

Deborah Mascalzoni, PhD
Center for research ethics and bioethics CRB,
Uppsala University

and

Center of Biomedicine
EURAC research
Bolzano/Bozen

1. Personal information

Date of Birth:

Nationality:

Education

April 2005 PhD in Bioethics at the university of Bologna (faculty of law)

2003.2004 One year fellowship in the program of Science and Society at Harvard University (Kennedy School of Government with professor Sheila Jasanoff)

January 2002. Awarded position as doctoral student in Bioethics - CIRSFID - University of Bologna – Italy

September 2001 Master in Environmental Education (research Paper on Environmental Ethics)

March 2000 Degree in Philosophy of Science cum Laude (Thesis in Bioethics: "Philosophy of Medicine: who should decide about Therapy and Care"), University of Bologna- Italy

Professional Updates:

Fall 2014 Academic Teaching training course (Uppsala University)

October 2014 Supervising at Doctoral Level (Uppsala University)

2. Research Interests

Deborah Mascalzoni's research interests include Bioethics, Law, Environmental Ethics, Science Policy, Philosophy of Politics. Special interests:

- Patients rights in research
- **Genetics and new technologies, Informed consent, privacy and Data Sharing, patient communication**
- PhD thesis on: “**Consenso informato e processi di partecipazione**”.
- Role of stakeholders in medicine (**Lay-experts, citizen juries, focus groups, interviews, democratic participation in health**)
- She leads the ethical legal aspects of research projects the Center of biomedicine at the EURAC and especially for the biobank based project CHRIS

3. Professional Experiences

- **2013-2015** appointed as senior researcher at Uppsala University, Center for Ethics and Bioethics. Management of the WP6 EU project RD-Connect on Rare disease in on Ethical and Legal issues.
- **2013-2015** Teaching ethics and bioethics at different courses at Uppsala University (85 hours)
- **2004- 2015 Joint position as Senior Researcher in Bioethics** at the European Academy: Institute of Genetic Medicine at the project CHRIS
 - She leads the ethical legal aspects of research projects at the Center of biomedicine at the EURAC and is especially committed with interactive dynamic consent projects in the biobank based project CHRIS
- **2004-2012** “Ethics in medicine” and “nursing ethics” courses at the Claudiana (University of Roma section of Bolzano, Claudiana) and for the local Health System
- **2006-2007** Appointment as “professore a contratto” for Bioethics at the Master in geriatrics Nursing, (appointed by the University of Roma section of Bolzano, Claudiana)
- **May 15-18 2006, July 17-20 2006, September 2007** Advisory work in Ethical Panels on FP6 Projects, European Commission, Bruxelles, Belgium,
- **2005_2006** Scientific coordination: research Project: Network “Bioetica Biobanche Italiane”
- **2005** Teaching Bioethics courses IPASVI , ECM
- **October 2001-2005** Assistant for courses in Legal Philosophy and Sociology of Law, CIRSIFID, University of Bologna - Italy

- **2004 - 2005** Research Project on Genetics and Ethics ; **Dissertation research paper** on “Informed consent in genomics: a participative process ”
- **2003-2005** responsabile della segreteria didattica dottorato di Bioetica Bologna
- **November 2002** Seminar on Environmental Ethics, Master in Environmental Education at the university of Bologna.

Institutional roles: Ethical and Review BOARDS

- ADOPT: Advisory Board member
- Committee for HUMAN research and Bioethics (HUB), IFOM (Fondazione Istituto Firc di Oncologia Molecolare, Milano),
- Member of the ELSI Group of BBMRI Italy
- Ethical Advisor for Telethon Italy
- Ethical Advisor for UNIAMO
- Member of the local Health system ethical board of Bolzano
- National referent for legal and ethical issues WIKI Platform of BBMRI (Biobanking and Biomolecular Resources Research Infrastructure)
- Member of BBMRI Italy ELSI working group
- Member of the ethical group of the International Genetic Epidemiology Society (IGES)
- P3G Member (public population Project in genomics)
- Bruxelles FP6- FP7 ethical expert for evaluation of European projects since 2006-2012
- Member of the provincial Ethical board of South Tyrol until 2013

