ITMO Santé publique

Le génome, Janus de la biologie
Entre patrimoine et individu

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& plateforme « Genotoul societal »

Séminaire “le gène dans tous ses états”; cycle “Les sciences comme biens communs”, Centre d’Alembert, Université Paris Sud, 12 février 2014

Janus….

- **Janus:** divinité romaine, dieu des commencements et des fins, des choix, des clés et des portes.
- Dieu de premier rang il est invoqué avant toutes les autres divinités.
- En tant que dieu introducteur il est un « dieu des portes » qui préside à l'ouverture de l’année et à la saison de la guerre.
- Janus a un double visage parce qu'il exerce son pouvoir sur le ciel, sur la mer comme sur la terre.
  - Wikipedia, 10/2/14

Janus….

- Dieu ambivalent à deux faces adossées, d'origine indo-européenne, *Janus*, créateur débonnaire, devint le dieu des portes, des transitions et des passages, marquant l'évolution du passé à l'avenir, d'un état à l'autre, d'une vision à l'autre.
- Il préside aux commencements, il intervient au début de chaque entreprise
- Son double visage signifie qu'il surveille aussi bien les entrées que les sorties, qu'il regarde aussi bien l'intérieur que l'extérieur, la droite que la gauche, devant et derrière, le haut et le bas, le pour que le contre. Il est la vigilance et peut-être l'image d'un impérialisme sans limite.

Source : Institut français du design

Génome

- Le **génome** est l'ensemble du matériel génétique d'une cellule, d’un individu, ou d’une espèce.
- Aujourd’hui, l’espèce humaine et chacun des individus qui la composent et presque chaque cellule de chaque individu

Quelques caractéristiques « du » génome

- Un ensemble de « doubles facettes »
  - Appartenance à l ’espèce (même structure du génome)
    - « Génome humain, « patrimoine » de l ’humanité »
  - Caractère unique de l ’individu (combinatoire)
    - « Carte d ’identité génétique, empreinte »
  - Elément du corps le plus privé et le plus partagé

Plan

- Génome patrimoine
- Génome hérité, partagé
- Génome permanent
- Génome ouvrant sur le passé
- Génome élément du corps
- Génome protégé
- Génome donnée de santé exceptionnelle

- Génome individu
- Génome personnel
- Génome évoluant
- Génome ouvrant sur l'avenir
- Génome information
- Génome exposé
- Génome donnée personnelle ordinaire

Génome patrimoine/individu

- Déclaration de l’UNESCO sur le génome humain et les droits de l’homme (1997)
- Préambule :
- « Reconnaissant que les recherches sur le génome humain et leurs applications ouvrent d'immenses perspectives d'amélioration de la santé des individus et de l'humanité tout entière, mais soulignant qu'elles doivent en même temps respecter pleinement la dignité, la liberté et les droits de l'homme, ainsi que l'interdiction de toute forme de discrimination fondée sur les caractéristiques génétiques, »

Déclaration de l’UNESCO sur le génome humain et les droits de l’homme (1997)

- Art. 1 « Le génome humain sous-tend l'unité fondamentale de tous les membres de la famille humaine, ainsi que la reconnaissance de leur dignité intrinsèque et de leur diversité. Dans un sens symbolique, il est le patrimoine de l'humanité. »
- Unité, dignité, diversité, patrimoine

Déclaration de l'UNESCO sur le génome humain et les droits de l'homme (1997)

- Art. 2
- « Chaque individu a droit au respect de sa dignité et de ses droits, quelles que soient ses caractéristiques génétiques.
- Cette dignité impose de ne pas réduire les individus à leurs caractéristiques génétiques et de respecter le caractère unique de chacun et leur diversité. »

Déclaration de l’UNESCO sur le génome humain et les droits de l’homme (1997)

- Art. 3
- « Le génome humain, par nature évolutif, est sujet à des mutations. Il renferme des potentialités qui s'expriment différemment selon l'environnement naturel et social de chaque individu, en ce qui concerne notamment l'état de santé, les conditions de vie, la nutrition et l'éducation. »
- Art. 4
- « Le génome humain en son état naturel ne peut donner lieu à des gains pécuniaires »

Génome « bien commun »? Science génomique « bien commun »?

