destruction of ID documents, displacement or exile.

3.
Some of the experiences described in this Guide show that this discipline can be of great use in documenting human rights violations, providing evidence in national and international proceedings and ensuring the right of victims of human rights violations and natural disasters, as well as society as a whole, to the truth.

4.
It is important to note that human identification is a process requiring contributions from a number of scientific disciplines working together in a coordinated and integrated manner. Although forensic genetics plays an important role in the process, the results obtained must be reviewed in the light of the findings of other disciplines, such as anthropology, medicine and dentistry.



Unmarked common grave, San Vicente, Córdoba Province, Argentina

B. Objectives and scope

5.
Through the identification of individuals, forensic genetics provides findings that serve as a basis for legal and historical inquiries to establish the facts about individual and collective acts of violence and for the pursuit of truth and justice. This discipline must therefore be used in accordance with the international obligations of States and scientific and ethical principles.

6.
The purpose of this Guide is to provide guidelines on the application of forensic genetics and the creation and management of genetic data banks as part of investigations to identify victims of human rights and international humanitarian law violations. These guidelines are provided in the light of the duty that States have to investigate human rights violations and identify the victims, while ensuring respect for the dignity and privacy of the people involved in the identification process. The guide is intended for people involved in or responsible for such processes, particularly members of the justice system, legislators, State officials, human rights defenders and the families of victims.

7.
The Guide first outlines the main international developments in the field of bioethics applicable to genetic identification. The second part traces the key legal developments in relation to the identification of victims of human rights and international humanitarian law violations, and the third part puts forward a series of recommendations on the organization and operation of biobanks and genetic data banks for DNA-based identification. The final section provides an overview of the scientific, technical and methodological requirements for sample collection, numbering and storage; DNA extraction and analysis; DNA profile development and management; and the creation and management of genetic data banks.

8.
It should be noted that this Guide is not intended to provide an exhaustive and detailed review of forensic genetics, but is rather a compendium of good practices, drawn from the experiences of various countries where this discipline has been used to establish the truth about human rights violations and bring the perpetrators to justice through the identification of human remains and living persons.

C. Ethical issues relating to the use of forensic genetics

9.
The international community has adopted a number of instruments which establish standards for the use of human genetics in compliance with bioethics. Such instruments include the Universal Declaration on the Human Genome and Human Rights, the International Declaration on Human Genetic Data and the Universal Declaration on Bioethics and Human Rights.

10.
The Universal Declaration on the Human Genome and Human Rights, adopted by UNESCO on 11 November 1997 and approved by the United Nations General Assembly in 1998, establishes that "Everyone has a right to respect for their dignity and for their rights regardless of their genetic characteristics" (article 2 a).

11.
The UNESCO International Bioethics Committee led a process of reflection lasting several years, which culminated in the adoption of the International Declaration on Human Genetic Data at the 32nd meeting of UNESCO's General Conference on 16 October 2003. The Declaration establishes principles to guide States in formulating legislation and public policy, with a view to ensuring respect for human dignity and the protection of human rights and fundamental freedoms in relation to the collection, processing, use and storage of human genetic data, in keeping with the requirements of equality, justice and solidarity, while giving due consideration to freedom of thought and expression, including freedom of research (article 1).

12.
Article 5 of the International Declaration on Human Genetic Data establishes that human genetic data may be collected, processed, used and stored for forensic medicine and civil, criminal and other legal proceedings and for any other purpose consistent with the Universal Declaration on the Human Genome and Human Rights and international human rights law.

13.
Article 12 of the International Declaration on Human Genetic Data establishes that the collection of human genetic data for the purposes of forensic medicine or in civil, criminal and other legal proceedings, including parentage testing and the collection of biological samples, *in vivo* or post mortem, can only be carried out in accordance with domestic law consistent with international human rights law.

14.
The Universal Declaration on Bioethics and Human Rights, the third instrument drafted and approved by UNESCO on the subject of bioethics, was adopted by acclamation at the 33rd meeting of the General Conference in 2005. It addresses ethical issues related to medicine, life sciences and associated technologies as applied to human beings, taking into account their social, legal and environmental dimensions. It provides a framework of principles and procedures to guide States in the formulation of their policies, legislation and codes of ethics.

15.
The first of the principles established in the Declaration is full respect for human dignity, human rights and fundamental freedoms (article 3).

Unmarked common grave, San Vicente, Córdoba Province, Argentina

16.
The Declaration also establishes the principle of respect for human vulnerability and personal integrity (article 8), the principle of privacy and confidentiality (article 9) and the principle of non-discrimination and non-stigmatization (article 11).

17.
Article 10 establishes the fundamental equality of all human beings in dignity, requiring that they be treated justly and equitably.

18.
Articles 5 and 6 of the Declaration establish the obligation to respect the autonomy of persons in making decisions and to obtain their consent, stating that exceptions to this principle should be made only in accordance with ethical and legal standards adopted by States, consistent with the principles and provisions set out in the Declaration and international human rights law.

19.
Article 27 establishes that limitations on the application of the principles enunciated in the Declaration should be made by law, consistent with international human rights law.

20.
Article 21 of the Declaration, which deals with international cooperation, urges States to promote international dissemination of scientific information and encourage the free flow and sharing of scientific and technological knowledge (see also articles 24 and 15).

21.
In addition to the general bioethics standards established in the instruments adopted by UNESCO, States also have obligations under international law regarding the use of forensic genetics in the identification of victims of serious human rights violations and people who were abducted and deprived of their identity.

II. RULES OF INTERNATIONAL LAW REGARDING THE IDENTIFICATION OF VICTIMS OF SERIOUS VIOLATIONS OF HUMAN RIGHTS, INTERNATIONAL HUMANITARIAN LAW AND THE RIGHT TO IDENTITY

22.
The main human rights and international humanitarian law treaties as well as their authorized interpretation, address the question of the duty that States have to facilitate the means necessary to effectively investigate serious human rights violations. Fulfilling this duty may require efforts to trace and identify victims of forced disappearances, to re-establish the true identity of victims of abduction and deprivation of identity or to identify people who have gone missing in international and internal armed conflicts. Such processes are essential to ensuring the right of victims and society as a whole to the truth.

23.
Forensic genetics can contribute to the identification of persons in various contexts: the implementation of crime prevention and prosecution policies; the determination of biological relationships in civil proceedings; the identification of the remains of people who are missing as a result of armed conflicts or natural disasters; the identification of victims of human rights and international humanitarian law violations; and the re-establishment of the true identity of victims of abduction and suppression of identity. Forensic genetics is a tool that States can use to fulfil their obligation under international law to guarantee the right to justice, identity and the truth and thereby ensure the right to human dignity and to make life choices. At the same time, the information contained in the DNA of each individual is highly sensitive and requires legal protection to ensure the right to privacy and prevent discrimination based on genetic characteristics.

24.
The main rules concerning the duty to investigate serious human rights and international humanitarian law violations, the right to the truth, the right to identity and the safeguards required to protect people's privacy and autonomy are described below.

A. Duty of States to investigate violations of human rights and international humanitarian law

25.
A State's obligation to investigate acts affecting the rights protected by human rights instruments derives from the general obligation established in them to ensure those rights, combined with the substantive right that must be safeguarded, protected or ensured, and the right to the protection of the law and due process safeguards. In light of this duty, whenever State authorities have knowledge of acts affecting the rights protected under these treaties, which are offences subject to public prosecution, they must promptly initiate a thorough, impartial and effective investigation using all legal means at their disposal to establish the truth and prosecute and punish the perpetrators.

26.
In the Universal Declaration of Human Rights[4], the duty of States to investigate acts affecting the rights established in it derives from the right of individuals to an effective remedy for acts violating their fundamental rights (article 8), together with the substantive right that must be protected.

27.
In the International Covenant on Civil and Political Rights[5], the duty to investigate human rights violations derives from the general obligation to guarantee the rights recognized by article 2.1 and 2.2, together with the duty to ensure the right to the protection of the law provided for in article 2.3 and 2.4 and the duty to ensure due process of law, as established in article 14. In General Comment No. 31[6] , the United Nations Human Rights Committee clarifies the content and scope of the obligations binding on the States Parties pursuant to article 2 of the International Covenant on Civil and Political Rights. In this regard, it stresses the general obligation imposed on States Parties to respect and ensure the rights recognized by the Covenant. Paragraphs 6 and 7 of the General Comment state that the legal obligation under article 2, paragraph 1, is both negative and positive in nature. This means that States Parties must refrain from violating the rights recognized by the Covenant and must also adopt legislative, judicial, administrative and other appropriate measures in order to fulfil their legal obligations, which include the obligation to provide accessible and effective remedies (article 2, paragraph 3). With regard to this point, paragraph 15 of the Committee's General Comment establishes that "[a]dministrative mechanisms are particularly required to give effect to the general obligation to investigate allegations of violations promptly, thoroughly and effectively through independent and impartial bodies (...) A failure by a

4 Adopted by the United Nations General Assembly in 1948.

5 The International Covenant on Civil and Political Rights, adopted and opened for signature and ratification by United Nations General Assembly resolution 2200 A (XXI) of 16 December 1966, entered into force on 23 March 1976, in accordance with article 49.

6 United Nations Human Rights Committee, General Comment No. 31 "Nature of the general legal obligation imposed on States Parties to the Covenant" adopted on 29 March 2004 at the Eightieth Session (CCPR/C/21/Rev.1/Add.13).

State Party to investigate allegations of violations could in and of itself give rise to a separate breach of the Covenant".

28.
The obligation to investigate human rights violations also derives from the right to the protection of the law recognized in other international instruments aimed at protecting the human rights of particularly vulnerable groups of people. For example, article 2 (c) of the Convention on the Elimination of All Forms of Discrimination against Women[7] establishes the general obligation to ensure the right of women to the protection of the law. Article 6 of the International Convention on the Elimination of All Forms of Racial Discrimination[8] establishes the duty to ensure effective protection and remedies through the competent tribunals and other State institutions. Article 3 of the International Convention for the Protection of All Persons from Enforced Disappearance establishes that each State Party must take appropriate measures to investigate cases of enforced disappearance, and article 10.2 establishes that a State Party in whose territory an offence of enforced disappearance is suspected to have been committed must immediately carry out a preliminary inquiry to establish the facts. The duty to investigate and ensure the right to the protection of the law is established in articles 12 and 17.2 (f).

29.
In the United Nations Declaration on the Protection of all Persons from Enforced Disappearance[9], the obligation to investigate enforced disappearances derives from the right to the protection of the law established in articles 9, 10 and 13. The letter of the Declaration goes further, as it not only ensures the right to effective remedies to establish the facts about what has happened, but also stresses the importance of these remedies as a means of determining the whereabouts of the missing person. Article 9.1 provides that "[t]he right to a prompt and effective judicial remedy as a means of determining the whereabouts or state of health of persons deprived of their liberty and/or identifying the authority ordering or carrying out the deprivation of liberty is required to prevent enforced disappearances under all circumstances". Article 13.1 provides that "[e]ach State shall ensure that any person having knowledge or a legitimate interest who alleges that a person has been subjected to enforced disappearance has the right to complain to a competent and independent State authority and to have that complaint promptly, thoroughly and impartially investigated by that authority. Whenever there are reasonable grounds to believe that an enforced disappearance has been committed, the State shall promptly refer the matter to that authority for such an investigation, even if there has

7 The Convention on the Elimination of All Forms of Discrimination against Women, adopted and opened for signature and ratification by United Nations General Assembly resolution 34/180 of 18 December 1979, entered into force on 3 September 1981, in accordance with article 27 (1).

8 The International Convention on the Elimination of All Forms of Racial Discrimination, adopted and opened for signature and ratification by United Nations General Assembly resolution 2106 (XX) of 21 December 1965, entered into force on 4 January 1969, in accordance with Article 19.

9 The Declaration on the Protection of all Persons from Enforced Disappearance was adopted by General Assembly resolution 47/133 of 18 December 1992.

Burial of a disappeared person whose remains have been identified

been no formal complaint. No measure shall be taken to curtail or impede the investigation".

30.
Under the Inter-American System for the protection of human rights[10], the Inter-American Commission and Court have interpreted the scope of the obligation to investigate human rights violations established by the American Convention[11] and other instruments that establish obligations binding on the member countries of the Organization of American States (OAS) in relation to human rights[12].

10 In April 1948, the OAS member States adopted the American Declaration of the Rights and Duties of Man, the first international instrument to catalogue fundamental human rights, including the right to life, personal security and the protection of the law. In 1959, the OAS member States set up the Inter-American Commission on Human Rights, formed by seven independent experts with a mandate to monitor the human rights situation in the region. In 1961, the Commission began to conduct on-site visits and adopt reports in fulfilment of its mandate. In 1965, it began to process petitions from alleged victims of violations of the rights recognized by the American Declaration.

11 In 1969, the OAS member States adopted the American Convention on Human Rights. This treaty, which has been ratified by the majority of OAS member States, increases the number of human rights originally listed in the American Declaration and establishes oversight and enforcement mechanisms operated by the pre-existing Inter-American Commission on Human Rights and the Inter-American Court of Human Rights, which was established in 1979.

12 After the American Convention had come into force in 1978, the OAS member States adopted a series of treaties concerning areas of particular interest for the protection of human rights in the region, including the Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights (Protocol of San Salvador), the Protocol to the American Convention on Human Rights to Abolish the Death Penalty, the Inter-American Convention to Prevent and Punish Torture, the Inter-American Convention on Forced Disappearance of Persons and the Inter-American Convention on the Prevention, Punishment and Eradication of Violence against Women (Convention of Belém do Pará). See Inter-American Commission on Human Rights, *Basic Documents pertaining to Human Rights in the Inter-American System*, OEA/Ser.L/V/I.4, rev. 13, 30 June 2010.

31.
Article 1.1 of the American Convention charges the States Parties with the fundamental duty to respect and ensure the rights recognized by the Convention. Any impairment of those human rights which can be attributed under the rules of international law to the action or omission of any public authority constitutes an act imputable to the State, which must assume international responsibility in the terms provided by the Convention and international law in general.

32.
The right to life and personal security are given particular importance in the American Convention. Pursuant to article 27.2, they form part of the core of rights from which no derogation is permitted, as they are established as rights that cannot be suspended even in the event of war, public danger or other threats. The State's obligation is not confined to refraining from violating these rights; it is also bound to adopt positive measures in response to the specific needs of those who require protection on account of their personal condition or the particular situation in which they find themselves[13]. States have the obligation to ensure the conditions required to prevent violations of the right to life. Compliance with article 4 of the American Convention, taken together with article 1.1, not only presupposes the principle that no human being should be arbitrarily deprived of his or her life, but also requires States to take all appropriate measures to protect and preserve the right to life pursuant to their duty to ensure the full and free exercise of the rights of all those under their jurisdiction[14].

13 Inter-American Court of Human Rights, *Case of Baldeón García v. Peru*, Judgement of 6 April 2006, Series C, no. 147, paragraph 81; *Case of the Sawhoyamaxa Indigenous Community v. Paraguay*, Judgement of 29 March 2006, Series C, no. 146, paragraph 154; and *Case of the Pueblo Bello Massacre v. Colombia*, Judgement of 31 January 2006, Series C, no. 140, paragraph 111. http://www.cidh.org/countryrep/Seguridad/seguridadiv.sp.htm - _ftn40

33.
Under the American Convention, States have a primary duty to ensure the right to life by establishing effective provisions under criminal law to deter people from committing crimes against others, backed by the machinery of enforcement to prevent, pursue and punish violations of those provisions. In some circumstances, this extends to a positive obligation requiring the authorities to take preventive operational measures to protect individuals or groups of individuals whose lives are endangered by the criminal acts of others. For such a positive obligation to arise, it must be established that the authorities knew or ought to have known, at the time that the crime was committed, of the existence of a real and immediate risk to the life of an identified individual or group of individuals from the criminal activities of others and failed to do all that could reasonably be expected of them to avoid the danger. In cases of gross and systematic human rights violations, the State's obligation to adopt positive preventive and protective measures pursuant to the obligations established under Article 1.1 of the American Convention[15] becomes particularly compelling.

14 Inter-American Commission on Human Rights, *Report on citizen security and human rights*, OEA/Ser.L/V/II, Doc. 57, 31 December 2009, paragraph 43.

15 Inter-American Court of Human Rights, *Case of Velásquez Rodríguez v. Honduras*, Judgement of 29 July 1988, Series C, no. 4, paragraph 177; *Case of the Pueblo Bello Massacre v. Colombia*, Judgement of 31 January 2006, Series C, no. 140, paragraph 142; *Case of Heliodoro Portugal v. Panama*, Preliminary objections, Merits, Reparations and Costs, Judgement of 12 August 2008, Series C, no. 186, paragraph 115; and *Case of Perozo et al. v. Peru*, Preliminary Objections, Merits, Reparations and Costs, Judgement of 28 January 2009, Series C, no. 195, paragraph 298.