Peer Reviewed Publications

Publications

1. Isabelle Budin-Ljøsne, Harriet Teare, Heidi Beate Bentzen, Luciana Caenazzo, Clive Collett, Flavio D'Abramo, Teresa Finley, Erica Jones, Višnja Katić, Amy Simpson, Jane Kaye, Deborah Mascalzoni, "Dynamic Consent: a possible solution to some of the challenges of modern biomedical research" BMC Medical Ethics (under review)
2. Isabelle Budin-Ljøsne, Deborah Mascalzoni, Sirpa Soini, Helena Machado, Jane Kaye, Heidi Beate Bentzen, Emmanuelle Rial-Sebbag, Flavio D'Abramo, Michał Witt, Geneviève Schamps, Višnja Katić, Dusanca Krajnovic, Jennifer R. Harris, "Feedback of individual genetic results to research participants: Is it feasible in Europe?", Biopreservation and Biobanking
3. Pauline McCormack, Anna Kole, Sabina Gainotti, **Deborah Mascalzoni**, Caron Molster, Hanns Lochmuller, Simon Woods "You should at least ask" The expectations, hopes and fears of rare disease patients on large scale data and biomaterial sharing for genomics research. EJHG 2016
4. James M. Roberts, Deborah Mascalzoni, Roberta B. Ness, Lucilla Poston, Collaboration to Understand Complex Diseases Preeclampsia and Adverse Pregnancy Outcomes, Hypertension, April 2016
5. Sabina Gainotti, Cathy Turner, Simon Woods, Anna Kole, Pauline McCormack, Hanns Lochmüller, Olaf Riess, Volker Straub, Manuel Posada, Domenica Taruscio, **Deborah Mascalzoni**, Improving the informed consent process in international collaborative rare disease research: effective consent for effective research., EJHG 2016
6. Viviana Meraviglia, Jianyan Wen, Luca Piacentini, Giulia Campostrini, Cheng Wang, Maria Cristina Florio, Valerio Azzimato, Lorenzo Fassina, Martin Langes, Johnson Wong, Michele Miragoli, Carlo Gaetano, Giulio Pompilio, Andrea Barbuti, Dario DiFrancesco, **Deborah Mascalzoni**, Peter P. Pramstaller, Gualtiero I. Colombo, Huei-Sheng Vincent Chen, Alessandra Rossini, "Higher cardiogenic potential of iPSCs derived from cardiac versus skin stromal cells", Frontiers In Bioscience, Landmark, 21, 719-743, January 1, 2016
7. Isabelle Budin-Ljøsne, Harriet Teare, Jane Kaye, **Deborah Mascalzoni** "Meta consent: Is it new and is it fit for purpose?" BMJ, 04. Nov 2015, [Http://www.bmj.com/content/350/bmj.h2146/rr-0](http://www.bmj.com/content/350/bmj.h2146/rr-0)
8. Pattaro C, Gögele M, **Mascalzoni D**, Melotti R, Schwienbacher C, De Grandi A, Foco L, D'Elia Y, Linder B, Fuchsberger C, Minelli C, Egger C, KofinkLs, Zanigni S, Schäfer T, Facheris MF, Smáráson SV, Rossini A, Hicks AA, Weiss

- H & Pramstaller PP, The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results, *Journal of Translational Medicine*, 2015;13:348
9. **Mascalzoni D**, Dove ES, Rubinstein Y, Dawkins HJS, Kole A, McCormack P, Woods S, Reiss O, Schaefer F, Lochmüller H, Knoppers BM & Hansson MG, International Charter of principles for sharing bio-specimens and data, *European Journal of Human Genetics* 2015;23:721-728
 10. Graham CE, Molster C, Baynam GS, Bushby K, Hansson M, Kole A, Mascalzoni D, Mora M, Monaco L, Bellgard M, Carpentieri D, Posada M, Riess O, Rubinstein YR, Schaefer F, Taruscio D, Terry SF, Zatloukal K, Knoppers B, Lochmüller H, Dawkins HJS, Current trends in biobanking for rare diseases: a review, *Journal of Biorepository Science for Applied Medicine* 2014;2:49-61
 11. Simpson CL, Goldenberg AJ, Culverhouse R, Daley D, Igo RP, Jarvik GP, Mandal DM, Mascalzoni D, Gray Montgomery C, Pierce B, Plaetke R, Shete S, Goddard KAB, Stein CM, Practical Barriers and Ethical Challenges in Genetic Data Sharing, *International Journal of Environmental Research and Public Health*, 2014;11(8):8383-8398
 12. Mascalzoni D, Paradiso A, Hansson M, Rare Disease Research: Breaking the Privacy Barrier, *Applied & Translational Genomics*, Available online 18 April 2014
 13. Destro Bisol G, Anagnostou P, Capocasa M, Bencivelli S, Cerroni A, Contreras J, Enke N, Fantini B, Greco P, Heeney C, Luzi C, Manghi P, Mascalzoni D, Molloy J, Parenti F, Wicherts JM & Boulton G, Perspectives on open science and scientific data sharing: an interdisciplinary workshop, *Journal of Anthropological Science*, 2014;92:1-22
 14. Mascalzoni D, Knoppers BM, Aymé S, Macilotti M, Dawkins H, Woods S, Hansson MG, Rare Diseases and Now Rare Data?, *Nature Review Genetics*, 2013, doi:10.1038/nrg3494, published online April 23
 15. Napolitano M1, Santoro F, Puopolo M, Donfancesco C, Galluzzo L, De Grandi A, Cevenini E, De Curtis A, Sevini F, Palmieri L, Mascalzon D, Roazzi P, Scafato E, Pramstaller P, Iacoviello L, Donati MB, Giampaoli S, Franceschi C, Belardelli F, Bravo E., Development of a pilot project on data sharing among partners of the Italian Hub of Population Biobanks (HIBP): association between lipid profile and socio-demographic variables., *Biopreserv Biobank*. 2014 Aug;12(4):225-33. doi: 0.1089/bio.2014.0001. Epub 2014 Jul 30.
 16. D. Mascalzoni, P. Pramstaller & C. Corradetti (2013). Patient Centric Initiatives (PCIs) - a Shift in the Governance of Science: Lessons From the Biobanks World. 2013, *Research Ethics* 9 (2):52-54.