Plan

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- Génome information
- Génome exposé
- Génome donnée personnelle ordinaire

Quelques articles/dossiers récents

- **"Ce que prédisent nos gènes"** Journal du CNRS. Juillet-Août 2013.
- **Science & Santé Mai 2013 ; Médecine personnalisée : les promesses du sur-mesure**

http://www.inserm.fr/actualites/rubriques/actualites-societe/medecine-personnalisee-les-promesses-du-sur-mesure

- Réflexion en cours à l'OPECST (2 auditions publiques en 2013; compte-rendus disponibles)
- Accessibles en ligne. Exemple cancer du sein.

La médecine personnalisée

- Importance du mot,
- L’expression peut devenir slogan,
- L’avantage et le danger : susciter l’espoir, mais sans faire de fausses promesses
- Le niveau de preuve à établir
- La caractérisation de la tumeur, de la maladie par rapport à la « personnalisation »: médecine de précision, stratification
- La nécessité d’études d’impact en santé publique.

Prédiction, prévision, prévention et anticipation

- **Prévision** inclut la décision et l’action, pas seulement le «dire »
- La **médecine prédictive ou de prévision** n’inclut pas que la génétique (radiologie, imagerie, échographie, biomarqueurs)
- Cependant la génétique constitue un exemple type
- La **prévention** conduit à la mise en place d’actions pour empêcher qu’un risque se réalise, qu’une pathologie survienne
- La **médecine anticipative** conduit à la mise en place de moyens pour faire face le jour où un risque se réalise.
- La **médecine personnalisée** inclut
  - les précédentes dimensions plus, pour le cancer,
  - les actions médicales fondées sur la caractérisation fine
    - de la maladie (pour le cancer, des tumeurs),
    - de son évolution.
  - le ciselage de la prescription ou du traitement lui-

CAMBON-THOMSEN A. Médecine de prévision, génétique et santé publique. *l’Observatoire de la génétique* (N° 19, http://www.ircm.qc.ca/bioethique/obsgenetique/

Les biomarqueurs, éléments centraux de la médecine personnalisée

- Plusieurs catégories de biomarqueurs
  - Constitutionnels (ne changent pas; génétique)
  - Liés à l’évolution de la maladie/du traitement
- Panels de marqueurs
  - La question du choix, du moment, de l’information, de l’accompagnement
- Intégration avec d’autres éléments (épigénétique, environnement)

Information et personnalisation

- La constitution de groupes (stratification)
  - Paradoxe?
  - Certains laissés de côté?
  - Certains stigmatisés?
- Le paradoxe de la personnalisation et de la quantité massive d'informations
- La gestion personnalisée d'une information globalisée (changement d'échelle)
- La personnalisation vue à travers la caractérisation biologique peut aussi être perçue comme réductrice : importance du dialogue.

La prédictivité dans la médecine personnalisée

- La disponibilité d'informations plus ou moins prédictives pose :
  - la question de l'interprétation
  - la question de la responsabilité individuelle du patient vis-à-vis de la gestion de sa santé
  - la question de la disponibilité et de l'accès aux informations
- Dans le cas de tests établis l'enjeu des brevets pour l'accessibilité de ces tests

Des questions complexes

- « … la médecine s’engouffre dans cette prédiction en pensant que plus on prédira un risque, moins il aura de chances de survenir. Il existe une confusion permanente entre l’usage que l’on fera de cette prédiction et la prédiction elle – même. »

Pr. Didier Sicard, ancien président du CCNE

Expo « Médecine prédictive », Cité des sciences et de l’industrie, Sciences actualités, Paris, 2008

Avis 46 CCNE, "Génétique et Médecine : de la prédiction à la prévention"

Enjeux en génétique (1)

Ils sont liés :

1) à la dimension familiale de l’information génétique pas toujours facile à différencier de l’information à valeur seulement individuelle
2) à la co-existence de différents types de tests, à valeur médicale inégale, notamment les tests de susceptibilité aux maladies multifactorielles, apportant une information souvent peu utilisable au niveau individuel ;
3) à la mise à disposition de tels tests en libre accès, sur internet ;

Enjeux éthiques en génétique (2)

Liés :

4) au changement d’échelle de la production d’information ;

5) à la complexité de l’interprétation des résultats requérant

- une réévaluation au fur et à mesure de l’avancée des connaissances,
- une formation et information spécifique des professionnels de santé aussi bien que des patients ;

6) au contexte historique de la génétique et aux promesses faites ces dernières années qui ont surestimé les capacités d’action découlant de la capacité à calculer des risques.