34.
A State's obligation to investigate violations of the rights protected under the American Convention derives from the general obligation established in article 1.1 to ensure those rights, taken together with the substantive right that must be safeguarded, protected and ensured and the due process safeguards and protection of the law established in articles 8 and 25. In light of this duty, whenever State authorities have knowledge of acts affecting the rights protected under the American Convention, which are offences subject to public prosecution, they must promptly instigate a thorough, impartial and effective investigation using all legal means at their disposal to establish the truth and prosecute and punish the perpetrators. During the course of the investigation and the legal proceedings, the victims or their families must be given ample opportunity to participate and be heard in the process undertaken to establish the facts and punish those responsible and the chance to pursue reparation. Such investigations must be undertaken by the State as an inherent legal obligation and not merely in response to private concerns, leaving it up to the victims or their families to take the initiative in bringing legal action and relying on evidence from private sources, without taking effective steps itself to determine the truth[16].

16 Inter-American Court of Human Rights, *Case of Barrios Altos v. Peru*, Judgement

35.
A State's duty to investigate acts affecting the rights protected under the American Convention applies regardless of who the alleged perpetrator may be. When the alleged perpetrator is an individual, the failure of the State to thoroughly investigate the charges would compromise its international responsibility as protector. When agents of the State might be involved, the State has a special obligation to establish the facts and bring those responsible to justice. Lastly, in cases involving serious human rights violations, such as torture, extrajudicial, summary and arbitrary executions and enforced disappearance, the Inter-American Court has established that amnesties, statutes of limitation and release from responsibility provisions, which seek to prevent the investigation, prosecution and punishment of those responsible, are inadmissible.

36.
In the case of crimes against humanity, the Inter-American Court has established that they violate non-derogable rights recognized by the American Convention and that these violations must not be left unpunished. The Court has also made it clear that such investigations must be conducted using all legal means available and seek to establish the truth and investigate, pursue, arrest, prosecute and punish all perpetrators and instigators, particularly when agents of the State are or might be involved. In such cases, the passing of amnesty laws leaves victims defenceless and perpetuates impunity for crimes against humanity. They are therefore clearly inconsistent with the spirit and letter of the American Convention[17].

37.
In cases in which a violation of the right to life also involves concealment of the body, the duty to investigate the crime, bring those responsible to justice and make reparations includes an obligation to identify the victim. In cases in which people are deprived of their identity, the duty to investigate, prosecute and make reparations involves re-establishing their identity. The scope of these two obligations is explained below.

1. Obligations of States regarding search, localization and identification of victims

38.
In the context of armed conflict, conventional and customary law and jurisprudence indicate that a State's obligation to identify victims is established and regulated by international humanitarian law[18].

39.
The Geneva Conventions of 1949 and their Additional Protocols regulate the obligations of the parties in relation to missing persons. Pursuant to these rules, the parties to a conflict are bound to adopt the required legislative and administrative measures to prevent people from going missing, identify people missing as a result of an armed conflict and investigate the circumstances of such disappearances. Customary international humanitarian law (Rule 98) establishes that enforced disappearance is prohibited in both international and non-international armed conflicts[19]. This prohibition encompasses a duty to investigate cases of alleged enforced disappearance.

40.
Article 32 of Additional Protocol I recognizes the right of families to know the fate of missing loved ones, and article 33 of the Protocol establishes that all the parties to a conflict must take all feasible measures to ascertain the whereabouts of people reported missing. There are also provisions on preventing disappearances in articles 136 and 141 of the Fourth Geneva Convention. Articles 15 and 18 of the Second Geneva Convention, article 16 of the Fourth Geneva Convention, and article 34 of Additional Protocol I require States to adopt visible measures to search for, recover and identify the dead and keep a record of the exact location and markings of gravesites and details of internees.

41.
Customary international humanitarian law (Rule 117) establishes that the parties to a conflict, whether international or non-international, must take all feasible measures to account for persons reported missing as a result of armed conflict and provide their family members with any information it has on their fate[20].

42.
Article 38 of the Convention on the Rights of the Child requires States to respect the rules of international humanitarian law concerning children applicable in armed conflicts. These rules include provisions on identifying the victims of humanitarian law violations.

43.
Within the framework of international human rights law, the International Convention for the Protection of All Persons from Enforced Disappearance imposes the duty to identify victims of such crimes. In this regard, article 15 establishes that "States Parties shall cooperate with each other and shall afford one another the greatest measure of mutual assistance with a view to assisting victims of enforced disappearance, and in searching for, locating and releasing disappeared persons and, in the event of death, in exhuming and identifying them and returning their remains".

44.
Current international standards on the processing of crime scenes in cases involving human rights violations stipulate that it is necessary to close off the area around the body and restrict access to it; collect physical evidence and samples of blood, hair, fibres and any other relevant items; examine the area for

of 14 March 2001, Series C, no. 75, paragraph 41; *Case of Almonacid Arellano et al. v. Chile*, Judgement of 26 September 2006, Series C, no. 154, paragraph 112; and *Case of the Ituango Massacres v. Colombia*, Preliminary Objections, Merits, Reparations and Costs, Judgement of 1 July 2006, Series C, no. 148, paragraph 402.

17 Inter-American Commission on Human Rights, *Report on Citizen Security and Human Rights*, OEA/Ser.L/V/II., Doc. 57, 31 December 2009, paragraph 46.

18 International humanitarian law is the set of rules of conventional and customary law applicable in international and non-international armed conflict situations, aimed at regulating methods and means of warfare and protecting those who are not, or no longer, taking an active part in hostilities.

19 See International Committee of the Red Cross, *Customary International Humanitarian Law, Volume I*, Chapter 32.

20 See International Committee of the Red Cross, *Customary International Humanitarian Law, Volume I*, Chapter 36.

shoeprints and any other impressions of an evidentiary nature; make a report detailing any observations at the scene; maintain the chain of custody of each item of forensic evidence; and keep a precise written record, supplemented, as appropriate, by photographs and other graphic elements, to document the chronological history of each item of evidence as it passes through the hands of the different investigators involved in the case[21]. The chain of custody can extend beyond the trial, sentencing and conviction of the accused. An exception to this is the remains of the victim once they have been positively identified. They can be returned to their families for burial, on the condition that they are not cremated, in case they are needed in new investigations undertaken at a later date[22].

45.
Under the Inter-American system for the protection of human rights, standards have been developed concerning the processing and management of information obtained for the purpose of identification in order to provide guidance to States in this field. The Inter-American Court of Human Rights has ordered the creation and updating of databases containing the personal details and any other relevant information available on victims of enforced disappearance; the creation and updating of databases containing all necessary personal information, in particular genetic data and biological samples collected from biological relatives of missing persons with their consent or by court order and stored by the authorities for the sole purpose of identifying missing persons; and the creation and updating of databases containing genetic data and biological samples collected from unidentified remains which could belong to persons who have disappeared or been killed in a given place. The Court has also established that States are required to protect all personal information stored in these databases[23].

46.
Resolutions 57/207, 59/189, 61/155 and 63/183 concerning missing persons adopted by the United Nations General Assembly stress the importance of forensic work in identifying missing persons and recognize the progress made in this field thanks to developments in DNA analysis. In the same vein, the Report of the Secretary-General of the United Nations[24] produced pursuant to above-mentioned resolution 61/155 establishes that in cases where missing persons are believed to be dead, the recovery, identification and proper and dignified management of their remains are essential. The report therefore concludes that the effective search for and identification of missing persons, including the use of forensic genetics, can make a significant contribution to efforts to identify such victims.

21 United Nations Manual on the effective prevention and investigation of extra-legal, arbitrary and summary executions (Doc. E/ST/CSDHA/.12), 1991.

22 Inter-American Court of Human Rights, *Case of González et al.* ("Cotton Field"), Preliminary Objection, Merits, Reparations, and Costs, Judgement of 16 November 2009, Series C, no. 205, paragraph 305, statement made before notary public by expert witness Clyde Snow on 17 April 2009.

23 Inter-American Court of Human Rights, *Case of González et al*. ("Cotton Field"), Preliminary Objection, Merits, Reparations, and Costs, Judgement of 16 November 2009, Series C, no. 205, paragraph 512. See also *Case of Molina Theissen v. Guatemala*, Reparations and Costs, Judgement of 3 July 2004, Series C, no. 108, paragraph 91; *Case of the Serrano Cruz sisters v. El Salvador*, paragraph 193; and *Case of Servellón García et al. v. Honduras*, paragraph 203.

24 Report of the Secretary-General of the United Nations on missing persons (A/63/299), 18 August 2008.

47.
The General Comment on the Right to the Truth[25] submitted by the United Nations Working Group on Enforced or Involuntary Disappearances[26] in 2010, highlights the importance of identifying victims of enforced disappearance, including through DNA analysis. On this subject, in paragraph 6 of the General Comment, the Working Group establishes that States ought to take the necessary steps to use forensic expertise and scientific methods to identify victims to the maximum of their available resources, including international cooperation.

48.
In its reports on the right to the truth, forensic genetics and human rights and the obligation of States to investigate serious violations of human rights and the use of forensic genetics[27], the Office of the United Nations High Commissioner for Human Rights concluded that the use of forensic genetics and the voluntary establishment of data banks play a crucial role in identifying the victims of human rights and international humanitarian law violations.

49.
The reports also highlight the need to raise the awareness of States about forensic genetics as a means of complying with their human rights obligations, in particular, but not exclusively, in relation to missing persons, and stress the requirement that such investigations be based on objective scientific methods, ensuring respect for human dignity and the protection of human rights and fundamental freedoms in the collection, processing and use of genetic data.

2. Obligation of States in cases of deprivation of identity

50.
In the face of serious violations of human rights and international humanitarian law involving cases of people who were illegally deprived of their identity, the obligation of the State under international law includes restoring their true identity. It should be noted that this obligation remains binding regardless of the time that has passed since the person's identity was suppressed, as it is considered a continuing crime until such time as the truth is established and the true identity of that person is restored.

51.
This obligation derives from the general obligation to ensure human rights established in international human rights

25 http://www.ohchr.org/Documents/Issues/Disappearances/GC-right_to_the_truth.pdf

26 Since it was set up in 1980, the Working Group on Enforced or Involuntary Disappearances has been examining reports of enforced disappearances and endeavours to contribute to determining the whereabouts of victims. The Working Group also prepares reports to provide input for the development of international standards in this field.

27 Reports of the Office of the United Nations High Commissioner for Human Rights on "The right to the truth and on forensic genetics and human rights", 24 August 2010 (A/HRC/15/26 of 2010) and on "The obligation of States to investigate serious violations of human rights and the use of forensic genetics", 4 July 2011 (A/HRC/18/25).

Preparing the remains of an identified disappeared for his burial in a coffin painted by his family

instruments. In this case, the State has the duty to ensure the effective exercise of the right to identity which, in its individual dimension[28], each person has from birth and includes the right to have a name for social, legal and family recognition, to acquire a nationality, to have a family and be raised in it and to preserve these conditions in the future. The obligation to ensure these rights also includes re-establishing the identity of any person who has been illegally deprived of some or all of the elements of his or her identity.

52.
In this regard, the right to identity is enshrined in various international human rights treaties. Article 24 of the International Covenant on Civil and Political Rights establishes that "1. Every child shall have, without any discrimination as to race, colour, sex, language, religion, national or social origin, property or birth, the right to such measures of protection as are required by his status as a minor, on the part of his family, society and the State. 2. Every child shall be registered immediately after birth and shall have a name. 3. Every child has the right to acquire a nationality". The United Nations Human Rights Committee adopted a view on the content and scope of the right to identity recognized in the International Covenant on Civil and Political Rights in the case of a minor who was deprived of her identity in the context of systematic human rights violations. In the view it adopted on this occasion, the Committee encouraged the State party to "persevere in its efforts to investigate the disappearance of children, determine their true identity, issue to them identity papers and passports under their real names, and grant appropriate redress to them and their families in an expeditious manner"[29].

53.
The Convention on the Rights of the Child[30] specifies the content of the right to identity and the general obligations of the State. Article 7 reflects the above-mentioned provision from the International Covenant on Civil and Political Rights, and article 8 provides that "States Parties undertake to respect the right of the child to preserve his or her identity, including nationality, name and family relations as recognized by law without unlawful interference".

54.
This same article concerning the obligations of States in relation to the right to identity, stipulates that "[w]here a child is illegally deprived of some or all of the elements of his or her identity, States Parties shall provide appropriate assistance and protection, with a view to re-establishing speedily his or her identity". In connection with this obligation, international human rights law has progressed towards the adoption of more specific provisions to ensure protection, mainly in response to historical processes characterized by large-scale and systematic human

28 The right to identity also has a collective dimension, that is, the identity of peoples. On this subject, see the Convention concerning Indigenous and Tribal Peoples in Independent Countries (Convention 169), International Labour Organization.

29 Human Rights Committee, *Case of Darwinia Rosa Mónaco de Gallicchio and Ximena Vicario v. Argentina*, 27 April 1995.

30 The Convention on the Rights of the Child was adopted and opened for signature, ratification and accession by the United Nations General Assembly on 20 November 1989 and entered into force on 2 September 1990.

rights violations, including enforced disappearances and the abduction of children of parents subjected to enforced disappearance or of children born during their mother's enforced disappearance and acts depriving them of their true identity.

55.
In this context, the international community adopted the Declaration on the Protection of all Persons from Enforced Disappearance[31], which establishes the obligation of States to prevent and suppress the abduction of children of parents subjected to enforced disappearance and of children born during their mother's enforced disappearance. It requires them to make every endeavour to search for and identify such children and to return them to their families of origin. It also provides that the abduction of children in such circumstances and the act of altering or suppressing documents attesting to their true identity, constitute an extremely serious offence, which must be punished as such[32].

56.
Within the framework of the OAS, article XII of the Inter-American Convention on Forced Disappearance of Persons[33] provides that "States Parties shall give each other mutual assistance in the search for, identification, location, and return of minors who have been removed to another state or detained therein as a consequence of the forced disappearance of their parents or guardians".

57.
The provisions of the International Convention for the Protection of all Persons from Enforced Disappearance include the obligation of States to criminalize the wrongful removal of children and confirm the principle that they must be returned to their families of origin. On this subject, the Convention also includes an express reference to the right of children who have been subjected to enforced disappearance to have their identity re-established[34] .

31 The Declaration on the Protection of all Persons from Enforced Disappearance was adopted on 18 December 1992.

32 See article 20 of the Declaration on the Protection of all Persons from Enforced Disappearance.

33 The Inter-American Convention on Forced Disappearance of Persons was adopted in Belem do Pará, Brazil, on 9 June 1994 and came into force on 28 March 1996.

34 On this subject, see article 25 of the International Convention for the Protection of all Persons from Enforced Disappearance, which provides as follows: "1. Each State Party shall take the necessary measures to prevent and punish under its criminal law: (a) The wrongful removal of children who are subjected to enforced disappearance, children whose father, mother or legal guardian is subjected to enforced disappearance or children born during the captivity of a mother subjected to enforced disappearance; (b) The falsification, concealment or destruction of documents attesting to the true identity of the children referred to in subparagraph (a) above. 2. Each State Party shall take the necessary measures to search for and identify the children referred to in paragraph 1 (a) of this article and to return them to their families of origin, in accordance with legal procedures and applicable international agreements. 3. States Parties shall assist one another in searching for, identifying and locating the children referred to in paragraph 1 (a) of this article. 4. Given the need to protect the best interests of the children referred to in paragraph 1 (a) of this article and their right to preserve, or to have re-established, their identity, including their nationality, name and family relations as recognized by law, States Parties which recognize a system of adoption or other form of placement of children shall have legal procedures in place to review the adoption or placement procedure, and, where appropriate, to annul any adoption or placement of children that originated in an enforced disappearance. 5. In all cases, and in particular in all matters relating to this article, the best interests of the child shall be a primary consideration, and a child who is capable of forming his or her own views shall have the right to express those views freely, the views of the child being given due weight in accordance with the age and maturity of the child".

58.
Lastly, both the United Nations Commission on Human Rights and the Human Rights Council have adopted resolutions that underline the obligation of States to assist persons deprived of their identity by re-establishing their true identity and highlight the role of forensic science in this task. The Commission on Human Rights has adopted resolutions acknowledging that the use of forensic science can contribute to reuniting children forcibly separated from their parents with surviving family members[35].

59.
In 2009, the United Nations Human Rights Council adopted by consensus resolution 10/26 on forensic genetics and human rights, with a view to promoting the development of international standards to serve as a guide for the application of forensic genetics in identifying victims of serious violations of human rights and, in armed conflict situations, of international humanitarian law. This resolution stresses the importance of restoring the identity of people separated from their families, including those taken away from their families when they were children. In this regard, the resolution includes an operative provision in which the Human Rights Council encourages States to consider the use of forensic genetics to contribute to restoring the true identity of persons separated from their families in such circumstances.

35 On this subject , see resolutions 1998/36, 2000/32, 2003/33 and 2005/26 on Human Rights and Forensic Science of the United Nations Commission on Human Rights.