17. Mascalzoni, Corradetti, Editorial to the special issue Ed. by Mascalzoni and Corradetti, special issue on Genomics and public participation, " *Studies in Ethics, Law, and Technology*" 2012
18. *Nature Reviews Genetics* 2012, From patients to partners: participant-centric initiatives in biomedical research, Jane Kaye¹, Liam Curren², Nick Anderson³, Kelly Edwards⁴, Stephanie M. Fullerton⁴, Nadja Kanellopoulou¹, David Lund⁵, Daniel G. MacArthur⁶, **Deborah Mascalzoni**⁷, James Shepherd⁸, Patrick L. Taylor⁹, Sharon F. Terry¹⁰ & Stefan F. Winter¹¹
19. Mascalzoni et al. "Comparison of participant information and informed consent forms of five European studies in genetic isolated populations" *EJHG* 2009
20. Mascalzoni, Deborah; Hicks, Andrew; and Pramstaller, Peter P. (2009) "Consenting in Population Genomics as an Open Communication Process," *Studies in Ethics, Law, and Technology*: Vol. 3 : Iss. 1, Article 2.
21. Mascalzoni D, Hicks A, Pramstaller P, Wjst M (2008) Informed Consent in the Genomics Era. *PLoS Med* 5(9): e192.
22. Pattaro C, Marroni F, Riegler A, Mascalzoni D, Pichler I, Volpato CB, Dal Cero U, De Grandi A, Egger C, Eisendle A, Fuchsberger C, Gogele M, Pedrotti S, Pinggera GK, Stefanov SA, Vogl FD, Wiedermann CJ, Meitinger T, Pramstaller PP. The genetic study of three population micro-isolates in South Tyrol (MICROS): study design and epidemiological perspectives. *BMC Med Genet.* 2007 Jun 5;8(1):29
23. Mascalzoni D., *Aspetti etici della genomica delle popolazioni: il caso deCODE Genetics "Kykeion"*, 11, 2004

Peer reviewed Books (peer reviewed):

Edited by Mascalzoni, *Ethics, Law and Governance of Biobanking: National, European and International Profiles*. D. Mascalzoni (ed.); Springer, Dordrecht, The Netherlands (2015)

Peer reviewed Bookchapters

1. **Mascalzoni D**, Introduction, in Edited by Mascalzoni, *Ethics, Law and Governance of Biobanking: National, European and International Profiles*. D. Mascalzoni (ed.); Springer, Dordrecht, The Netherlands 2015

2. D Mascalzoni, Tra diritti individuali deboli e diritti comunitari assenti, in Mordacci ed., Bruno Mondadori, 2012
3. D. Mascalzoni, in Trattato di Biodiritto, Biobanche e genetica delle popolazioni, Giuffre'editore, 2010
4. Etica pubblica ed ecologia, in "Etica pubblica ed ecologia", ed. G.L. BRENA, Messaggero, Padova, 2005
5. "Etica ambientale e Responsabilita`" in Miur 2000, CIRSFID (University of Bologna), 2003

Peer reviewed and defended Thesis:

1. Mascalzoni, Informed Consent in Genomics: a new contract between science and Society, PHD thesis, 2005
2. Mascalzoni D, Il principio di responsabilita´in etica ambientale (Environmental Ethics and the Imperative of Responsibility), Master Thesis, 2001
3. Mascalzoni, "Philosophy of Medicine: who should decide about Therapy and Care", 2000, University of Bologna

Official EU deliverables

1. Children involvement in longitudinal research
2. Return of Results in rare diseases
3. Data Sharing in a Rare diseases consortia
4. Ethical Framework for a Rare diseases consortia
5. Carrier Status: ELSI issues in the communication of carrier status in research for rare disease families

Published abstracts and Posters:

1. ESBB
2. UNESCO Conference
3. Mexico
4. Mascalzoni Bio-banks evolution. Reevaluation of the role of the patient as a partner, International journal of molecular biomarkers, 2010

Grants:

- 2015 EU COST Action Chip me: STSM grant for researcher mobility in Oxford
- 2013 EU COST Action CHIP me: co-coordination of the WP1 Group ELSI
- 2009 FIRB Italian Ministry grant project on ethical and legal issues for biobanks, funded by the MIUR (Italian ministry of education), EURAC
- 2004 Travel Grant International Conference in Iceland “Ethical legal and Social Aspects of Human Genetic Databases”
- 2003 European Grant Marco polo (University of Bologna-Harvard University): international mobility grant
- 2001 2 years Research Grant as “assegno di ricerca” for the national Project "Principle-based Universalism and Cultural relativism", Bologna university, CIRSFID, Faculty of Law
- 2000: Grant by the SouthTyrol Region for the Bachelor Thesis defended in Bologna on the 28th March 2000.
- 1997 - 1998 ERASMUS: one year grant at the University of Freiburg ((Project on Intercultural Pedagogy).

Biobank’s policy and applied ELSI work:

- Information leaflets for Population research
- IT based Beta model for Dynamic Consent in research (2015)
- ELSI regulation for the biobank CHRIS (2011)
- Informative Brochure for CHRIS Study (version 1 and 2) (2011/2014)
- Survey on the perception of Dynamic consent in the South tyrolean population participating in the CHRIS Study (600 participants)
- Qualitative interviews with Healthy participants of the project CHRIS about the perception of Dynamic Consent (2014)
- Development of an Information Movie to support informed consent (2010)
- Development of a Policy regulation on Access and Sharing of Bio-samples and Data (EURAC Biobank) (2014)
- Collaboration structure between research biobanks and e-health care system in Southtyrol
- Development of an MTA template for Data and Sample Sharing (2013)
- Policy on return of incidental Findings (Chris project) (2013)
- Dynamic Consent tool for CHRIS project (2009)
- Privacy structure of the CHRIS project (2011)
- Communication Strategy (ongoing information) for the CHRIS Project (2008-2009)

- Perception among GP's of Genomics (South Tyrol) Report available in German, 2011)
- Informational Meetings with the local population of the Southtyrolean valleys for community consent (2005-2010)
- 2007-2008 Survey with the local population on perception of the genomic Project MICROS
- Focus Group on perception of Genomics among the population in South Tyrol (Ladin valley)
- Interview with a midwife of a Ladin valley about the perception of genomics in the population
- 2005-2006: informed consent for the population biobank MICROS

Policy Documents based on scientific advice:

PHG Foundation guidelines

Scientific Outreach

- Blog: Consent and Engagement in Research, editor in CHIEF
- Science Cafe' Salute etica. Il dilemma della libertà di decisione del paziente
- Science Cafe' 16 June 2005 : "Non" decido per te! Incontro sul futuro della ricerca biomedica e della fecondazione assistita in Italia
- January 2003 – July 2003 : Scientific Board for the «Citizens School»(I Lumi Della Ragione, Scuola di Etica Laica) on Bioethical Issues, held in Bologna.