Tests génétiques : challenges collectifs

- La place de l’information génétique au sein du système de santé
- Le flot d’information produite, qui dépasse l’information utile au temps T
  - Masse d’information à gérer, à protéger
  - Que faire des informations devenant disponibles, mais non recherchées
- Education professionnelle, accompagnement

Avantage ou fardeau économique?

- La question économique se pose :
  - Question du coût
  - Question de l’accès équitable (pb des brevets)
- D’un côté des médicaments mieux adaptés, et une plus grande sécurité
- De l’autre des patients encore plus « catégorisés » peut-être pas toujours à leur avantage

Plan

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- Génome donnée personnelle ordinaire

Des corpus de règles variés

- Eléments du corps humain
- Données de recherche
- Données personnelles et en leur sein données de santé

Le génome est difficile à cerner du point de vue légal aussi

Le partage (ou non) est au cœur de la plupart des considérations

Au sujet du partage aujourd'hui

- Introduction
  - Large scale biology, big data and data sharing
- The ethical dimensions of data sharing
- Identifiability and anonymity
- Relevant evolution of data sharing legal framework
  - Revision of the EU data protection Directive
- Context of the global alliance

What to share and when?

« One of the lessons from the Human Genome Project (HGP) was the recognition that making data broadly available **before publication** can be profoundly valuable to the scientific enterprise and lead to public benefits. This is particularly the case when there is a community of scientists that can productively use the data quickly — beyond what the data producers could do themselves in a similar time period, and sometimes for scientific purposes outside the original goals of the project.” NATURE|Vol 461|10 September 2009, 168-170

In Research,

Data Sharing = set of practices aiming at allowing and facilitating the re-use of data gathered or produced in the context of (publicly funded) research by other researchers (does not imply necessarily free access)

International collaboration

"…The full benefits for which the subjects gave their samples will be realized through maximizing collaborative high quality research. **Therefore there is an ethical imperative to promote access and exchange of information.**"

Data Storage and DNA Banking for Biomedical Research: Technical, Social and Ethical Issues, ESHG, 2001, art. 17.

What does prevent sharing of bioresources/ data ?

Technical aspects?

Protection of individuals?

Intellectual / institutional protection?

Lack of rewarding mechanisms?

Balancing values evolve

“Ethics does not consist of a static set of theories or principles that can unproblematically be ‘applied’ to new situations.”

Knoppers and Chadwick, Nat Rev Genet, 2005

“However, informed consent is far from being a magical solution to ethical preoccupations,…”

Cambon-Thomsen, Nat Rev Genet, 2004

“A focus on informed consent is also highly convenient for researchers and their institutions, and above all commercial enterprises. The reason is simple: insofar as individual rights delimit the domain of ethics, they shield other substantive issues from critical scrutiny.”

Garrath, Genomics, Society, Policy, 2005

Why is sharing data (and samples) important?

- Enable research
- Build on existing resources
- Construct communities
- Optimise fund investment in research
- Ethical imperative
- Policies in place (in light of open access)
- But…

Why are such resource sharing important…. and poorly done?

- Much biomedical/epidemiological research is based on using bioresources

- Their access to all relevant researchers is essential
- Promoting their sharing is crucial
- **There are today ~ no incentive to that.**

An OECD text of reference

- In 2004, the ministries of science and technology asked OECD to define guidelines based on agreed principles in order to facilitate access to digitalised data issued from publicly funded research.
- **OECD Principles and guidelines for access to data from publicly funded research (2007)**

http://www.oecd.org/dataoecd/9/60/38500823.pdf

Landscape of data sharing (1)