B. Right to the truth

60.
The right to the truth is defined under international law as the right of victims of serious violations of human rights and international humanitarian law and their families to know the truth regarding such violations, in particular the identity of the perpetrators, the causes and the facts of the violations, and the circumstances in which they occurred. In addition to this individual dimension, the right to the truth also has a collective dimension, associated with the right of society as a whole to know the truth about its past in order to build historical memory and preserve it for the future, particularly in transition processes following a period in history marked by a systematic pattern of human rights violations and the use of violence.

61.
The consensus of the international community on this point is reflected in the provisions of international humanitarian law[36] and international human rights law[37], and in case law decisions adopted by international courts[38] which have progressively clarified the content and scope of this right.

62.
The individual and collective right to the truth is one of the main elements in the fight against impunity and entails a series of obligations that States must fulfil to ensure effective enforcement. In this regard, it is important that States establish proper and effective mechanisms to enable society as a whole and the families of victims in particular to know the truth about serious violations. This is a duty of care and not a duty to achieve a specific result, which is not an alternative to the obligations that States have in these cases to ensure access to justice.

63.
In this context, the use of forensic science in general and forensic genetics in particular, is regarded as a useful tool which can be used by States to fulfil their obligations in terms of ensuring the effective exercise of the right to the truth in the context of human rights or international humanitarian law violations involving enforced disappearance and/or abduction and deprivation of identity[39]. In such cases, the obligation to ensure the effective exercise of the right to the truth involves the adoption of measures to identify the victims of violations and re-establish the true identity of people illegally deprived of their identity.

64.
On this subject, the International Convention for the Protection of All Persons from Enforced Disappearance, the first binding international human rights instrument to recognize the right to the truth, establishes that victims[40] of enforced disappearance have the right to know the truth about the circumstances of the enforced disappearance, the progress and findings of the investigation and the fate of the missing person. It establishes that each State Party must take appropriate steps to ensure this[41].

36 Article 32 of Additional Protocol I to the Geneva Conventions relating to the protection of victims of international armed conflicts recognizes the right of families to know the fate of missing relatives. In this regard, article 33 of the Protocol establishes that, as soon as circumstances permit, each party to the conflict must search for the persons who have been reported missing.

37 On this subject, see, for example, resolution 2005/66 of the United Nations Commission on Human Rights and resolutions 9/11 and 12/12 of the United Nations Human Rights Council on the right to the truth. See also the International Convention for the Protection of all Persons from Enforced Disappearance (Preamble and article 24). Within the framework of decisions by the treaty bodies of the international system for the protection of human rights, the United Nations Human Rights Committee established, in a case involving enforced disappearance, the right of the family of the victim to know what had happened to her and underlined the obligation of States to investigate the events to establish the truth and determine the whereabouts of the missing person (Decision of 21 July 1983, *Case of María del Carmen Almeida de Quintero v. Uruguay*, Communication no. 107/1981, paragraphs 14, 15 and 16).

38 On this subject, within the Inter-American System, the Inter-American Court of Human Rights recognized, in the *Case of Velásquez Rodríguez*, the existence of the rights of the families of victims of enforced disappearance. The Court stated that the "duty to investigate facts of this type continues as long as there is uncertainty about the fate of the person who has disappeared. Even in the hypothetical case that those individually responsible for crimes of this type cannot be legally punished under certain circumstances, the State is obligated to use the means at its disposal to inform the relatives of the fate of the victims and, if they have been killed, the location of their remains." (*Case of Velásquez Rodríguez*, Judgement of 29 July 1988). A similar position was expressed in the *Case of Godínez Cruz*. In the cases of *Castillo Páez and Blake*, the Court also made reference to the need of families to know the whereabouts of victims. Within the framework of the European human rights system, the European Court of Human Rights in Strasbourg issued its opinion on the duty of States to determine what has happened to people who have gone missing in situations of armed conflict or internal violence. On this subject, the Court established that the failure to disclose information available on persons detained or missing as a result of an armed conflict to their families constitutes inhuman treatment under the rules of the European Convention on Human Rights (European Court of Human Rights, *Case of Kurt v. Turkey*, paragraph 188; *Case of Timurtas v. Turkey*, paragraph 188; and *Case of Cyprus*, paragraph 189).

39 Resolution 9/11 on "The right to the truth", United Nations Human Rights Council (A/HRC/9/L.12).

40 Pursuant to the provisions of article 24.1 of the Convention, "victim" means the missing person and any other individual who has suffered harm as the direct result of an enforced disappearance.

41 See article 24 of the International Convention for the Protection of All Persons from Enforced Disappearance (United Nations). In the same vein, the preamble of this Convention establishes "the right of any victim to know the truth about the circumstances of an enforced disappearance and the fate of the disappeared person".

C. Right to privacy

65.
Scientific methods employed in forensic genetics require the use of biological samples in order to obtain personal information about identified or identifiable individuals. The information contained in the DNA of each person is highly sensitive, as it can reveal information on ethnic origin, biological relationships, health and other private matters. In addition to the obligation to clarify and ensure reparation for serious human rights violations by identifying victims and restoring identity, States must also take into account the rights relating to the protection of an individual's private life, which are also established under international law.

66.
In this regard, article 17 of the International Covenant on Civil and Political Rights provides for the protection of people from arbitrary or unlawful interference in their privacy, family, home or correspondence and from unlawful attacks on their honour and reputation. This right to the protection of privacy must be guaranteed in the face of all such interferences and attacks whether they emanate from State authorities or from natural or legal persons. The obligations established pursuant to this article require that the State adopt legislative and other measures to ensure the effective protection of the right to privacy.

67.
Relevant legislation must specify in detail the precise circumstances in which such interferences may be permitted. The decision to make use of such authorized interference can only be made by the authority designated under the law, and on a case-by-case basis[42]. Interference is considered "unlawful" except in cases envisaged by the law. Interference authorized by States can only take place on the basis of law, which itself must comply with the provisions, aims and objectives of the Covenant on Civil and Political Rights. Interference provided for under the law can also be considered "arbitrary" when it is contrary to the provisions, aims and objectives of the Covenant and unreasonable in the particular circumstances of the case in question[43].

68.
The gathering and holding of personal information on computers, data banks and other devices, whether by public authorities or private individuals or bodies, must be regulated by law. Effective measures have to be taken by States to ensure that information concerning an individual's private life does not reach the hands of persons who are not authorized by law to receive, process and use it, and that it is never used for purposes incompatible with the Covenant. In order to have the most effective protection of their private lives, individuals should have the right to ascertain, in an intelligible form, what personal data, if any, is stored in automatic data files and for what purposes. Every individual should also be able to ascertain which public authorities or private individuals or bodies control or may control their files. If such files contain incorrect personal data or have been collected or processed contrary to the provisions of the law, individuals should have the right to request its rectification or elimination[44].

69.
With regard to the use of forensic genetics to identify people in the field of criminal policy, the European Court of Human Rights has ruled that the improper retention of biological samples and DNA profiles, including indiscriminate collection and indefinite retention, can result in the violation of the right to privacy of the individual affected[45]. The UNESCO International Declaration on Human Genetic Data establishes that human genetic data and biological samples collected from a suspect in the course of a criminal investigation should be destroyed when they are no longer necessary, unless otherwise provided for by domestic law consistent with international human rights law, and that those used for forensic purposes and civil proceedings should only be available for as long as they are necessary for these purposes, unless otherwise provided for by domestic law consistent with international human rights law.

70.
In relation to identification processes in cases involving human rights violations, the United Nations Commission on Human Rights urges States to make every effort to ensure that personal information, including medical and genetic data, is not used in a way that may affect or infringe human rights[46].

71.
Article 19 of the International Convention for the Protection of All Persons from Enforced Disappearance provides as follows: "(1) Personal information, including medical and genetic data, which is collected and/or transmitted within the framework of the search for a disappeared person shall not be used or made available for purposes other than the search for the disappeared person. This is without prejudice to the use of such information in criminal proceedings

42 Human Rights Committee, General Comment No. 16, "The Right to Respect of Privacy, Family, Home and Correspondence, and Protection of Honour and Reputation (article 17 of the International Covenant on Civil and Political Rights)", 32nd session, 1988, paragraph 8.

43 Human Rights Committee, General Comment No. 16: The Right to Respect of Privacy, Family, Home and Correspondence, and Protection of Honour and Reputation (article 17 of the International Covenant on Civil and Political Rights)", 32nd session, 1988, paragraphs 3 and 4.

44 Human Rights Committee, General Comment No. 16, "The Right to Respect of Privacy, Family, Home and Correspondence, and Protection of Honour and Reputation (article 17 of the International Covenant on Civil and Political Rights)", 32nd session, 1988, paragraph 10.

45 European Court of Human Rights, Case of Van Der Velden v. The Netherlands, application 29514/05, and Case of S. and Marper v. The United Kingdom, paragraphs 125 and 126.

46 On this subject, see United Nations Commission on Human Rights resolution no. 2005/26 on Human Rights and Forensic Science.

relating to an offence of enforced disappearance or the exercise of the right to obtain reparation. (2) The collection, processing, use and storage of personal information, including medical and genetic data, shall not infringe or have the effect of infringing the human rights, fundamental freedoms or human dignity of an individual".

72.
Human genetic data has a special status because it can be predictive of genetic predispositions concerning individuals. It can therefore have a significant impact on the family, including offspring, extending over generations, and in some instances on the whole group to which the person concerned belongs. In some cases, the significance of the information it reveals is not known at the time of the collection of the biological samples. Such data may also have cultural significance for persons or groups[47].

73.
Due attention must therefore be paid to the sensitive nature of human genetic data, and an appropriate level of protection for such data and biological samples must be established. Human genetic data and biological samples associated with an identifiable person should not be disclosed or made accessible to third parties, in particular, employers, insurance companies, educational institutions and the family of the person in question[48].All such information must therefore be protected in accordance with the principle of privacy and the rules of confidentiality[49].

47 International Declaration on Human Genetic Data, article 4.

48 International Declaration on Human Genetic Data, article 14.

49 The UNESCO Universal Declaration on the Human Genome and Human Rights establishes inter alia that "[g]enetic data associated with an identifiable person and stored or processed for the purposes of research or any other purpose must be held confidential in the conditions set by law" (article 7).

D. Personal autonomy and the use of forensic genetics to identify victims of human rights and international humanitarian law violations

74.
Forensic genetics is a useful tool for the identification of victims of human rights and international humanitarian law violations and people whose identity is in doubt. It therefore helps States to fulfil their obligations in ensuring the right to justice, identity and the truth and, in this way, guaranteeing the right to human dignity and to make life choices. At the same time, the information contained in the DNA of each individual is of a highly sensitive nature and requires legal protection in order to ensure the right to privacy.

75.
As mentioned above, international instruments concerning bioethics establish a series of safeguards concerning confidentiality, transparency and legality in order to protect the right to non-discrimination in the use of DNA samples, mainly in the light of experiences involving the use of genetics for medical and research purposes. In such cases, the collection of samples is subject to the consent of the donor.

76.
In cases requiring biological samples from a particular donor for the purpose of establishing the identity of a victim of a serious human rights violation or restoring a person's true identity, there are special considerations to be taken into account, particularly in relation to the exercise of personal autonomy and the decision to cooperate voluntarily in the collection process.

77.
It should be noted that processes to investigate serious human rights violations require the participation of all those who are able to provide the evidence needed to effectively determine the truth about what happened, the damage caused and the reparation that should be made to victims, including re-establishment of their true identity.

78.
In cases in which it is not possible to secure the voluntary participation of individuals whose biological samples could be critically important in determining the facts about such crimes, the competent courts must intervene, rule on the matter and issue a reasoned decision. The decision of the court should be made balancing the necessity, reasonableness and proportionality of the measure for that particular case, after exhausting all other less drastic options (non-invasive procedures).

79.
This strict reasonableness and proportionality test must take into account the right to personal autonomy not only of those individuals who refuse to give their consent to the collection of samples, but also of other people affected, whose rights and life choices must also be protected. It must also take into account the imperative public interest in ensuring the effective

Federal Judge stamping boxes containing samples

resolution of the case, which is linked not only to the obligation of States to investigate human rights violations, but also to the obligation to ensure the effective exercise of the victims' right to identity and the right of victims and society as a whole to have the full facts about such violations.

III. BIOBANKS, PERSONAL DATA BANKS AND DNA BANKS FOR USE IN THE INVESTIGATION OF HUMAN RIGHTS AND INTERNATIONAL HUMANITARIAN LAW VIOLATIONS

80.
As stated above, States have an obligation to adopt appropriate administrative, legal and political measures for the investigation and clarification of cases of serious human rights violations, including the identification of victims of such crimes as forced disappearance and the abduction of children, using the most effective technical and scientific methods. To this end, States should establish genetic information systems to facilitate kinship analysis and the identification of victims[50]. The establishment of such genetic information and analysis systems requires the development and maintenance of forensic biobanks and DNA banks.

81.
The identification of human remains or a living person whose identity is in doubt requires the efforts of various forensic disciplines, including medicine, dentistry, anthropology, fingerprinting and genetics. As explained in greater detail below, human genetic identification involves analyzing the DNA in samples taken from human remains or living persons and comparing it with DNA obtained from reference samples left by the missing person prior to his or her death (ante-mortem) or disappearance or from reference samples collected from family members. As the victims' DNA profiles have to be compared with a large number of reference samples, and the samples and data stored for long periods, forensic genetics laboratories need storage systems known as *forensic databases* and *biobanks*[51].

82.
FORENSIC BIOBANK. A forensic biobank is defined as the collection of the original biological samples as well as the DNA extracted from them and used to generate DNA profiles for the

50 Inter-American Court of Human Rights, Case of Anzualdo Castro v. Peru, Judgement of 22 September 2009 (Preliminary Objection, Merits, Reparations and Costs), Series C, no. 202, paragraph 189. Examples of experiences in genetic data bank development and management include the mechanisms in place in Argentina, Chile, El Salvador and the Balkans. In Argentina, the National Genetic Data Bank (BNDG) was set up in 1987 by Law 23511/87 and extended by Law 26548, with a view to kinship analysis in cases involving the enforced disappearance and abduction of children who disappeared with their parents in the period 1974-1983 and DNA-based identification of the remains of victims of enforced disappearance. An estimated 3,500 DNA samples were collected between 1987 and 2009. The Argentine Forensic Anthropology Team (EAAF) also contributed to these identification efforts. The International Commission of Missing Persons (ICMP), created in 1997 in the context of the Balkan conflict, developed a database containing over 88,000 samples from the families of 29,000 missing people, which was used to identify some 15,900 victims. The Asociación Pro-Búsqueda de Niños y Niñas Desaparecidos, created in El Salvador by the Catholic Church in 1994, managed to trace 300 missing children. In 2007, Chile created the Medico-Legal Service Human Rights Programme and started up its Genetic Data Bank for use in cases of serious human rights violations committed in the period 1973-1990. By 2011, the Genetic Data Bank had collected around 3,500 family reference samples.

51 Article 17 of Recommendation 4 (2006) of the Committee of Ministers of the Council of Europe defines "population-based biobank" as follows: "a collection of biological materials that has the following characteristics: i. the collection has a population basis; ii. it is established, or has been converted, to supply biological materials or data derived therefrom for multiple future research projects; iii. it contains biological materials and associated personal data (…); iv. it receives and supplies materials in an organised manner".

Sampling relatives in FTA card

purpose of genetic identification. The following are the types of sample stored in forensic biobanks for the purpose of identifying human remains and missing people:

- Unknown samples from human remains (*post-mortem* samples) or samples from unidentified victims and the corresponding DNA extracts.
- Reference samples left by the missing person or victim (known as *ante-mortem* samples) and the corresponding DNA extracts.
- Reference samples collected from members of the victim's family and the corresponding DNA extracts.

83.
Standard forms should be used when collecting samples for the biobank to record identification details, track the chain of custody and document informed consent.

84.
When developing a biobank for the purpose of DNA-based identification, it is essential to guarantee the quality, security and traceability of the data and samples stored in it. It is also necessary to ensure the confidentiality of the personal information associated with the samples.

85.
It is recommended that the identification data and the analytical results yielded by the biological samples be electronically recorded using information auditable management systems, to enable management and updating available information, identification of samples, assignment of testing and management of analytical results in a traceable and reliable manner.

86.
The laboratories or entities that manage biobanks must have restricted and controlled access areas to ensure the proper storage, custody and preservation of biological samples while investigations are ongoing. They must also establish final disposition procedures (archival preservation or destruction) to be implemented when the genetic identification process has come to an end.

87.
Genetic data banks must comply with the following requirements: they must respect the principles of human dignity and non-discrimination; they may not charge victims and their families for their services; they must ensure the confidentiality of the information provided; they must ensure that samples are used only for identification purposes in accordance with respective informed consents; and they must ensure that data privacy and protection standards are maintained.

88.
DNA PROFILES DATABASES. DNA profiles databases are defined as those that store the DNA profiles generated from the samples contained in the biobank. The profiles can be digitized and stored in different databases, so that systematic searches can be made to compare and match profiles, in order to identify unknown samples from victims.