International Events organized:

Workshop in Oxford on Dynamic Consent

Workshop Uppsala on Children

12. 17-18 October 2013, Stakeholder Conference, Brussels (scientific committee chair)

26-27 10/2011 Rome, International consensus Workshop on patient centric initiatives (Scientific Chair)

27/29 10/2011 Rome international Conference on Patient Centric approaches (Chair of the scientific organizing committee)

International Conferences (invited)

19-21 Nov 2013, Geneva, Brocher Foundation, Returning Genetic Results in Biobanks:

Opening an International Dialogue, Presentation on Incidental Findings policies and ethics

12. 17-18 October 2013, Stakeholder Conference, Brussels (scientific organization and presentation)

11. 15-17 2013 Prague BT-Cure annual meeting , Report on models for consent in Rare Diseases , Presentation on Dynamic Consent in Rare Diseases Research

9. 15 June 2013 Stockholm , Invited Presentation on Informed consent in Clinical trial, Annual Conference EHA (European Hematology Association) Invited

8. 12-14 June 2013, Geneva, Brocher Foundation, Exploring innovative mechanisms to build trust in human health research biobanking; Organized by Jennifer Harris (Norwegian Institute of Public Health, Norway); invited poster on Informed consent: a challenge won by trust

4.25-27 April 2013, London Workshop on E-Health and invited Lecture on Consent in Biobanking

6.28-31 May 2013, BBMRI-Italy meeting for Ethical Issues in Biobanking, Rome (invited speech)

3.29- 30 2013 January 2013, EFGCP Annual Conference; Virtual Future: An exploration of the ethical dimensions of emerging technologies in clinical trials and research 2013 - Palais des Académies, Brussels, Belgium; lecture on Dynamic consent in Clinical trials (Keynote speaker)

25-29. 06. 12 World conference on Bioethics: “Bioethics and the Future: the future of bioethics”

24-25. 01.12 Brussels , EFGCP annual conference: Consent: how less could be more (invited, workshop chair)

20. 01. 12 Rome CCM Project Conference, “Elsi issues for Population biobanks” (invited speaker)

25-06. 12 Rotterdam: Workshop ELSI 2.0 , satellite meeting of the world conference on Bioethics: “Bioethics and the Future: the future of bioethics”

25-29. 06. 12 World conference on Bioethics: “Bioethics and the Future: the future of bioethics”

26-27 10/2011 Rome, International consensus Workshop on patient centric initiatives (scientific organizer and Chair)

27/29 10/2011 Rome international Conference on Patient Centric approaches (Scientific chair organizer, presenter)

2011 Cambridge, COGS workshop: ELSI and organizational uncertainties in the implementation of screening. (invited expert)

2010 Oxford Workshop on Patient Centric Initiatives, The Helix Center (invited)

2010 Oxford , International Data Sharing Conference, St Hugs college: “the virtual patient: towards a shared Governance” (presentation)

7-10/7 2010 Cambridge UK , Consensus conference the COGS Project (invited)

2009 Biochip development for Cancer diagnosis, International conference on translational medicine, Venezia (invited speaker)

2009 Luxembourg, P3G (Public population project on genomics), conference.
(presentation on dynamic consent)
2009 Brussels Joint conference on biobanks PHOEBE, P3G, BBMRI, Informed consent as
a process: presentation
And Poster: Interactive communication with participants
2009 Trieste: 4rd International meeting on genetics of complex diseases and isolated
populations, Ethical issues for population projects (invited keynote speaker)
2008 April ELSI (Ethical, Legal, and Social Issues)-Meeting for the EU-funded project
EUROSPAN (EUROpean Special Population resArch Network) (organizer and chair)
2007 3rd International meeting on genetics of complex diseases and isolated populations,
Torino, Italy, May 26-29, 2007 (invited presentation)
2007 Chair of the ethical panel of “ la giornata dell’infermiere” IPASVI (invited speaker)
2007 International conference, Translational medicine and public health policy; Geneva
(invited speaker)
2006 Trieste Scienza e democrazia al bivio: riforma o restaurazione? La partecipazione
pubblica nella scienza (invited speaker)
2006 National conference: “Le Biobanche per la Ricerca e la terapia”, Genova (ECM)
(invited speaker)
2006 "Genomics and Global Health, Presentation of a United Nation Report", Kennedy
School of Government, Boston, U.S.A, (invited speaker)
2006 Trieste: National conference: Participation in policy making (Scienza e democrazia
al bivio: riforma o restaurazione? Seminario di studio (invited speaker)
2005 Giornata dell’infermiere Bolzano, IPASVI, Concetti di salute per una nuova forma di
professionalità (invited speaker)
2005 November, Geneva International Conference “Value and Risk of Genetic Data
Collections”
2002 Participation as Coordinator and moderator of the school for citizens : I lumi della
ragione: a course on public ethics, Bologna (organizer)
2001 Congress "Bioethical Matters and the Courts: Do Judges Make Law?" Pavia

Languages:

Italian mother-tongue

English: very good (IELTS)

German: very good (Oberstufe Pruefung, Zweischprachigkeitspruefung A)