- Principles for rapid release of genome sequence data from the HGP formulated at a meeting in Bermuda 1996;
- Bermuda meetings (1996, 1997 and 1998)
- 2003 Fort Lauderdale meeting,
- – rapid prepublication release (resource for scientific community)
- – responsibilities of the resource producers, resource users, and the funding agencies.
- – 2004 : OECD
- 2008 Amsterdam meeting extended to proteomics
- 2009 Toronto statement on pre-publication data release
- 2010 : Oxford Int Conf on data sharing (+P3G)

The landscape of data sharing (2)

- 2011: Sharing research data to improve public health: joint statement of purpose by 17 major health funding agencies (+ Comment in Lancet, 10/1/11)
- 2011: A data sharing Code of Conduct for international genomic research (Knoppers et al. *Genome Medicine* 2011, **3**:46)
- 2012:
  The tension between data sharing and the protection of privacy in genomics research.
  J. Kaye. Annu Rev Genomics Hum Genet 13, 415 (2012)

The landscape of data sharing (3)

- **June 2013: Global alliance; Geneticists push for global data-sharing**
- International organization aims to promote exchange and linking of DNA sequences and clinical information:
- initially 69 institutions in 13 countries,
- now 116 in 18 countries on 6 continents.
- Nature 498, 16–17 (05 June 2013) | doi:10.1038/498017a

http://www.nature.com/news/geneticists-push-for-global-data-sharing-1.13133

The 7 main principles
(code of conduct)

1. **Quality**
2. **Accessibility**
3. **Responsibility**
4. **Security**
5. **Transparency**
6. **Accountability**
7. **Integrity**

The 7 main principles

(code of conduct)

1. **Quality**

   bona fide researchers.

   Proof of academic or other recognized peer reviewed standing is essential.

2. **Accessibility**

   Facilitation of both the deposit of data and secure access to data are the foundations of data sharing.

   Curators of databases should promote sharing to generate maximum value.

The 7 main principles

(code of conduct)

3. Responsibility

- Responsible governance should be shared between funders, generators and users of data.
- Investments in databases require coordination, strategy and long-term core funding.
- Mechanisms for building interoperability should be encouraged and appropriate management anticipated.
- Capacity building and recognition of all the data generators contributes to best practices.

The 7 main principles

(code of conduct)

4. Security

- Trust and the promotion of data sharing rely on data management, security mechanisms, oversight
- Mechanisms for identifying and tracking data generators and users should be international.

5. Transparency

- Key policies on publications, intellectual property, and industry involvement should be public.
- Websites that are accessible to the general public serve to provide feedback on general results.

The 7 main principles

(code of conduct)

6. Accountability

- Inter-agency co-operation and funding fosters efficient monitoring and good governance.
- Provisions should be made for ongoing public engagement that is tailored to local cultures.

7. Integrity

- Mutual respect and professional integrity.
- Prevention of harms and anticipation of public concerns
- Irresponsible research practices should be reported.
- Sanctions for breach of this Code must be clear.

An example of initiative on rewarding mechanisms

- BRIF : Bioresource research impact factor

The Bioresource Research Impact Factor initiative, a work in progress

Anne Cambon-Thomsen (*leader*), **Laurence Mabile** (*project manager*)
on behalf of the BRIF working group

Inserm et Univ. Toulouse III UMR 1027, Toulouse, France

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BRIF

Bioresource Research Impact Factor

Objective: increase sharing
Purpose: recognition of the effort involved in establishing, maintaining and sharing resources
Challenge: how to overcome the feeling of loss that may prevail

Idea: Transforming a loss into an advantage.
« The more it is shared, the more it contributes to science, the more it is recognised »
Virtuous circle!

http://www.gen2phen.org/groups/brif-bio-resource-impact-factor

Key elements towards BRIF

- Creating a bioresource unique identifier, or digital ID
- Standardizing bioresource acknowledgement in papers using this identifier
- Identifying parameters to take into account when creating the BRIF metrics
  - Bioresource parameters (ex: size; age…)
  - Bioresource policy parameters (analysing bioresource data access and sharing policies and classifying their impact on usability)
  - Parameters reflecting the use of the bioresource (ex: nb of projects supported /year; nb of MTA or DTA)
- Creating a BRIF metrics and test it
  Implement it (involvement of stakeholders)

‘BRIF MAIN PUBLICATIONS

RECENT PAPERS:

-CAMBON-THOMSEN A, THORISSON GA, MABILE L on behalf of the BRIF workshop group. **The role of a bioresource research impact factor as an incentive to share human bioresources.** *Nat Genet, 2011, 43: 503-4*

-BRAVO E, CAMBON-THOMSEN A, DE CASTRO P, MABILE L, NAPOLITANI F, NAPOLITANO M, ROSSI AM. **Citation of bioresources in journal articles: moving towards standards**. *European Science Editing* 2013;39(2) 36-38

-MABILE et al and the BRIF working group. **Quantifying the use of bioresources for promoting their sharing in scientific research.** http://www.gigasciencejournal.com/content/2/1/7

INITIAL PAPERS:

-CAMBON-THOMSEN A. **Assessing the impact of biobanks**. *Nat Genet*, 2003, 34, (1) : 25-26

-KAUFFMANN F., CAMBON-THOMSEN A. **Tracing biological collections: between books and clinical trials.** *JAMA*, 2008;299(19): 2316-2318

Des exemples….

- Introduction
  - Large scale biology, big data and data sharing
- The ethical dimensions of data sharing
- Identifiability and anonymity
- Relevant evolution of data sharing legal framework
  - Revision of the EU data protection Directive
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Some examples of issues

Ex1. Testing Racial Purity

ESHG condemns use of testing to establish 'racial purity'

Wednesday, June 13, 2012

The use of genetic testing to establish racial origins for political purposes is not only scientifically foolish, but also unethical and should be condemned, the European Society of Human Genetics (ESHG) said. The society said that the use by a member of parliament from the Hungarian far-right Jobbik party of a genetic test to attempt to prove his 'ethnic purity' was ethically unacceptable.

The company Nagy Gén scanned 18 positions in the MP's genome for variants that it said were characteristic of Roma and Jewish ethnic groups and concluded that Roma and Jewish ancestry could be ruled out.

GENETICS

Genome test slammed for assessing 'racial purity'

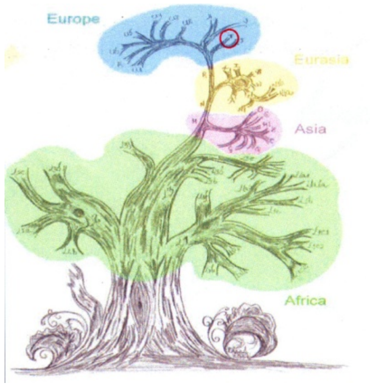
Hungarian far-right politician certified as 'free of Jewish and Roma' genes.

BY ALISON ABBOTT

14 JUNE 2012 | VOL 486 | NATURE | 167

nature

The rise of the right: Hungary's Jobbik party rallies supporters in Budapest.



Ex2. Genome Identification

Identifying Personal Genomes by Surname Inference

Melissa Gymrek,[1,2,3,4] Amy L. McGuire,[5] David Golan,[6] Eran Halperin,[7,8,9] Yaniv Erlich[1]*

Sharing sequencing data sets without identifiers has become a common practice in genomics. Here, we report that surnames can be recovered from personal genomes by profiling short tandem repeats on the Y chromosome (Y-STRs) and querying recreational genetic genealogy databases. We show that a combination of a surname with other types of metadata, such as age and state, can be used to triangulate the identity of the target. A key feature of this technique is that it entirely relies on free, publicly accessible Internet resources. We quantitatively analyze the probability of identification for U.S. males. We further demonstrate the feasibility of this technique by tracing back with high probability the identities of multiple participants in public sequencing projects.

www.sciencemag.org SCIENCE VOL 339 18 JANUARY 2013

Summary

- Sharing sequencing data sets without identifiers has become a common practice in genomics.
- Surnames can be recovered from personal genomes by profiling short tandem repeats on the Y chromosome (Y-STRs) and querying recreational genetic genealogy databases.
- A combination of a surname with other types of metadata, such as age and state, can be used to triangulate the identity of the target.
- This technique entirely relies on free, publicly accessible Internet resources.
- Quantitative analyze the probability of identification for U.S. males.
- Demonstrate the feasibility of this technique by tracing back with high probability the identities of multiple participants in public sequencing projects.