89.
In order to safeguard the confidentiality of DNA profiles stored in such databases, they are assigned identifying codes to keep personal information separate. The following aspects must be clearly defined:
- the nature of the DNA profiles stored in the database;
- the use and disclosure of DNA profiles and any matches made;
- the laboratories or entities qualified to perform testing;
- sample preservation;
- the right to cancel, correct and access data.

90.
The software solutions used to manage DNA databases for the purpose of DNA-based identification of human remains and missing persons should have been validated in the forensic community. They typically should provide the following capabilities:

- Where possible, electronic upload of DNA profiles to avoid transcription errors arising from manual data entry;
- Systematic matching (all against all) of a large number of DNA profiles arranged into different search indexes (samples from human remains, family reference samples, *ante-mortem* samples, etc.);
- Use of different search algorithms, including: direct matching (DNA profiles from unidentified human remains screened against each other and against profiles obtained from *ante-mortem* samples of missing persons) and indirect matching or kinship analysis (DNA profiles from unidentified human remains screened against DNA profiles from family reference samples) of DNA profiles with autosomal inheritance; maternal inheritance (mitochondrial DNA typing) and paternal inheritance (Y-chromosome typing).
- Automatic integration of a statistical calculation of the likelihood of matches between autosomal DNA profiles in order to determine the significance or strength of a particular match;
- Generation of reports on the DNA matches found for use by those responsible for producing expert reports at the request of competent authorities (e.g. truth commissions, judges and courts).

91.
The entity responsible for maintaining forensic biobanks and DNA profile databases should operate independently, albeit under the responsibility of the State and ensure civil society involvement. International human rights and international humanitarian law organizations can also be involved. With a view to ensuring a clear and transparent sample management policy, this entity must be regularly audited by an independent body duly competent in the field of genetics.

92.
In the instrument that sets up the genetic data bank, rules should be established on administration, purpose, custody, confidentiality, access, duration, separation from other data banks[52], storage, security, interaction with official bodies,

52There are often various genetic data banks in a country with different purposes or different mandates, e.g. criminal casework and the identification of missing persons. These data banks should be managed and treated separately, and the information they contain should not be centralized in a single data bank.

Sample count in court for onward shipment to genetic laboratory

relations with sample donors, transparent management and auditing and civil, criminal and administrative liability arising from misuse of the samples and information stored in the data bank. The facilities must possess infrastructure and appropriate equipment to protect the samples and prevent unauthorized access to databases. Biobank regulations must be consistent with international standards for the quality control of procedures in this field. Biobanks can also promote national and international agreements on sample collection and testing.

IV. SCIENTIFIC, TECHNICAL AND METHODOLOGICAL ASPECTS OF HUMAN GENETIC IDENTIFICATION

A. General considerations concerning human genetic identification

93.
A *genome* is the complete set of genetic material of a given species. The human genome is made up of deoxyribonucleic acid (DNA) and other molecules contained in structures called *chromosomes*, located in the cell nucleus (nuclear DNA). There is also DNA outside the nucleus in cytoplasmic organelles called *mitochondria* (mitochondrial DNA). DNA is the main component of the genome and contains information enabling the cell to produce proteins and other structural and regulatory molecules. The structure of DNA consists of two long chains of units called *nucleotides*. There are four nucleotides in DNA named according to the base that is attached: adenine (A), guanine (G), cytosine (C) and thymine (T). The human genome is around three billion nucleotides long. The *sequence* of the four types of nucleotides along the length of the DNA strand contains the genetic code which, among other functions, instructs the cell about the aminoacid sequence that each protein produced must have.

94.
Human nuclear DNA is comprised of 23 pairs of chromosomes (46 in total) in most cells. Each pair consists of one copy of genetic material inherited from the father and another from the mother. Twenty-two of them are what are known as autosomal chromosomes, and the remaining one is the sex-chromosome pair (XX in females and XY in males). Therefore, in somatic cells, the genetic content is duplicated (diploid), unlike in reproductive cells (e.g. sperm), which have only one set of chromosomes (haploid). A defined position in the genome is called a *locus* (plural *loci*), which means “place” in Latin. Each locus is represented twice in the diploid cell – different versions of the same locus are called *alleles* and occupy the same locus on homologous chromosomes.

95.
Each human being has a unique and distinct genetic make-up (except in the case of monozygotic twins, who are genetically identical and indistinguishable). This human genetic diversity is the result of variations in DNA sequence caused by spontaneous genetic changes (*mutations*), which have occurred throughout human evolution over tens of thousands of years and continue to occur. It has been calculated that human beings differ from each other in at least one out of every thousand nucleotides in a DNA sequence consisting of three billion nucleotides. Human genome sequencing and subsequent developments have shown that the very extensive genetic variability in humans is mainly observed in DNA that is not used by cells to code for protein synthesis (non-coding DNA, which accounts for approximately 98% of the human genome). These variations, referred to generically as *genetic markers*, include repeated sequences of nucleotides, the length of which varies from one individual to another: *minisatellites* (variable number tandem repeats – *VNTRs*) and *microsatellites* (short tandem repeats – *STRs*). In addition, it is estimated that there are at least three million locations in the DNA sequence where individuals vary from each other in the type of nucleotide present (single nucleotide polymorphisms – SNPs). Furthermore, as mentioned above, in addition to nuclear DNA (located in chromosomes in the cell nucleus), there is also DNA in organelles in the cell cytoplasm, called *mitochondria* (mitochondrial DNA or mtDNA). Only the mother passes mitochondrial DNA on to her offspring. It is used in human genetic identification to determine maternal lineage (mother, maternal aunts and uncles and maternal grandparents) and also to identify human remains. An additional feature of mtDNA is that it is present in multiple copies in a cell, thereby increasing the likelihood of recovering sufficient DNA from tissues, even when there is a high degree of molecular degradation.

96.
Genetic identification involves analysing the DNA extracted from a biological sample containing the cells of a deceased or living person. The DNA can be extracted from samples collected directly from the person (blood, saliva, hair, skeletal remains, etc.) or left by the person prior to their disappearance or death (blood stains, saliva, cells found in used underwear and on other objects, etc.). As explained below, DNA-based identification involves generating the individual’s DNA profile by analysing various genomic loci. However, it is not possible to identify an individual by simply analysing his or her DNA; it is necessary to match this DNA profile to DNA extracted from reference samples left by that person before he or she died or went missing or from reference samples collected from people who could be biologically related to him or her. It is important to remember that “identification requires matching”. It is not possible to identify someone simply by carrying out DNA testing (or any other technique) unless the unknown DNA profile can be compared and matched to profiles obtained from known biological samples (e.g. match between the DNA from an unknown victim sample and the DNA of biological relatives).

97.
Forensic genetics is a subdiscipline of human genetics, the aim of which is to identify individuals or biological traces through DNA typing. The international forensic community has agreed on a standard pool of markers useful in forensic analysis and human identification. The most commonly used are the above-mentioned short tandem repeats (STRs) and single nucleotide polymorphisms (SNPs). STRs are found in nuclear DNA, both in autosomal chromosomes and the pair of sex chromosomes (X and Y). SNPs are found in both nuclear and mitochondrial DNA.

98.
STRs located on the Y chromosome (Y-STRs) are passed on from father to son and are therefore markers of paternal lineage. On the other hand, mitochondrial DNA is only passed on to offspring by the mother and is therefore used in forensic identification as a maternal lineage marker.

99.
Autosomal genetic markers are preferred because they can uniquely identify an individual, owing to their high power of discrimination. Nevertheless, lineage markers (Y-STRs and

Genetic Laboratory of the Argentine Team of Forensic Anthropology in Córdoba, Argentina

mtDNA) are also useful when attempting to match samples from people several generations removed or from more distantly related family members.

100.
Some markers that are useful in human identification can also provide information on an individual's ancestry. Their use must therefore be subject to strict ethical standards and criteria to prevent any kind of discrimination.

101.
The following sequence of steps is established for DNA-based identification of missing persons:

a) collect biological samples from the victim and family reference samples.
b) assign a code number to victim and reference samples for inclusion in biobanks.
c) Ship samples to the forensic genetics laboratory ensuring the chain of custody.
d) Perform the extraction of DNA from samples and, when necessary, its quantification.
e) Analysis of DNA markers.
f) Generate DNA profiles.
g) Store digitized DNA profiles in databases.
h) Compare DNA profiles of victim samples against DNA profiles of reference samples to find matches.
i) Perform statistical evaluations and interpret results.
j) Reconcile the results with the findings obtained by other forensic identification disciplines.
k) Report results.

B. Identification of remains of missing persons

102.
Determining the identity of missing persons involves different forensic disciplines. Complexity factors include: (1) the number of people assumed to be missing compared to the number of people actually missing (possible discrepancies between assumed, reported and actual cases); (2) number of individuals presumed missing; (3) "closed" incidents compared to "open" or "mixed" incidents[53]; (4) number of victims recovered; (4) condition of the remains recovered (state of preservation); (5) degree to which remains are disarticulated and scattered; (6) degree to which remains are commingled; and (7) availability and type of reference samples. Another factor is the financial resources available to implement the identification process and see it through to completion.

103.
The process of identifying a body, whatever its condition, involves comparing two types of data, as explained above. *Ante-mortem* data (records and information about the person prior to death) and *post-mortem* data (data obtained from the dead body). The nature of the comparison and the type of data depend on two key factors: a) the condition of the body (e.g. in the case of skeletal remains, there are no fingerprints to be matched); and b) the quality and quantity of ante-mortem information available. Although contextual evidence (clothing, personal items, circumstances of the disappearance, discovery of the body, eyewitness accounts by perpetrators, etc.) is not, on its own, sufficient to make an identification, it can support other means of identification and contribute to the process. It is important to realize that these elements, on their own, may not often provide sufficient evidence for an identification. In order to make a positive identification, all these elements should be analysed and compared in a comprehensive forensic report prepared with input from medical doctors, anthropologists, odontologists and geneticists.

104.
It is important to evaluate the capacity of the available laboratory or laboratories to process and analyse a large number of samples. It is advisable to select laboratories with proven experience in forensic genetic testing, as the analyses required in investigations of this kind are often very complex, requiring a high level of technical expertise and expensive equipment. In cases in which there is a paucity of forensic evidence or post-mortem samples, the failure to handle them correctly can result in the destruction of what evidence there is. When more than one laboratory is involved in processing and analysing biological samples, the genetic tests to be performed in different cases must be standardized and compatible, so that the data can be compared.

53 In a "closed" incident there is a list of victims (e.g. air accident), while in an "open" incident, although the identity of some of the victims is thought to be known a priori, the list is not complete or is not as reliable as in a closed incident. A "mixed" incident is a combination of the two.

C. Reference sample collection

105.
The main purpose of this section is to describe the basic procedures that should be implemented in investigations in which missing persons need to be identified using DNA analysis techniques. The following steps should be implemented in an ordered manner: (1) collect *post-mortem* evidence, (2) collect different types of reference samples, (3) produce a family tree of the family member reporting the disappearance, showing his or her relationship to the missing person and (4) address legal and ethical aspects involved in the families giving their consent to conduct genetic studies.

106.
As explained above, in the identification process, DNA collected from a deceased or living person (or from personal items), whose identity is unknown, is matched to DNA extracted from known reference samples. A known reference sample is one provided by a donor whose identity is known or can be verified (e.g. samples provided by biological relatives of the missing person).

107.
The task of collecting reference samples is one of the most sensitive stages in the process, not only from a technical and legal point of view, but also from a psychological and ethical perspective. The collection of reference samples should therefore be carried out by trained personnel who are familiar with the requirements which must be met in connection with certain key issues, such as maintaining the chain of custody, obtaining the informed consent of family members providing samples, ensuring that there is no cross-contamination of samples, uniquely identifying each sample, determining the exact relationship with the missing person and addressing the psychosocial needs of the victims.

1. Considerations relating to reference sample collection

108.
It must be taken into account that the context in which a donor provides a reference sample (either as a relative of a missing person or as someone whose identity needs to be re-established) is emotionally traumatic. Interviewers should therefore take into account the psychosocial needs of donors. It is important to remember that the act of providing a reference sample has a symbolic significance in the search for a missing family member or for one's own identity. Great care must therefore be taken to ensure that donors are treated with sensitivity and respect and not forced into making a decision about providing a sample.

109.
The transparency of the process is another key tenet. It is advisable to inform donors which institution is in charge of the investigation, all progress made, the scope, limitations and possible outcomes of the investigation and the time it may take. They should also be informed that they will not be charged for the DNA testing and that all information is completely confidential. It is important not to raise false hopes, promise donors results or establish deadlines that cannot be met in practice. It is important to explain the limitations of

forensic genetics, particularly in cases when it is not possible to extract and analyse DNA.

110.
It is crucial to create a safe and trusting atmosphere for donors and interviewees. The safety of a donor should never be compromised even when the sample is important to the investigation in question, and sample collection arrangements should be properly planned.

111.
Consideration should be given to circumstances that could hinder the collection of reference samples from relatives. All possible assurances should be given to help them to provide the sample, so that the required testing can be performed. Measures that can be taken to solve these problems include professionals travelling to the incapacitated person to take the sample, using equipment and materials that ensure compliance with biosafety requirements and offering potential donors the possibility of a less frightening sample collection procedure, for example, saliva or hair instead of blood, which they may regard as less invasive. If they still do not wish to give a sample, they could be asked to provide a personal item, such as a toothbrush, shaver or hairbrush. In such cases, however, it should be made clear that there could be uncertainty about the results yielded by the cells recovered, if the item has been used by more than one person.

112.
It is recommended to set up a family assistance centre or specific unit (sample collection facility) within the governmental or non-governmental organization responsible for the investigation and identification process, with a view to centralizing all sample collection in a facility dedicated specifically to this task. This will ensure that appointments and interviews are arranged in an orderly and efficient manner and facilitate protocol standardization, sample coding, the release of information through the media, donor contact management, collection of ante-mortem information about the victim and the informed consent procedure for the collection of biological samples from donors.

113.
When family reference samples are collected, the donor should be interviewed and personal information about the victim which could contribute to forensic identification methodically recorded, following established guidelines and using standard ante-mortem information forms. The information that should be recorded includes full name, identity document numbers, place and date of birth, general description, physical characteristics, distinguishing marks, tattoos and any other information that could prove useful in the investigation.

114.
It is also recommended to record relevant medical information relating to the donor, for example, whether they have received a transplant or have any genetic disorder that could skew the interpretation of the DNA analysis.

115.
It is recommended that each donor be given the opportunity to contact the sample collection personnel or centre again if they wish. Sometimes, there is information (e.g. about paternity) that they would prefer not to disclose in the presence of other family members, but which they might later clarify if they can contact the centre. It also enables donors to provide further information about the family tree or additional reference samples from other relatives.

116.
Issues relating to sample coding, informed consent, information confidentiality agreements, etc. are dealt with in greater detail below. There are two types of reference samples: samples provided by relatives of the missing person (Table 1) and samples taken from personal items used by the victim or biological samples stored at medical facilities and blood banks (Table 2). When possible, both types of reference sample should be collected.

2. Biological relatives of the missing person[54]

(a) Type and classification of relationship with the missing person:

117.
Samples should be collected from as many relatives of the missing person as necessary, preferably first-degree relatives (parents, siblings and offspring) (see Table 1).

118.
The exact nature of the donor's biological relationship to the victim is of crucial importance in the forensic investigation. In the interview with the victim's family members, it is therefore important for the person collecting the sample to accurately record the relationship of the relative(s) to the victim, along with all relevant information about the family pedigree. A clear description of the relationship will facilitate the selection of the genetic markers that the genetics laboratory will analyse for subsequent matching.

119.
The forensic genetics laboratory will decide which relatives are most suitable for the collection of reference samples (see Table 1). It is recommended that a pictorial representation of the family structure be produced, particularly in the case of second and third-degree relatives. A family tree should be drawn using standard symbols:

- Male = □
- Female = ○
- Unknown = ◇

120.
It is important to describe how the donor is related to the missing person, with the donor at the centre of the family tree, and not the other way round. For example:
- full brother/sister of the victim;
- paternal or maternal half brother/sister of the victim;
- brother/sister of the victim's mother (rather than maternal aunt or uncle);
- in the case of nieces and nephews, it must be clearly specified whether they are related on the mother's side or on the father's side, for example: son/daughter of the victim's brother, son/daughter of the victim's sister, etc.

54 The same recommendations apply in cases involving re-establishment of the identity of living persons by analysing their relationship to purported biological relatives.

121.
It is important to avoid abbreviating the biological relationship, because although it is more tedious to record the full description, it helps the forensic genetics laboratory to decide what kind of DNA testing is possible or preferable. For example, the use of the term “paternal male cousin” will result in valuable information being lost, as it could mean “son of the father’s brother” or “son of the father’s sister”. Y-chromosome analysis could be used In the first case, but not in the second. An abbreviated description of the relationship would therefore withhold information required to make decisions about testing. As mentioned above, it is recommended that family trees be drawn to represent relationships accurately.

122.
Table 1 shows the possible combinations of samples from relatives that can be used in studies to identify missing persons. It is always preferable to have reference samples that are as widely representative of the family as possible.