En 2013,
« buzz on identifiability »

Lots of articles following that one
on re-identification….

Science 18 January 2013: Vol. 339 no. 6117 pp. 321-324 - Report + Interview
+ 39 p Supp. Material.

Identifying Personal Genomes by Surname Inference

Melissa Gymrek[1,2,3,4], Amy L. McGuire[5], David Golan[6], Eran Halperin[7,8,9], Yaniv Erlich[1]

Science 18 January 2013: Vol. 339 no. 6117 p. 262 - News & Analysis Genetics

Genealogy Databases Enable Naming of Anonymous DNA Donors

John Bohannon

Science 18 January 2013: Vol. 339 no. 6117 pp. 275-276 - Policy Forum - Research Ethics

The Complexities of Genomic Identifiability

Laura L. Rodriguez1, Lisa D. Brooks1, Judith H. Greenberg2, Eric D. Green1

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[2] *National Institute of General Medical Sciences, NIH, Bethesda, MD , USA.*

Science 8 February 2013: Vol. 339 no. 6120 p. 647 - *Letter*

Legal Limits to Data Re-Identification

Stephen Wilson

Lockstep Consulting Pty Ltd, Five Dock (Sydney) NSW 2046, Australia.

Science 1 March 2013: Vol. 339 no. 6123 p. 1032 - *Letter*

Data Re-Identification: Prioritize Privacy

Amy Gutmann. *Chair, Presidential Commission for the Study of Bioethical Issues and President, University of Pennsylvania, Philadelphia, PA, USA*

Hastings Cent Rep. 2013 May-Jun;43(3):15-8.

Found your DNA on the web: reconciling privacy and progress.

Gutmann A, Wagner JW.

Science 1 March 2013: Vol. 339 no. 6123 p. 1033 - *Letter*

Data Re-Identification: Protect the Children

David Gurwitz *Human Molecular Genetics and Biochemistry, Sackler Faculty of Medicine, Tel Aviv University, Israel*

Science 1 March 2013: 1032-1033 - *Letter*

Data Re-Identification: Societal Safeguards

Russ B. Altman, Ellen Wright Clayton, Isaac S. Kohane, Bradley A. Malin, and Dan M. Roden

Perspective

- In authors view, the appropriate response to genetic privacy challenges is not for the public to stop donating samples or for data sharing to stop.
- Rather, establishing clear policies for data sharing, educating participants about the benefits and risks of genetic studies, and the legislation of proper usage of genetic information are pivotal ingredients to support the genomic endeavor.

Related policy forum

- Reconsidering whether a simplistic distinction between identifiability and non identifiability remains adequate as a metric for describing expectations about participant protections.
- Modalities
  - "citizen science" initiatives
  - US current patchwork of extant and potential legal standards for acquiring and managing such information adds uncertainty
- Time for the research community to engage in a rigorous and open discussion about data identifiability and how to balance effectively the benefits of broad data sharing and the imperative to respect and protect research participants.

Outline

- Introduction
  - Large scale biology, big data and data sharing
- The ethical dimensions of data sharing
- Identifiability and anonymity
- Relevant evolution of data sharing legal framework
  - Revision of the EU data protection Directive
- Context of the global alliance
- Conclusion

EU Data protection revision: potential implications for health research

Slides prepared in collaboration with with
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Introduction

- The main instrument for the legal protection of personal data in the EU: Directive 95/46/EC on the protection of individuals with regard to the processing of personal data and on the free movement of such data.

  - **Purpose**: To find the right balance between a high level of protection for the privacy of individuals and the free movement of personal data within the EU.

- The Directive is currently under revision ➔ Proposal for a Regulation on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation) , 25/01/2012, then various comments and amendments proposals. Final vote expected Spring 2014.

EUROPEAN COMMISSION

Brussels, 25.1.2012
COM(2012) 11 final

2012/0011 (COD)

Proposal for a

REGULATION OF THE EUROPEAN PARLIAMENT AND OF THE COUNCIL

on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation)

Objectives of the revision

- **To modernize** the EU legal system regarding technological developments;
- **To improve the clarity and coherence** of the EU legal framework ;
- **To achieve effective legal implementation of data protection** and of the rules allowing their lawful free movement;
- **To continue guaranteeing a high level of individuals' protection (personal rights)** in the processing of personal data in all areas of the EU's activities.

Agenda

I/ Health data protection in the proposal of regulation:

1/ From "Directive" to "Regulation"
2/ **New category of sensitive data: genetic data**
3/ Enhancement of fundamental rights
4/Reinforcement of international collaboration: transfer to Third Countries
5/Data protection impact assessment
6/New mechanisms of data protection
7/ Data protection authorities : EC and supervisory authorities

II/ Emergence of legal regime for the processing of data of a child in the proposal of regulation

Some proposed amendments to the Regulation proposal for « Data protection »

- Deleted exception “***or for historical, statistical and scientific research purposes.***”
  - *Justification: “Processing of sensitive data for historical, statistical and scientific research purposes is not as urgent or compelling as public health or social protection. Consequently, there is no need to introduce an exception which would put them on the same level as the other listed justifications.”*

Some elements to discuss in Regulation proposal: consent (1)

1/ Initial proposal (January, 2012): “Consent should be given explicitly by any appropriate method enabling a freely given specific and informed indication of the data subject's wishes, either by a statement or by a clear affirmative action by the data subject, ensuring that individuals are aware that they give their consent to the processing of personal data.”

- For all Sensitive Data
- For Data processing concerning…scientific research consent could be one condition but not the only one

Some elements to discuss in Regulation proposal: consent (2)

2/ EU Parliament amendments on the legal regime (Dec., 2012, fisrt voted by a EP Committee Oct 21 2013) "The amendments to paragraphs (…) ensure that health data, which is extremely sensitive, may only be used without the consent of the data subject if it serves a (exceptionally) high public interest and in this case must be anonymised or at least pseudonymised using the highest technical standards."

- For processing of personal data concerning health which is necessary for … scientific research purposes, shall be permitted only with the consent of the data subject (…)

Next steps

- The provisions exposed are still not final : *necessity to wait for the final version of the Regulation;*
- More *clarification* regarding the data processing for *scientific research purposes* (definition) and their *legal regime* (consent/assent and exemption);
- Necessity to integrate specific provisions *on processing of data of children in research studies;*
- *Respect of other relevant legislation for the processing of personal data* for the purposes of historical, statistical or scientific research
- Implementation of the Regulation
- Consequences and practical aspects for research

Outline

- Introduction
  - Large scale biology, big data and data sharing
- The ethical dimensions of data sharing
- Identifiability and anonymity
- Relevant evolution of data sharing legal framework
  - Revision of the EU data protection Directive
- Context of the global alliance
- Conclusion

Global alliance in context

- **Geneticists push for global data-sharing**
- International organization aims to promote exchange and linking of DNA sequences and clinical information.
- Nature 498, 16–17 (05 June 2013) | doi: 10.1038/498017a
- Now 116 institutions in 18 countries
- http://www.nature.com/news/geneticists-push-for-global-data-sharing-1.13133

EU Commission funded projects establish their data sharing policy :

a laboratory of practices

CageKid Cancer Genomics of the Kidney	www.cng.fr/cagekid	Cancer Genomics of the Kidney –	R&D
GEN2PHEN	*www.**gen2phen**.org*	Genotype-To-Phenotype Databases: A Holistic Solution	R&D
ESGI	www.esgi-infrastructure.eu	European Sequencing and Genotyping Infrastructure	IS
GEUVADIS CONSORTIUM	www.geuvadis.org	Sharing capacity across Europe in high-throughput sequencing technology to explore genetic variation in health and disease	CA

International Cancer Genome Consortium

ICGC - International cancer genome consortium
http://icgc.org/

En conclusion… International collaboration

"…The full benefits for which the subjects gave their samples will be realized through maximizing collaborative high quality research. Therefore there is an ethical imperative to promote access and exchange of information."

Data Storage and DNA Banking for Biomedical Research: Technical, Social and Ethical Issues, ESHG, 2001, art. 17.

What does prevent sharing of (bio) resources ?

- **Protection of individuals : approach by governance and legislation and implication of stakeholders (participants)**
- **Lack of rewarding mechanisms : set up tools**

The ethics of sharing is at the heart of scientific research

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Merci pour votre attention