Table 1 – EXAMPLE OF USEFUL FAMILY REFERENCE SAMPLES

Monozygotic twin (identical twin)
Both parents
Four grandparents
One parent, spouse and offspring
Child and spouse
Two grandparents and a parent from different sides of the family
One parent and one sibling
Two or more siblings

123.
If it is not possible to collect reference samples from the victim’s first-degree relatives, samples should be collected from second-degree relatives. In this case, a greater number of relatives is required, as they are less informative. In special cases, when there are not enough informative relatives, a request can be made to exhume the bodies of relatives in order to take bone samples and obtain valuable genetic information. It is important to take into consideration that the threshold to be established to consider a reliable identification (DNA match) will be highly influenced by the number of victims involved.

124.
In some cases, the samples provided by first-degree relatives of the missing person may provide genetic information that is valid for one type of genetic analysis (e.g. autosomal STR markers in nuclear DNA), but not suitable for others (e.g. mitochondrial DNA or Y-chromosome analysis). For example, in the case of a missing man where blood samples are available from the mother and sister, the analysis of STR and mitochondrial DNA markers could be performed, but not Y-chromosome analysis. In such cases, in order to supplement the reference genetic information, it is necessary to obtain samples from other relatives who, although more distantly related, are useful in providing lineage markers (Y-STR for paternal lineage and mtDNA for maternal lineage).

125.
It is recommended to collect samples from relatives who can provide genetic information that is not repeated. For example, if blood samples have been collected from the mother and the father of a missing person, it is not necessary to take samples from siblings too, as they will provide the same information as the parents’ profiles, which are already available. By the same token, if the sister of a missing person has donated a sample, then a sample taken from her daughter (niece of the missing person or daughter of the missing person’s sister) will provide no additional genetic information to that already obtained from the mother. Information obtained from the niece’s sample would therefore be redundant.

126.
In cases in which biological samples from the victim’s descendants (offspring) are available, it is advisable to also take reference samples from the spouse (biological parent), with a view to partially reconstructing the victim’s DNA profile and increasing the power of discrimination in the genetic study. It is also necessary to determine whether the victim’s offspring are full siblings (whether they have the same mother and father), with a view to avoiding inconsistencies when the family group is analysed as a whole.

(b) Types of reference samples collected from relatives of the missing person

127.
- **Blood:** Blood samples are taken by fingerstick or venipuncture, after disinfecting the puncture site with alcohol. Several drops of blood are placed on a suitable absorbent card (blood card, filter paper, etc.). The samples must be labelled and signed by the donor. Once dried, they should be stored in envelopes/foil pouches and can be stored at room temperature for long periods of time. Gloves should be worn when handling samples in order to avoid inadvertent cross-contamination. This is an invasive sample collection procedure.
- **Buccal swabs:** There are currently a variety of commercially available sample collection materials which can be used to collect epithelial cells non-invasively from the inside of the cheek (2-3 swabs). Once dried, the swabs can be preserved freezed or at room temperature . Kits specifically designed for the collection of this kind of sample can also be used.
- **Saliva:** Another alternative is the collection of saliva samples (1 to 2 ml). Saliva is placed on a suitable absorbent card (blood card, filter paper, etc.), air dried and stored at room temperature or freezed.
- **Nail clippings:** Nail clippings provided by the victim’s relatives are another long-lasting, stable source of genetic material which can be used for DNA matching. It is recommended to take clippings from several fingers.
- **Plucked hairs (with root):** Several hairs are required to obtain results and repeat testing if necessary. The hair must be plucked, not cut, and must have the follicle attached. Hairs may be collected just in case that the preferred material describe above is not available.

128.
Samples of hair with the root attached are particularly useful in the case of people who have had an allogeneic bone marrow transplant, because their blood yields a DNA profile that is no longer their own, but that of the transplanted bone marrow.

3. Informed consent

129.
Securing informed consent must be addressed as a communicative process between a qualified personnel and the sample donor, assisted by an interpreter when necessary and involving the exchange of clear and comprehensive information. Donors must fully understand how the sample will be used and what significance the genetic testing and results will have. After the interview and the oral explanation provided by the interviewer, donors are required to make a written statement, confirming that they have understood the purpose and scope of their participation and giving their consent for the sample to be taken. In the case of children, adolescents and people whose capacity is impaired at the time of the interview, the procedure should be adjusted to comply with the legislation in force in the country in question.

130.
The consent form must contain the following information about the donor:

- Given name and surname
- ID document number and type
- Date of birth
- address and contact details
- Photograph taken at the time of sample collection (optional)
- Right/left thumbprint (optional)
- Signature
- Relationship with the victim or missing person recorded as explained above
- whether the donor has received a bone marrow transplant or a recent blood transfusion.

131.
The consent form must explain the scope, objectives, limitations, benefits and potential detriments of genetic identification studies. It is suggested that the consent form includes the following:

a. authorize the inclusion of the sample donor's DNA profile in the genetic databases operated by the body responsible for identifying victims;
b. authorize the screening of the sample donor's DNA profile against the profiles in missing person databases;
c. specify the objective, limitations, benefits and potential detriments of genetic studies, clearly stating that DNA profiles will be used solely for identification purposes;
d. provide assurances of confidentiality, protection and restricted access to the information;
e. provide assurances that the genetic data obtained will not be used for purposes other than the identification of the victims who are the object of the study;
f. authorize the anonymous use of the data collected in order to produce statistical population data should it be necessary to create population frequency databases;
g. specify that donors have the right to request access to their DNA profile(s), as well as their withdrawal from process;

Blood samples from relatives

h. specify sample collection place and date and the person responsible (signature);
i. specify final sample and profile disposition (it is recommended to destroy them once the purpose of the genetic analysis has been fulfilled).

132.
The form should use clear, plain language which is easily understandable to the sample donor, avoiding ambiguous wording.

133.
The donor must be given a copy of the consent form signed by him or her and by the person responsible for collecting the sample.

4. Victim samples

134.
The recovery of biological trace evidence (samples) from items containing cellular material from a victim is another way of obtaining reference samples when death or disappearance is relatively recent. It is recommended that the victim's family members be asked to provide this type of sample wherever possible. As with family reference samples, personal items belonging to the victim which could provide this type of sample should be collected by trained personnel. As with forensic samples, personnel handling them must work in clean controlled conditions to avoid contamination with foreign DNA or with microorganisms which could cause degradation of the genetic material present in the samples selected for testing. As with all trace evidence, strictly speaking, they cannot be considered known samples, but they can provide a DNA profile that can be used for comparison with other samples.

135.
Personal items provided as reference samples might not have been used solely by the victim (e.g. hairbrush), or family members could inadvertently provide items that did not belong to the victim. It is therefore advisable to verify that samples from personal items do actually belong to the victim by matching them, wherever possible, to family reference samples by means of DNA analysis. Once it has been verified that the DNA recovered from the personal item does in fact belong to the victim and does not contain mixed DNA profiles, the sample can be used in DNA matching to make an identification.

136.
The use of samples from the victim is extremely useful, because the power of discrimination (potential for individualization) is much stronger in direct matching, which compares a DNA profile obtained from a personal item known to belong to the victim with a DNA profile from post-mortem samples, than in indirect matching or kinship analysis, which compares the missing person's DNA profile with those of biological relatives.

137.
As explained in greater detail below, items considered to be of use in such studies must be placed separately in clean, labelled paper envelopes and stored appropriately, along with accompanying documentation, until such time as they are tested.

138.
Table 2 provides a detailed list of the kind of items suitable for testing. It also indicates the quality of the DNA usually extracted from samples recovered from these items.

Table 2 - CLASSIFICATION OF PERSONAL ITEMS FROM WHICH THE DNA OF A MISSING PERSON CAN BE RECOVERED FOR USE AS A REFERENCE SAMPLE

DNA QUALITY	COMMONLY AVAILABLE	MIGHT BE AVAILABLE
Good	Toothbrushes, electric shavers, razor blades, hairbrushes and combs.	Samples from a bone marrow donor programme, newborn metabolic screening card, forensic and laboratory paternity testing databases, reference samples from military personnel, clinical blood samples, sperm bank samples, dried umbilical cord, deciduous teeth (primary or milk teeth), extracted teeth and paraffin-embedded pathology specimens.
Fair	Lipsticks, deodorant sticks, pillowcases, used cups and glasses and used underwear.	Cervical smears, fingernail clippings, cigarette butts, pipes, motorcycle and other sport helmets, hats and caps, inner clothing items (bra, T-shirt and socks), ear plugs, earphones, eye glasses, pens with teeth marks and mailed envelopes and postcards.
Poor	Jewellery, wrist watches, outer clothing, towels, shoes, hair bands and earmuffs.	Baby hair, dentures, hair rollers and nail clippers, scissors and files

Note: taken in part from Forensic Science International Genetics 1(1), 3-12.

D. Sample collection from human remains (*post mortem*)

1. Coordination of teams responsible for collecting human remains for testing

139.
Depending on the circumstances, the remains can be evaluated at the recovery site or at a morgue or forensic anthropology laboratory. The personnel assigned to this task (qualified morgue personnel, assistants, anthropologists, etc.) should have the necessary training to collect post-mortem samples in accordance with aseptic handling and personal protection requirements, to decide what kinds of sample to collect, to classify the samples and to prevent sample contamination by using gloves, masks and other necessary apparel. Prior to sample collection, the body or skeletal remains should be examined (medico-legal autopsy or forensic anthropological analysis respectively) in order to record findings before any segments are removed and to evaluate the nature of the case, so that the most appropriate sampling strategy can be chosen (particularly important in cases of fragmented, incomplete or commingled remains). A careful assessment must be made of the condition of the post-mortem samples, as this will influence DNA typing success rates.

140.
It is advisable to consult with the forensic genetics laboratory beforehand in order to establish the most suitable types of post-mortem sample for DNA analysis. Ideally, different tissue samples should be collected from the best preserved parts of the remains. The possibility of taking duplicate post-mortem samples of tissues for additional DNA testing should

also be considered, as post-mortem samples that are poorly preserved or degraded as a result of environmental conditions, the passing of time or physical and chemical factors may require more than one test procedure to obtain reproducible results. Having an additional sample also permits cross-testing and analysis by other laboratories involved in the DNA-based identification process and the storage of genetic material, if permitted or ordered by the competent authority.

141.
In the case of fragmented or commingled remains, criteria should be defined in advance for the identification of fragments (minimum fragment size to be typed for identification, number of samples that can be analysed, taking into account available financial resources, etc.). Families and/or family associations and the local authorities concerned should be fully informed about this particularly sensitive issue and involved in decisions. It is recommended to plan step-by-step sample collection, so that the case can be reassessed and further samples collected depending on the results obtained. This is particularly important in cases involving a large quantity of commingled remains.

142.
Anatomical parts identified by other means (fingerprinting, anthropology, odontology, etc.) are very important in the reassociation of a victim's remains or as reference samples to identify other people from the missing person's family.

143.
Different soft tissue and bone samples can be collected, depending on the state of preservation of the remains (Table 3).

144.
Containers used for collecting post-mortem samples should be sterile, one-use and spill-proof and allow for secure labelling. It is preferable to use commercially available kits specifically designed for the collection of forensic evidence or clinical samples.

145.
The correct labelling and identification of samples and accompanying documentation is vitally important.

Table 3 - COLLECTION OF POST-MORTEM SAMPLES FROM HUMAN REMAINS

CONDITION OF BODY	RECOMMENDED SAMPLE
Non-decomposed, complete body.	Deep muscle tissue, blood (on card or swab), buccal swabs, nail clippings.
Non-decomposed, fragmented remains.	If available, blood and/or deep muscle tissue (0.5-1.0 g).
Decomposed complete body and fragmented remains.	Sample from diaphysis of long, compact bone (4-6 cm section) or/and 3 healthy teeth without dental work (preferably molars).
Severely burnt bodies (not charred).	Any of the samples listed above or swabs from inside the urinary bladder.
Skeletal remains.	Preferably teeth (3 teeth) or/and long compact bone diaphysis (4-6 cm section), particularly femur ,or tibia Always take samples from visibly well-preserved parts of the remains. If other types of bone are collected, compact bone with dense tissue is preferred.

2. Skeletal remains

146.
The types of post-mortem sample that can be collected from skeletal remains are listed in Table 3. They are collected by means of archaeological techniques commonly used in forensic anthropology. Once the anthropology laboratory tests have been completed, two or three teeth can be removed or a window section cut from a bone, preferably a long bone (femur, tibia). This strategy ensures that the integrity of the remains is maintained until they are identified and released for burial. If the bones referred to above are not available, the anthropologist or forensic pathologist in charge of the case should take samples from the dense compact part of the bone to be tested, avoiding spongy bone. It is not recommended to collect whole skeletal elements (e.g. a whole femur or humerus), because they will take up precious storage space at the forensic genetics laboratory, when only a few grams are required for DNA testing (Table 3).

3. Soft tissue

147.
When fresh remains or recent burial sites are to be analysed, the remains may contain soft tissue, such as muscle. In such cases, soft tissue samples can be used for DNA extraction, if they are sufficiently well preserved.

148.
The samples should collected in suitable, separately labelled containers and stored, if possible, at a temperature between -20° C and -80° C until such time as they can be tested at the forensic genetics laboratory. The different types of post-mortem soft tissue samples that can be collected are shown in Table 3. Soft tissue samples should preferably be taken from deep muscle to prevent cross-contamination with DNA from other sources. When the remains are in an advanced state of decomposition, bone samples are the best source of DNA.

149.
If freezers are not available for the storage of soft and semi-decomposed tissue samples, pure medical alcohol or some other commercially available preservative suitable for biological specimens should be added to the collection container. Do not preserve samples in formalin, as it can cause molecular changes to the DNA, which can prevent DNA analysis.

Dental sample for genetic analysis

E. DNA-based identification of living persons

150.
From the perspective of forensic genetics, the methodology used to identify living persons, including people who were deprived of their identity when they were abducted as children or in other circumstances, does not differ substantially from that described above. In cases in which the biological parents are deceased, there are generally other relatives of the person whose identity is in doubt who can donate reference samples for DNA-based identification (grandparents, aunts, uncles, siblings, etc.).

151.
In investigations to identify living persons whose identity was changed when they were abducted and/or illegally adopted, samples are collected from them for DNA testing. These samples are then compared with family reference samples. The biological samples that can be collected from living victims in these cases are the same as those collected from the victim's relatives or any other living person: blood, saliva, swabs, hair with root attached and personal items (Table 2). DNA typing and matching of these samples from living persons with family reference samples is carried out in the same way as for forensic evidence and post-mortem samples. In some countries, there are genetic databases that store the DNA profiles of the relatives of missing people's sons and daughters who were abducted as children. The DNA profiles of living persons whose identity is in doubt are compared with the profiles stored in these databases.

152.
As with post-mortem samples, samples from living persons must be managed in accordance with general procedures and standards concerning sample collection, storage, transportation and chain of custody, the creation of genetic databases and the scope and limitations of genetic studies conducted with the samples.

F. Sample coding

153.
Special care must be taken with coding both reference samples (samples from biological relatives and direct samples from victims) and samples from the remains to be identified, as the use of a logical numbering system, with no duplication, ensures the correct individualization and effective tracking of samples. It is essential to adopt mechanisms to ensure that all original sample code numbers are traceable, particularly when they might change during the process.

154.
The numbering system must identify samples individually and unequivocally. The identifiers assigned to samples must be unique and centralized in a single database in order to avoid duplication.

155.
It is recommended to use a simple numbering system. Where possible, barcodes should be used, as they ensure fast, easy, fail-safe sample management and minimize transcription errors.

156.
A system that assigns a unique number (identifier) to each reference sample and the corresponding victim has a number of advantages, as it:

- minimizes typographical and spelling errors when transcribing the name and avoids problems arising from victims or donors having the same name.
- facilitates use in DNA matching software data handling.
- allows for the construction of family groups by linking various family members and the victim under a single case number, specifying the relationship of each one to the victim.
- maintains the confidentiality of the information, as the number replaces the name.

G. Sample collection, storage, transportation and chain of custody

157.
Samples collected for DNA analysis (reference and post-mortem samples) should be individually placed in separate containers, assigned a code number, duly labelled and accompanied by all pertinent documentation (photographs, description and any other relevant information). All chain-of-custody requirements must be met. In some cases, a court order may be required to collect samples and it may even be necessary for an officially appointed officer to be present to witness sample collection. It is important to ensure that all requirements relating to court orders and safeguards have been met prior to sample collection.

158.
In the case of personal items left by the missing person, the following details should be recorded:

a. type of sample/object;
b. place (home, hospital, etc.) and date of sample collection;
c. in the case of samples from hospitals, blood banks, sperm banks, etc., the name and signature of the person releasing the sample;
d. photograph of the sample at the time of collection;
e. personal details and signature of the family member who authorizes collection of the object;
f. name and signature of the person responsible for collecting or taking delivery of the sample.

159.
How samples are stored depends on the type of specimen:

a) *Dry samples* (bone samples, samples on solid supports, etc.): paper bags, cardboard boxes or paper envelopes are recommended (always avoid cellophane and nylon as they retain humidity). They can be stored at room temperature although it is preferable to freeze them.
b) *Wet samples*: place them in airtight, sterile containers specifically designed for the collection of biological samples and, if possible, freeze them as soon as possible. Avoid the use of formaldehyde solutions of any kind.

160.
It is advisable to close and seal the sample containers, so that they are tamperproof.

161.
All related information must be stored with the sample, including post-mortem sample collection records and consent forms for reference samples. Whenever the samples are moved or when they are shipped to the genetics laboratory, the chain-of-custody must be strictly maintained and all transfers of custody tracked throughout the process. Even when the samples are not required for a court case, keeping a record of who has had access to them ensures clarity and transparency throughout the process.

162.
Depending on circumstances and arrangements with the genetics laboratory, additional information can also be included

in the chain-of-custody documentation, such as hypotheses about identity, samples to be compared, the condition of the remains, etc.

163.
It is recommended to electronically upload sample data and photographs into databases in order to avoid transcription errors and optimize sample tracking.

H. Technical procedures

164.
This section describes DNA extraction and quantitation procedures. It provides a brief overview in layman's terms, as technology in this field is advancing very rapidly and forensic genetics laboratories have their own updated technical and scientific guidelines. Although the procedures described refer mainly to the identification of human remains, those used for DNA-based identification of living persons who were deprived of their identity when they were abducted as children or in other circumstances are basically the same in technical terms.

165.
The international forensic community uses various methodologies for extracting DNA from biological samples, including automated and robotized techniques, which are recommended when large numbers of samples need to be processed, as they are faster and reduce operator errors. Based on the type of forensic sample to be tested, the laboratory should choose the most suitable procedure for obtaining the best quality DNA, which may be assessed according to the samples being processed.

166.
The laboratory must be properly equipped and prepared to process minute and limited biological samples, including blood, swabs, saliva, biological material on fabric, paper, etc. It must also be capable of processing severely degraded, decomposed and burnt samples, skeletal remains of different ages, etc.

167.
Laboratories assigned such work must operate to the highest quality control and assurance standards, including ISO 17025/2000, and their procedures validated and subjected to periodic checks. When more than one laboratory is involved, it is advisable for them to agree on common procedures and protocols.

1. DNA extraction and quantitation

168.
Forensic genetics laboratories currently use a variety of methodologies to extract and purify DNA with different commercially available kits and protocols. Laboratories must endeavour to use the most reliable DNA purification methods, particularly in the case of post-mortem samples. As the quality and quantity of DNA that can be extracted from a forensic sample varies greatly, the laboratory should quantitate the DNA extracted. This produces better results in DNA profiling and sequencing.

169.
There are various methods for quantitating the DNA in samples. The most widely used in recent times is the real-time polymerase chain reaction (PCR) method. As with DNA extraction, the forensic laboratory must use a validated method for DNA quantitation, particularly in the case of critical samples.

2. Analysis of DNA markers

(a) Analysis of autosomal STR markers

170.
As mentioned above, the most widely used markers today are autosomal STRs. The international forensic community has agreed on a set of duly validated markers for typing.

171.
There are commercial kits that are widely used and validated by forensic genetics laboratories throughout the world. They type various genetic markers in a single reaction; some kits are suited for the analysis of highly degraded material.

172.
When DNA profiles are obtained from degraded or compromised post-mortem samples, the electropherograms (graphs showing DNA sequencing) should be checked with special care and proper procedures to confirm any conclusions drawn.

(b) Analysis of Y-chromosome markers

173.
The analysis of STR markers on the Y chromosome (Y-STRs) can be useful for matching with male relatives of the missing person.

174.
It is important to note that genetic lineage markers (mitochondrial DNA and Y-STRs) come from just one parent (mtDNA from the

mother and Y-STRs from the father) and are shared by all the individuals of the same lineage (maternal or paternal). Special care should be taken in the case of matching using lineage markers when there is more than one victim from the same line, as they will have the same markers. There are also SNPs located on the Y chromosome (Y-SNPs), which have been used extensively in DNA testing. However, special consideration should be given to ethical issues, as this type of analysis can provide information on ancestry.

(c) Analysis of X-chromosome markers

175.
Like Y-STRs, STR markers on the X sex chromosome (X-STRs) can be useful in matching profiles when a generation is missing and there are male relatives available for testing, as they pass the X chromosome on unaltered to their daughters as they received it from their mothers (except for mutational events).

(d) Analysis of mitochondrial DNA

176.
In highly degraded samples (burnt remains or old skeletal remains), analysis of STR markers may not be successful. In such cases, mtDNA analysis is more likely to yield results, as this type of DNA is present in high copy numbers in each cell. Additionally, as it is a lineage marker (maternal), it increases the possibility of making an identification as the profile can be matched to the profiles of reference family members who are only distantly related to the victim. However, this very characteristic means that care must be taken when interpreting DNA match results, in cases in which more than one person from the same maternal lineage is missing.

177.
It should be noted that a match made using mtDNA and/or Y-chromosome is not as powerful (in terms of discrimination or individualization) as one made using STR markers.

178.
When various laboratories are involved, it is recommended that they agree on quality standards for electropherograms to ensure reporting consistent with recommendations made by the international forensic community.

(e) Other genetic markers

179.
In addition to autosomal STR markers and uniparental genetic markers (mtDNA, Y-STRs and X-STRs), there are other binary genetic markers which can be useful in identifying people particularly when samples are degraded, namely SNPs or indels (acronym of insertion/deletion).

180.
Single nucleotide polymorphisms (SNPs) are markers which are useful in human identification. While the power of discrimination of an SNP is weaker than that of an STR marker, the analysis of four or five SNPs provides the same power of discrimination as the analysis of one STR marker. There are currently techniques for typing approximately 50 SNPs, which provide a power of discrimination equivalent to that of the 15 STRs included in commonly used commercial kits. An advantage of analysing SNPs rather than STRs is that they can be analysed in short DNA segments, which means that they are particularly useful for the testing of highly degraded samples, such as old, dry bones.

181.
Short nucleotide insertions and deletions (*indels*) have proved to be a useful tool in forensic genetics. Like SNPs, *indels* are particularly effective for typing degraded samples, as this type of testing analyses short DNA segments.

3. Comparing the victim`s sample DNA profile against the reference sample database

182.
As explained above, there are three types of sample from which DNA profiles are obtained for DNA matching: (a) ante-mortem reference samples, such as those taken from personal items belonging to the victim, for direct matching with samples from the remains; (b) samples from blood relatives of the victim or missing person used to establish biological relationships; (c) post-mortem or forensic samples.

183.
When disappearances involve a large number of people, the amount of data to be stored and compared can be enormous. This includes DNA profiles obtained from post-mortem samples, family reference samples, ante-mortem samples from the missing person, details of the missing person and any other relevant information. It is important for all this data to be loaded onto computer media suited to this purpose.

184.
Once the DNA profile obtained from a sample has been analysed and confirmed, it should be uploaded using automatic systems to avoid transcription errors.

185.
While genetic laboratories should always attempt to generate complete DNA profiles for post-mortem samples, partial profiles can also be used for matching, although they are more likely to yield “false positive” or “false negative” results. In particular, partial DNA profiles obtained from post-mortem samples can be used for direct matching with ante-mortem reference samples from the victim (personal items, for example) or with other post-mortem samples for reassociation in the case of scattered or commingled skeletal remains.

186.
Although DNA testing can be performed by different laboratories, involve different teams of experts (anthropologists, dentists, pathologists, police, public prosecutors, investigators, etc.) and take place at a number of different locations, it is recommended to centralize information and data analysis to ensure consistency and quality, so that potential matches are not missed. It is also recommended that all the information be brought together within one agency or organization that is part of the investigation, gathering all the information according to the principles set forth in this Guide.

4. Statistical evaluation and interpretation of results

187.
When there is a strong presumption of the identity of the remains of a victim, specific reference samples can be selected for matching to confirm the identification of the remains. In cases of large-scale disappearances, when there are enormous amounts of genetic data to be stored and compared, the use of specifically designed computer programmes is recommended.

188.
The use of computer programmes with low stringency search options is recommended, so that no potential matches are missed.

189.
The profile matching software application should allow comparison of the DNA profile obtained from a post-mortem sample against reference samples from the victim's relatives (kinship analysis), against ante-mortem reference samples (direct matching) and against other post-mortem samples.

190.
In the case of large-scale disappearances in which people related to each other could be buried at the same site, the search options should be configured accordingly, with a view to grouping potential family members together and avoiding false associations and identifications.

191.
In some situations, it may be necessary to add the profile of a victim who has already been identified, to the reference sample database, so that it can be used to identify other biological relatives who are also missing. When there are two or more closely related victims from the same family group, it is important to factor this into the analytical programme, as the likelihood ratio (statistical probability of a true match) falls sharply for each individual identification. This means that if there are two or more biological relatives missing, it is more difficult (sometimes even impossible) to individually identify each one based on DNA matches with family members who donate samples. They might share lineage markers, such as mtDNA and/or Y-STRs, because they belong to the same family group. In some cases, it might prove impossible to distinguish between two biologically related people through DNA typing, unless there are direct biological samples left by the victim when alive (personal items, etc.) or samples from his or her offspring available for matching. The reconciliation of DNA results with findings by experts in other fields (anthropology, dentistry, fingerprinting, etc.) is especially recommended in such cases.

192.
A match between DNA profiles consistent with the reported genetic relationship does not necessarily mean that an identification can be made. The match must be mathematically evaluated. Once a match has been found between the DNA sample from remains and a reference sample or samples, statistical calculations must be made before reporting the

Sampling relatives in Bolivia

match, based on the criteria and material used for the comparison of the DNA profiles. The two basic approaches to DNA matching are explained below.

(a) Direct matching

193.
In direct matching, the DNA profile from a victim's post-mortem sample is compared to a profile developed from personal items left by the missing person prior to death or disappearance to determine whether they come from the same source. This method may be preferred over kinship matching, because it avoids the problem of inaccurate information about relationships and is statistically more powerful. It can be reported as a *random match probability* or preferably as *likelihood ratio*. However, it should be taken into account that false negatives can occur, resulting in an identification being missed. This can happen when the family inadvertently provides a reference sample that does not contain the missing person's DNA or as a result of the problems described in paragraph 184.

(b) Relationship index

194.
The *relationship index* (RI) is calculated based on the statistical analysis of DNA profiles obtained from post-mortem samples and reference samples taken from the missing person's biological relatives. The relationship index, also referred to as the *likelihood ratio* (LR), defines the statistical significance of the match between the DNA profiles of biological relatives in the family analysed and the remains sample under two hypotheses: the first hypothesis is that the remains sample is related to the family as reported, and the second hypothesis is that the remains sample is unrelated to the family.

(c) Other statistical considerations

195.
The Bayesian method is recommended for the calculation of statistical significance, combining non-DNA evidence and data (quantified into the prior odds) with the findings of the DNA analysis (likelihood ratio or relationship index) to compute the posterior probability of identification.

196.
The use of prior values and other ante-mortem data reduces the probability of error in the identification of a missing person. They are combined with the likelihood ratio to increase the reliability of the results in the identification. The prior probability value is obtained by quantifying all the non-DNA information ascertained prior to the inclusion of the DNA information and should be calculated by the identification committee, according to the circumstances of the case and taking into consideration the number of missing persons. This value should be reported to the genetics laboratory, so that it can be combined with the likelihood ratio computed in the DNA analysis to calculate the overall probability of identification (or posterior probability). The prior probability value is calculated by analysing the human remains and forensic evidence recovered to determine whether it is a case with a closed list (e.g. common grave with four skeletons and four prior presumptive identifications) or a case with an open list (no prior presumptive identifications). Prior probability will be easier to establish when the list is closed than when it is open. In closed incidents, the prior value is the inverse of the minimum number of human remains, that is, 1/n of individuals missing. Hence, the higher the number of potential missing persons, the lower the probability of making a correct identification, which means that more genetic markers or more family reference samples must be tested, or that ante-mortem reference samples left by the missing person, which have a high power of discrimination, must be obtained for testing.

197.
In incidents with open lists, the priors should be determined by analysing other factors to obtain the most accurate values (morgue records, police records, records of human rights organizations, cemetery records, civil records, military records, etc.). It is advisable to establish in advance the posterior probability threshold to be met before an identity match is reported, depending on the type of investigation being carried out. A posterior probability threshold of 99.9% is usually recommended for an identification to be considered valid (prior probability combined with the likelihood ratio = posterior probability). However, it should be taken into account that science and technology are constantly evolving, and new human identification methodologies developed in the future could permit higher probabilities than those currently used. The threshold for considering an identity match valid could therefore vary as technology improves, and should be agreed by all parties involved in the investigation.

198.
In cases involving the re-association of body fragments or skeletal remains commingled in mass graves, it is advisable to use direct matching of DNA profiles (fragment-to-fragment comparison) and the likelihood ratio or random match probability. When the established minimum threshold is met, the best of the profiles obtained from the re-associated fragments (belonging to the same person) should be selected for screening against reference samples from personal items belonging to the missing person or samples from biological relatives for kinship analysis.

199.
In large-scale screening that involves extensive databases containing information relating to evidence (remains) and references, special consideration must be given to the possibility of adventitious or random matches or false positives. It is therefore always advisable to combine prior values and reconcile DNA results with the findings of experts in other fields to increase the reliability of the identifications made.

(d) Population allele frequencies

200.
It is important to select the proper allele frequencies for the population group to which the missing person belongs. An incorrect population database can distort the significance of allele sharing, as common alleles in one population group may be rare in another. There may be very marked differences between population groups, especially with regard to lineage markers (Y-STRs and mtDNA). If allele frequencies are

not available for the population to which the missing person belongs, it is recommended to create a population database by analysing randomly selected, unrelated individuals from that population. There is a great deal of information on allele frequencies in different populations published in scientific journals of international renown, which can be used for matching.

201.
If necessary, calculations should be performed using various databases containing the most representative allele frequencies in the population to which the victims belong, particularly in the case of lineage markers (mtDNA and Y-STRs). The results of the most conservative estimations should be reported.

202.
Annex 3 provides a list of websites where population databases can be found for autosomal STR, Y-STR and mtDNA markers. In addition to the websites listed in Annex 3, there are many scientific publications presenting allelic frequencies in different populations.

5. Reconciliation of genetic data with information from other disciplines

203.
As mentioned elsewhere, it is very important to reconcile all ante-mortem (background investigations, fingerprint records, etc.), anthropological and genetic information before drawing a final conclusion and reporting the identification.

204.
The identification process must be carried out as a multidisciplinary effort. It is therefore recommended that the system designed for the exchange of information among the different experts and areas be as flexible and efficient as possible.

205.
The evident limitations of each individual discipline involved in the victim identification process will be minimized if available information is considered globally. This will also help to resolve problems of false positives and missed matches.

206.
It is recommended to establish an identification review board (or assign dedicated personnel), tasked with reviewing each case and coordinating with the different areas involved to organize the exchange of information obtained and determine the steps to be taken.

6. DNA results report

207.
The DNA results report must comply with certain basic standards and be consistent with international recommendations.

208.
The report should explain the results in a clear manner, so that they can be reviewed and interpreted by other specialists. The report should also present the conclusions in a way that allows them to be interpreted by other professionals involved in the project with no training in genetics, such as magistrates, judges and the victims' families. Terms used in forensic genetics are often misinterpreted by people unfamiliar with the jargon (e.g. in genetics "negative result" can be mistakenly used to indicate that it was not possible to obtain a DNA profile from the sample, although it is usually understood to mean that no match was made, that is, "exclusion of identity"). Special attention should therefore be paid to the terminology used in reports to prevent erroneous or ambiguous interpretations.

209.
It is recommended that the reports contain the following information:

a) description of the forensic material received and processed by the laboratory (barcode or other identifier, sample type, case number and chain of custody);
b) description of the reference samples against which forensic samples were screened (identifier, sample type, relationship to the victim, etc.) and any other sample-related information considered relevant;
c) means and methods used to analyse DNA: extraction, quantitation, kits used for STR profiling, sequencing, software applications used, etc.;
d) tables showing results (autosomal STR, Y-STR and mtDNA profiles, etc.);
e) population frequency databases consulted for comparison with the frequencies of the DNA profiles obtained;
f) statistical analysis (indicating LR, prior probability and posterior probability) and software applications used for profile screening;
g) results, interpretation and conclusions.

7. Confidentiality of genetic data

210.
Genetic information falls within the category of personal data that is regulated domestically and internationally by personal data protection laws and regulations.

211.
All DNA results (DNA profiles, electropherograms, raw data and any other genetic information produced) must therefore be treated as confidential. The use and disclosure of any such information must be restricted to the actors involved in the victim identification process (sample donor, institution responsible for the identification process, technical personnel, etc.).

212.
There must be a confidentiality agreement between the victim's relatives who provide samples and the institution responsible for the identification process (genetics laboratory, anthropology laboratory, institute of legal medicine, health authorities, etc.). It should be included as part of the consent form.

213.
The agreement must clearly and expressly state how the sample can be used in the testing process (DNA profiles developed for identification purposes only) and establish limits on access to the samples and profiles and their storage and custody. It must recognize the donors' right to access their profiles and establish limitations on access by third parties.

214.
If population databases need to be developed (to establish allele frequency parameters required for mathematical calculations), with the inclusion of the DNA profiles of the donor relatives, the samples and profiles can only be used for the publication of anonymous allele frequency data in scientific journals. In such cases, written consent must be obtained from the relatives in advance.

8. Special situations in the identification of the remains of missing persons

215.
In the identification of victims of human rights violations, the remains are often highly fragmented and commingled. This can be due to the circumstances of death, burial or recovery. It is therefore not unusual to have to deal with complex cases in which it is not only necessary to identify the victims, but also to individually identify fragments, in order to be certain that the greatest possible proportion of the remains is returned to the family. Genetic profiling, combined with forensic anthropology, is a key tool in the task of reassociating body parts or fragments (generally skeletonised remains).

216.
In such cases, small body parts (hands, feet or ribs) or fragmented remains may not be suitable for DNA analysis, because they would be destroyed in the process required to develop the profile. This means that once the victims have been identified, there may be biological material that it has not been possible to re-associate with a particular set of remains.

217.
Once the case is closed and no further analysis is to be performed, the situation should be clearly explained to the family, with the permission of the institution responsible, and a joint decision taken on what is to be done with the commingled remains or fragments that could not be re-associated (e.g. collective burial).

218.
When victims have been identified, but there is a possibility that more remains may be discovered in the future, the situation should be clearly explained to the family, with the knowledge and permission of the institution responsible, so that they can decide whether they prefer to receive the remains identified up to that point for burial (and bury any further remains that might be discovered at a later date) or whether they prefer to wait until the process is considered to have reached an end (even though it might take years).

219.
In cases involving highly fragmented or incomplete remains, it may be that the only piece of biological material that can be identified as belonging to the victim is destroyed during the DNA testing performed to obtain the profile. In such cases, in addition to explaining the situation clearly and unambiguously to the family and the institution responsible, it is recommended that an, albeit symbolic, tangible artefact of the missing person's remains be returned, for example, the residue of the bone powder used in the extraction, leftover DNA extract, etc.

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VI. TERMINOLOGY

A. Definitions given in the UNESCO International Declaration on Human Genetic Data

- **Biological samples:** any sample of biological material (for example blood, skin and bone cells or blood plasma) in which nucleic acids are present and which contains the characteristic genetic make-up of an individual.
- **Consent:** any freely given specific, informed and express agreement of an individual to his or her genetic data being collected, processed, used and stored.
- **Cross-matching:** matching of information about an individual or a group contained in various data files set up for different purposes.
- **Data irretrievably unlinked to an identifiable person:** data that cannot be linked to an identifiable person, through destruction of the link to any identifying information about the person who provided the sample.
- **Data linked to an identifiable person:** data that contain information, such as name, birth date and address, by which the person from whom the data were derived can be identified.
- **Data unlinked to an identifiable person:** data that are not linked to an identifiable person, through the replacement of, or separation from, all identifying information about that person by use of a code.
- **Invasive procedure:** biological sampling using a method involving intrusion into the human body, such as obtaining a blood sample by using a needle and syringe.
- **Non-invasive procedure:** biological sampling using a method which does not involve intrusion into the human body, such as oral smears.
- **Human genetic data:** information about heritable characteristics of individuals obtained by analysis of nucleic acids or by other scientific analysis.

B. Technical terminology

- **Adventitious match:** in direct matching, it is when the sample from the victim shares, by chance, the same DNA profile as a person other than the one from whom the sample was taken. In kinship analysis, it is when a victim sample is matched to a family group by chance and not because they are biologically related. It is indirectly defined as a false positive, as it is a coincidental match which can result in incorrect identifications. The bigger the databases in which the search is made, the greater the possibility of adventitious matches and false positives.
- **Allele:** one of the different forms of a gene. While the genetic markers commonly analysed in human identification are located in non-coding regions, in forensic genetics, the term allele is commonly used to define this type of sequence variation in non-coding regions of DNA.
- **Allelic dropout:** it occurs when an allele cannot be visualized in a DNA profile in a given sample. It is frequent in DNA extracted from samples with a small quantity of DNA, poor quality DNA or degraded DNA. It can lead to erroneous conclusions in the interpretation of the results of genetic testing of degraded samples, as a heterozygous specimen can appear to be homozygous.
- **Amelogenin:** a gene located on the sex-chromosome pair (X and Y). In forensic genetics, it is analysed to determine sex in any type of forensic sample.
- **Autosomal markers:** markers located on autosomal chromosomes.
- **Discontinuous family tree:** a family tree in which there is a gap between two generations. In a case in which the parents are missing, the grandparents and the grandchildren are an example of a discontinuous family tree, because the generation between them is missing.
- **DNA:** the DNA molecule consists of two long chains of nucleotides coiled into a double helix. DNA contains the genetic information needed for the development and functioning of cells in all living organisms.
- **DNA extraction method:** any method used to purify DNA from a biological sample, such as blood, saliva, semen, hair, bone, used garments, etc.
- **DNA or genetic profile:** in forensic genetics, it is an encrypted set of numbers reflecting the genetic make-up of an individual for the specific genetic markers analysed. Each genetic marker analysed can be expressed as two numbers (e.g. 8-10) in the case of heterozygosity or one number (e.g. 8) in the case of homozygosity.
- **DNA quantitation method:** any method used to determine the quantity of DNA in a given biological sample.
- **Electropherogram:** graphic representation of the results of DNA profiling or DNA sequencing.
- **Extranuclear DNA:** the DNA not located inside the cell's nucleus. In general, it refers to mitochondrial DNA.
- **Genetic markers:** a DNA segment with a known physical location in the genome or on the chromosome and with a known inheritance pattern that can be tracked.

- **Genetic polymorphism:** a form or variation of a particular DNA sequence in a gene or DNA segment which can exist among the individuals of a population or species.
- **Genetic recombination:** the process by which a DNA sequence is cut and then rejoined to a different DNA molecule.
- **Haploid genetic markers in genealogy:** DNA markers or sequences passed on to offspring by just one of the parents. For example, mitochondrial DNA is passed on to offspring by the mother only.
- **Heterozygosity:** condition of an individual having two different alleles (one from the father and one from the mother) of a certain gene or genetic marker.
- **Homozygosity:** condition of an individual having two identical alleles (one from the father and one from the mother) of a certain gene or genetic marker.
- **Human genome:** all the genetic information contained in human DNA.
- **Likelihood ratio (LR):** it is a logical way of presenting the results of DNA analysis, as it takes into account two mutually exclusive hypotheses. For example, a) the remains sample belongs to a particular missing person or b) the remains sample does not belong to that missing person and therefore belongs to another person. The ratio is the quotient of these two opposing possibilities. In forensic genetics, the LR is used to compare the significance or strength of a DNA match.
- **Microsatellites (or STRs):** regions of DNA with adjacent short sequences repeated a certain number of times, which can vary from one individual to another. These repetitive stretches of short DNA sequences are like train wagons hitched in tandem, which is why they are called variable number tandem repeats. In forensic genetics, microsatellites or short tandem repeats (STRs) are the markers most commonly used in human identification.
- **Mitochondrial DNA:** circular DNA found inside structures called mitochondria located in the cell's cytoplasm.
- **Monozygotic twins:** also known as "identical twins", because they come from one egg which splits early on in development to produce two genetically identical individuals. Unlike monozygotic twins, dizygotic twins or "non-identical twins", although resulting from the same pregnancy, come from two separate eggs independently fertilized by two different sperm cells, so that genetically they are full siblings.
- **Mutation:** change in a DNA sequence in the genome which can be passed on to offspring. In forensic genetics, mutations must be taken into account, because otherwise the interpretation of DNA data can lead to incorrect results.
- **Nuclear DNA:** the DNA found inside the cell's nucleus. Nuclear DNA is arranged and packaged on chromosomes; there are 22 pairs of autosomal chromosomes and one pair of sex chromosomes, making a total of 23 pairs.
- **Nucleotide sequence:** the arrangement or sequence of nucleotide bases (C, G, A or T) in a segment of DNA or RNA (ribonucleic acid).
- **PCR inhibitors:** any substance that inhibits DNA amplification by polymerase chain reaction (PCR).
- **Polymerase chain reaction (PCR):** it is an enzymatic process in which a specific DNA sequence is replicated or copied thousands of times. PCR can be likened to a "molecular photocopier" of DNA sequences, producing thousands of identical copies of the target DNA sequence.
- **Primer:** a short synthetic DNA segment used to initiate and continue a polymerase chain reaction (see PCR).
- **Random match probability:** measures the probability that an individual chosen at random from the general population will have the same genetic profile as a forensic sample.
- **Relationship index:** term used to refer to the likelihood ratio (LR) in a biological relationship test.
- **Short tandem repeats:** see microsatellites (or STRs).
- **Single nucleotide polymorphism (SNP):** a variation in a DNA sequence that affects a single nucleotide base (C, G, A or T) of a sequence in the genome.
- **SNPs:** see single nucleotide polymorphisms.
- **Spongy bone:** unlike compact bone, this type of bone does not contain osteons; the interstitial lamellae are irregularly arranged forming a lattice of thin columns of bone called trabeculae. It is found in the epiphyses (or ends) of long bones, in vertebral bodies, the sternum, pelvis, kneecap, etc.
- **STRs:** see microsatellites (or STRs).
- **Stutters:** amplified products with fewer repeats (normally one or two fewer) than the true allele; they can also have more repeats (one or more) than the true allele, although this is less common.
- **X-STRs:** microsatellites or STRs located on the X sex chromosome.
- **Y-SNPs:** single nucleotide polymorphisms (SNPs) located on the Y sex chromosome.
- **Y-STRs:** microsatellites or STRs located on the Y sex chromosome.

VII. ANNEXES

ANNEX 1

Tables

POPULATION FREQUENCY DATABASES FOR GENETIC MARKERS USED IN FORENSICS		
STRs	STRbase	www.cstl.nist.gov/biotech/strbase/
	ENFSI	www.str-base.org/index.php
	ALFRED	http://alfred.med.yale.edu/alfred/index.asp
Y-Chromosome haplotypes	Y-HRD.ORG	http://www.yhrd.org/
	Y-filer Haplotype database	http://www6.appliedbiosystems.com/yfilerdatabase/
mtDNA haplotypes	EMPOP	www.empop.org
	Mitomap	www.mitomap.org

Software solutions designed to provide DNA database management for forensic identification purposes and/or statistical evaluation of the significance of matches found by screening the DNA from unidentified human remains against DNA from reference family members.

(DNA-DATABASE MANAGEMENT REVIEW AND RECOMMENDATIONS, ENFSI (European Network of Forensic Science Institutes) DNA Working Group, August 2012, http://www.enfsi.eu/page.php?uid=98)

PROGRAM NAME	PRODUCER	REFERENCE
Anonymous	Petr Linhart	http://www.fsigenetics.com/article/S1872-4973(08)00182-8/abstract
Bonaparte	SNN/Smart Research	www.bonaparte-dvi.com
CODIS 6.0	FBI	http://www.fsigenetics.com/article/S1872-4973(10)00105-5/abstract
DNAStat	Jaroslaw Berent	http://www.ncbi.nlm.nih.gov/pubmed/17907628
DNAView	Charles Brenner	http://dna-view.com/
EasyDNA	Wing Kam Fung	http://www.hku.hk/statistics/EasyDNA/
EasyPat	Michael Krawczak	http://www.uni-kiel.de/medinfo/mitarbeiter/krawczak/download/
Familias	Petter Mostad	http://www.math.chalmers.se/~mostad/familias/
FINEX	R.G. Cowell	http://www.ncbi.nlm.nih.gov/pubmed/12850417
FSS DNA Lineage	FSS	http://www.promega.com/geneticidproc/ussymp21proc/abstracts/poster_89.pdf
Genoproof	Qualitype	http://qualitype.de/genoproof/
Genotype	Kvant s.r.o.	http://www.dip.sk/typo3/dip.sk/index.php?id=9&no_cache=1&L=1
Grape	DNA-soft	http://www.DNA-soft.com
Hugin	Hugin	http://www.hugin.com/productsservices/demo/hugin-lite
M-FISys	Gene Codes Forensics	www.genecodesforensics.com
MPKin	Institute of Investigative Genetics	www.investigativegenetics.com/content/1/1/8
PatCan	Jose Antonio Riancho	Forensic Science International Volume 135, Issue 3, 27 August 2003, Pages 232-234
Patern	Michael Krawczak	http://www.uni-kiel.de/medinfo/mitarbeiter/krawczak/download/
Paternity Index	Michel Jung	FSI Genetics Volume 3, Issue 2, March 2009, Pages 112-118
PatPCR	Juan Antonio Luque	http://www.fsigenetics.com/article/S1872-4973(08)00182-8/abstract

ANNEX 2

Useful websites

STRBase: a short tandem repeat DNA database for the human identity testing community. Ruitberg C.M., Reeder D.J., and Butler J.M. (2001) Nucleic Acids Research 29, 320–322. (http://www.cstl.nist.gov/div831/strbase/)

DNAAdvisory Board (2000) Statistical and population genetic issues affecting the evaluation of the frequency of occurrence of DNA profiles calculated from pertinent population databases. Forensic Science Communications 2 (http://www.fbi.gov/hq/tab/fsc/backissu/july2000/dnastat.htm)

Interpol:
www.interpol.int/Public/Forensic/DNA/

Federal Bureau of Investigation. FBI Laboratory:
http://www.fbi.gov/hq/lab/labhome.htm

Federal Bureau of Investigation. CODIS Information:
http://www.fbi.gov/news/testimony/the-fbis-codis-program/

Association of Chief Police Officers of England, Wales and Northern Ireland (National DNA Database reports):
https://www.gov.uk/government/publications/ndnad-annual-report-2011-to-2012

GeneWatch UK:
www.genewatch.org

American Association of Blood Banks (2004) Annual report summary for testing in 2004:
www.aabb.org/content/Accreditation/ParentageTestingAccreditation Program/ptprog.Htm

ENFSI – European Network of Forensic Science Institutes:
http://www.enfsi.org/

FSS – The Forensic Science Service (UK):
http://www.forensic.gov.uk/
http://www.forensic-science-society.org.uk/

ISFG – International Society for Forensic Genetics:
http://www.isfg.org/

GEHP-ISFG – Spanish and Portuguese Speaking Working Group of ISFG:
http://www.gep-isfg.org/ISFG/English/portada.php

SNPforID Project:
http://spsmart.cesga.es/snpforid.php

NIFS – National Institute of Forensic Science Australia:
http://www.nifs.com.au/home.html

Forensic Mathematics. Contains information of the kinship software with DNA-ViewTM and articles/discussions focused

on the statistical/mathematical interpretation of DNA profiles:
http://dna-view.com

ENFSI DNA WG STR Population Database. Calculates the profile frequency of a SGM Plus profile using 24 European allele frequency databases:
http://www.str-base.org/index.php

ChrX-STR.org:
http://www.chrx-str.org/

popSTR
http://spsmart.cesga.es/popstr.php

ANNEX 3

SIMPLIFIED GUIDE FOR VICTIMS AND THEIR FAMILIES

This guide is intended for victims of serious human rights and international humanitarian law violations and their families, and aims to help them understand the DNA-based human identification process.
There are international conventions and protocols which require signatory countries to adopt measures to prevent forced disappearances, identify missing persons whether dead or alive and investigate the causes and circumstances of their death or deprivation of identity. This requires political will on the part of governments and the active involvement of victims' families. This guide answers the questions most frequently asked by people searching for a missing family member.

In what way can I help forensic institutions and facilitate the process to identify my missing family member?
In cases involving human remains, the identification process is based on a technical comparison of the findings of the analysis of the remains and information about the missing person provided by the victim's family or acquaintances. The forensic team usually interviews family members to obtain this information and asks them to provide records and evidence to facilitate identification, such as X-rays, photographs and personal items belonging to the victim. Often there are files containing photographs of unidentified bodies which you can look through for your missing family member. It is also important for the most informative family members to be selected for DNA-based identification.
In cases in which the missing person is found alive with a different identity, his or her DNA is compared to that of putative family members. If they give their consent, biological samples (drops or blood or saliva) taken from family members can be used as reference samples for comparison with unidentified human remains.

Are there other ways of identifying missing persons in addition to DNA typing? Are they as reliable?
Yes. Before performing DNA analysis, forensic experts normally exhaust all other methods of identification, including:

1. Fingerprint analysis. Although this is one of the fastest and most conclusive ways of identifying a body, it can only be used when the bulb of the finger is intact. Fingerprinting is therefore not an option in the case of skeletonised remains.
2. Dental examination is useful if the victim's dental records are available. Often this ante-mortem information is not available, however, and even when teeth are recovered with the remains, they might not be useful for identification purposes.
3. A forensic anthropological study yields information on the height, age, sex and ancestral pattern of the remains, providing valuable pointers, although they are not generally sufficient to report a positive identification.
4. Lastly, the findings of the medico-legal autopsy, such as signs of previous fractures, bone deformities, tattoos, surgical implants, etc. and the description of personal items and clothing found on the body can also be checked against information supplied by the family. Such findings can often provide direct evidence to make at least a presumptive identification, which can then be corroborated using other techniques.

What is DNA testing?
As mentioned above, DNA typing for human identification involves comparing the DNA profile of the remains against the DNA profiles of possible relatives of the missing person or against the DNA profile developed from the victim's personal items or biological samples stored in medical facilities. In the case of missing persons who are found alive (abducted children, etc.), the DNA profile of the person whose identity is in doubt is compared against those of possible relatives.

What does DNA analysis involve?
DNA is the molecule containing hereditary information present in all cells in the human body. They all carry the same genetic information, which means that it does not matter which part of the body the sample used in the study is taken from. For convenience, in the case of living donors, blood or saliva samples are preferred, as just a few drops of blood or cells gently scraped from the inside of the cheek with a cotton swab are sufficient for testing. In the case of skeletal remains, DNA can be preserved and analysed years after death.
There are two types of DNA in cells: nuclear DNA and mitochondrial DNA. Both can be used for human identification. Nuclear DNA is inherited from both parents, half from the father and half from the mother (biparental markers). Specific variations in the DNA molecule are determined and compared with the variations found in possible family members, as explained in greater detail below.

The Y chromosome is found in the nuclear genome of males. It is a structure formed by DNA specific to males and is therefore used to identify samples from men. The Y chromosome is passed on, generally intact, from father to son (uniparental marker) and is shared by all the males in the family with a common male ancestor. It is useful in relating an individual to a specific paternal lineage but, unlike biparental nuclear markers, this uniparental marker does not enable an individual identification to be made; it just links an individual to a particular family group which, in the case of missing persons investigations, can be very useful, especially when there are no first-degree relatives available for testing.
Mitochondrial DNA, like the Y chromosome, is a uniparental marker, but is passed on by the mother to all her offspring regardless of sex. It can be used when only family members from the maternal line are available for testing, even if they are only distantly related, for example, a female cousin (daughter of the victim's aunt), the victim's brother (son of the same mother), etc.

What is a DNA profile?
It records the DNA sequence variants detected in a person's DNA analysis. This information is practically unique to the individual. It is produced by examining DNA regions that are highly variable from one individual to another. In the case of nuclear DNA, two components (called alleles) are identified for each of them: the variant inherited from the father and the variant inherited from the mother. They are represented as pairs of numbers. DNA profiles normally used in forensic genetics take into account around 15 pairs of numbers. Once the DNA profiles of the putative family members have been obtained, it can be determined whether they are related to the missing person. If, for example, the son has variants 6 and 9, the mother must have passed on one of the two numbers to him and the father the other. The parents, in turn, have their own pair of variants, one of which must coincide with one of the son's. For example, the father of the son 6/9 may be 6/10 and the mother 9/11. The biparental markers of the nuclear genome can be used to identify people with a high degree of confidence.

Is any family member suitable for DNA matching?
First of all, it is absolutely essential for the person providing the sample to be biologically related to the missing person. Samples from stepfathers, stepmothers, adoptive parents or foster families in general are of no use. It is also important to specify the exact nature of the relationship between each sample donor and the missing person. Any uncertainty should be reported.
In general, relatives within the first degree of consanguinity are most useful for DNA matching; the mother, the father and the offspring of the missing person are the first choice. They are followed by siblings of the missing person. In this case, it must be specified whether they are full siblings sharing the same mother and father or half-siblings. A third alternative is the paternal and/or maternal grandparents. Lastly, the DNA of other relatives from the paternal lineage or the maternal lineage of the missing person can also provide useful information.

What sources of DNA can be used for matching with the victim's DNA?
When the missing person is found dead, DNA from the skeletal remains can be compared with DNA obtained from:

1. deceased or living relatives of the missing person;
2. personal items used by the missing person before death, such as toothbrushes, razors, hairbrushes and combs, etc.;
3. medical samples taken when the victim was alive and kept in hospital archives; pathology samples, such as biopsies or smears, may have been collected from the victim and stored in medical facilities.

Skeletal remains can be directly matched to the samples listed above or screened against DNA databases stored in genetic data banks.
When missing persons are found alive, the DNA obtained from a blood or saliva sample from the victim or personal items belonging to him or her is compared with that of the putative family members or, as in cases involving skeletal remains, screened against databases containing the DNA profiles of relatives of missing persons.

What kind of DNA samples are collected from deceased victims?
This depends on the state in which the body is found. When a body is found within hours of death, it is possible to take post-mortem blood samples or small fragments of soft tissue when no decomposition has taken place.
In the case of completely skeletonised remains, the samples collected for testing are sections of bone or whole teeth.

Why is DNA analysis sometimes not possible?
As mentioned above, some degree of degradation is expected in skeletal remains. Today, however, thanks to technological advances, it is often possible to obtain useful and reliable genetic information even when the sample is degraded.
Nevertheless, the genetic material present in a fragment of bone may be so severely degraded that it cannot be analysed. In such cases, the forensic expert tests the bone again or tests other bone samples taken from the same skeleton and is sometimes able to develop a DNA profile suitable for matching purposes. There are, however, situations in which it proves impossible to generate a DNA profile, and no comparison can be made with reference samples. It is important to note that a negative result indicates that the study did not yield results and not that the remains do not belong to the missing person. When this happens, the competent judicial authority should assess non-DNA evidence in the case before taking a decision on the identity of the remains.

How long does the process take?
The time it takes to identify a body depends on the circumstances of the DNA analysis and on other unrelated factors.
The time it takes to make an identification can vary greatly. Sometimes it can take months or even years, depending on the steps taken by the authorities to exhume or retrieve the remains of the missing person.
Once the skeletal remains are in the laboratory, the time required varies depending on how long it takes to locate relatives of the missing person and collect the necessary reference samples. This is one of the most complex stages of the process, because the families of victims are often afraid or wary about being interviewed and having a sample taken for the study.

Once the laboratory has all the elements it needs to proceed with matching (skeletal remains and reference samples), the time it takes to analyse the human remains depends on the following factors:

1. The laboratory's capacity in terms of throughput and the demand for forensic identification services in the country in question
   In countries where the demand for human identification testing is high, the workload may overwhelm the capabilities of available laboratories. Cases are dealt with in order of arrival, which means that a body may remain in storage for several weeks or even months before the laboratory starts testing. If there is no backlog, full DNA testing can be completed in less than a week under ideal conditions. There are, however, other factors unrelated to the laboratory's throughput capacity and speed, such as the individual characteristics and state of preservation of the skeletal remains to be analysed, which can also affect the time required to complete the process.
2. Individual characteristics and state of preservation of the skeletal remains to be analysed
   The state of preservation of DNA in human remains can vary greatly. Certain types of tissue with large quantities of bone cells are more likely to have a sufficient concentration of DNA for analysis. It is not, however, only the quantity that matters; the degree of DNA degradation can also affect the results and therefore the time it takes to complete the process. A sample containing few cells and highly degraded DNA is unlikely to yield useful results for the identification study. In such cases, the procedure has to be repeated, and it may prove necessary to use additional bone samples. This can take the laboratory several months to complete.
3. Sometimes only people distantly related to the victim are available to provide reference samples for the genetic study. There may still be a high degree of uncertainty about the identity of the body after the genetic study has been completed. In such cases, it is necessary to look for other more informative relatives, which can take several months. It should be noted that the authorities may, at any time during the process, request permission to exhume a member of the victim's family with a higher degree of consanguinity, for example, the victim's mother, in order to take a DNA sample as a reference for matching.

What is a genetic data bank?

It is an electronic archive used to store the DNA profiles of unidentified skeletal remains, living persons whose identity is in doubt (blood or saliva samples) and relatives of missing persons. They can also contain DNA profiles developed from the missing person's personal items and samples stored in medical archives.

The information is uploaded to the databases of the genetic data bank. The system is programmed to compare the files and detect matches between the DNA profiles of relatives and unidentified remains or living persons whose identity is in doubt. It is important to note that the genetic data bank provides pointers for potential matches, but does not make identifications on its own. Each apparent match detected in the search must then be confirmed by experts at the laboratory before reporting a positive genetic identification of the missing person and returning the remains to the family or re-establishing the true identity of a living person.

The relatives of victims of forced disappearance have the right to be included in the genetic data bank. The authorities and the forensic institutions responsible for managing the data bank must arrange for and facilitate the inclusion in the data bank of the DNA profile of any relatives of missing persons who so wish. When required, the State must also make the necessary arrangements to collect samples from people living abroad and have them sent to the forensic laboratory responsible for the identification process.

What information am I entitled to when I provide a sample for the genetic data bank?

You can request information from the competent authorities about where your biological sample is being stored, whether it has been used in searches, whether a positive match has been made and whether it has been destroyed once the process to identify your missing family member has been completed.

You are also entitled to receive a copy of your DNA profile if you wish.

Can I ask for my sample to be removed from the data bank once my relative has been identified?

Yes, once your relative has been identified, you can ask the competent authority to instruct the data bank administrators to destroy your biological sample, any elements generated during processing and the DNA profile that the data bank used for the identification.

In legal cases in which a criminal investigation has been initiated to establish criminal liability, it is advisable not to remove your profiles and samples from the data bank until the proceedings are over.

If the case is not part of a criminal investigation, you can have your sample/profile eliminated from the data bank if you so wish, even when your missing family member has not been identified, provided that the authority responsible permits such elimination in legal cases.

Can anyone else use my DNA profile for purposes other than the identification of my missing family member?

In some cases, there may be social security companies, health insurance companies or legal authorities with an interest in using your DNA profile for investigations other than enforced disappearance.

It is very important for the consent form signed with the forensic institution that collects the sample to specifically include limitations on use, with a view to protecting biological samples and any genetic information obtained from them.

Can I participate in the identification process?

Yes, in addition to collaborating in the process by providing samples, the families of missing persons can also participate as observers in the identification process, taking an active part in meetings at which progress reports are presented in order to ensure transparency, impartiality and thoroughness in the forensic investigation.

The information that family members can provide about the missing person (physical description, medical history, details about the disappearance, photographs, etc.) is just as important to the identification process as the biological sample.

Can the genetic study reveal unexpected family relationships?
Yes, the genetic study can detect family relationships that were not reported and reveal that people are not related in the way that they think they are. It is important to be aware that incomplete or inaccurate information about relationships can lead to false conclusions about the identity of a body. Most often, it results in a false exclusion, that is, it is wrongly concluded that the remains analysed are not those of the relative you are searching for.
These are situations in which errors can be made in DNA matching if the information recorded during the interview with the forensic expert is not clear and accurate. If you need to inform the forensic expert about any sensitive issues, you can request that the information provided be kept completely confidential. In this way, information about the family will not be made public in the geneticist's report. An agreement of this kind can be established in the consent form signed when the blood sample is collected for the genetic study.

Could the genetic study be wrong?
Yes, any forensic study is subject to error, and there have been cases of laboratories reporting errors in identification testing. This can be due to the incorrect handling of the samples before shipment to the laboratory, particularly in large-scale processes when there is a large number of victims involved in a single incident, or to problems caused by poor quality DNA results when samples are highly degraded, which can lead to an erroneous interpretation, such as a false exclusion. Laboratory analysts are aware of this risk and implement increasingly stringent process and product control systems. All genetic studies must be reviewed by a second analyst before an identification is reported. Furthermore, the tendency now is for a decisions committee to discuss the DNA results in the light of the findings of the other forensic disciplines, with a view to evaluating the coherence of the studies of the different studies as to the identity of the body. For the past ten years, forensic genetics laboratories have been implementing quality assurance and management systems, and many of them are accredited and certified to international quality standards, which means that quality control tools are incorporated into methodological processes at all levels.

Will all the skeletal remains of my family member be DNA typed?
No, DNA testing is performed using just one or two pieces of bone selected by a forensic anthropologist or forensic pathologist, following individualization of the skeletal remains. It is not necessary to carry out DNA testing on all the body's 206 bones. If the bone samples tested yield a positive identification, the body will be considered to have been identified. If the result is exclusion, this also applies to the whole body.

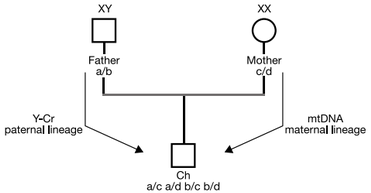
If a disease is detected when my DNA profile is examined, will I be automatically informed or can I ask not to be told?
In general, the genetic markers used for the purpose of forensic identification do not reveal the presence of a disease or a genetic predisposition to a particular disease. Such information is present in what are known as coding regions of DNA, while forensic testing uses non-coding regions. There are, however, some markers routinely used to determine the sex of a set of remains which do detect genetic abnormalities. Such findings put analysts in a difficult position, because they give them information that falls outside the scope of their duties as forensic experts. The consent form signed to authorize the taking of the biological sample should therefore specify whether you, as the sample donor, would prefer to be informed in such an event or whether you would prefer not to be told.

Can I receive psychosocial care during the identification process?
Yes, the families of victims require psychosocial care not just when the remains are returned, but throughout the whole process, including the search, recovery and identification of the missing person. It should also include guidance on collateral aspects of the disappearance affecting their lives, such as their civil and legal situation, financial matters and forced displacement. If you wish, you can apply to the institution responsible for referral to psychosocial specialists, which should have a mechanism in place for this purpose.

ANNEX 4

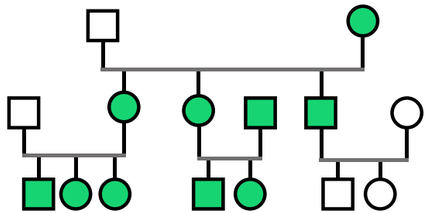
FIGURES

FIGURE 1
Father-to-child inheritance of genetic markers



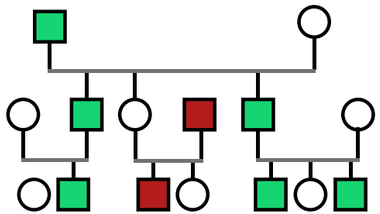
Autosomal chromosomes are represented as a,b,c and d. Autosomes are passed on in part from both parents to offspring and proportionally from all ancestors. Transmission of the four a,b,c, d autosomes are independently assorted as a/c, a/d, b/c or b/d in offspring.
Sexual pair is depicted as XX (mother) and XY (father).
Y-Chromosome is passed on complete from fathers to sons (paternal lineage). In the figure, the children (Ch) inherits complete the same Y-Chromosome than his father.
mtDNA is passed on complete from mother to all children, but only daughters will pass on complete again (maternal lineage) to the offspring. In the figure, Ch inherits the same mtDNA haplotype from his mother.

FIGURE 2
Pattern of maternally inherited mitochondrial DNA



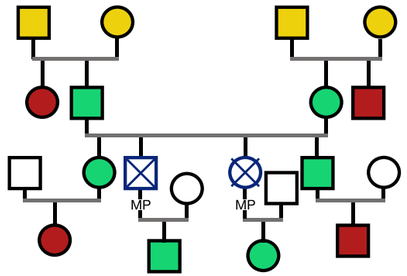
Green color indicates individuals sharing same mtDNA

FIGURE 3
Paternally inherited Y-chromosome



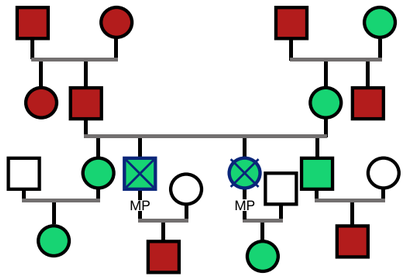
same colour represents male individuals sharing the same Y-haplotype

FIGURE 4
Selection of family members for the collection of reference samples for nuclear autosomal dna analysis



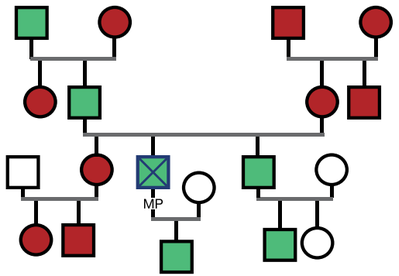
Colour coding is used to indicate how informative the samples are. Green: good. Yellow: moderate. Red: poor. MP indicates the missing person.

FIGURE 5
Selection of family members for the collection of reference samples for mitochondrial dna analysis



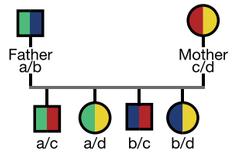
Colour coded as follows. Green: family member suitable for comparison. Red: family member not suitable for comparison. MP indicates the missing person.

FIGURE 6
Selection of family members for the collection of reference samples for Y chromosome analysis



Colour coded as follows. Green: family member suitable for comparison. Red: family member not suitable for comparison

Figure 7
Parents to children autosomal inheritance pattern



each parent passes one of each homologous allele pair to each child

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