europass Curriculum vitae

PERSONAL INFORMATION

Alessandra Renieri

WORK EXPERIENCE

1993–1998 **Assistente Medico - Medical Genetics**
Azienda Ospedaliera Universitaria Senese (Italy)

1998–2000 **Researcher - Medical Genetics**
University of Siena (Italy)

2000–2007 **Associate Professor - Medical Genetics**
University of Siena (Italy)

2002–Present **Director of the Medical Genetics division**
Azienda Ospedaliera Universitaria Senese (Italy)

2007–Present **Full Professor - Medical Genetics**
University of Siena (Italy)

EDUCATION AND TRAINING

–June 1983 **Maturità classica degree**
High School: Liceo Classico Virgilio of Empoli, Italy (Italy)
final marks: 60/60

–June 1989 **M.D. degree**
University of Siena (Italy)
School of Medicine
Final marks: 110/110 summa cum laude.
Thesis entitled "Aspetti genetici e molecolari dell'alfa-1- antitripsina"
Medical Genetics, Tutor: Prof. Mario De Marchi,
1990 European School of Medical Genetics Certification
Third Course, Sestri Levante

–June 1994 **PhD degree**
University of Turin (Italy)
Human Genetics
Thesis entitled "Genetica Molecolare della sindrome di Alport"
Tutor: Prof. Mario De Marchi

–November 1998 **Specialty degree**
University of Florence (Italy)
Medical Genetics
Final marks: 70/70 summa cum laude.
Thesis entitled "AMME: una nuova sindrome da geni contigui in Xq22.3"
Tutor Prof. Enrico Montali

ADDITIONAL INFORMATION

Expertise

Alessandra Renieri graduated in Medicine at the University of Siena and obtained a PhD in Human Genetics at the University of Torino. Subsequently she received a specialist degree in Medical Genetics at the University of Florence and she then went back to Siena where she worked first as Medical Assistant and then as researcher. In 2000 she was appointed Associate Professor and from 2007 she is Full Professor of Medical Genetics at the School of Medicine of the University of Siena.

From 1992 to 2002 she personally performed 1240 second-level genetic counselling activities at the division of Medical Genetics of Siena, which imply identifying a case on the basis of clinical genetics, recommending a possible molecular diagnosis, coordinating the implementation of the research, assessing the recurrence risk for relatives and, sometimes, making pre-symptomatic diagnosis. She is the director of the Medical Genetics Unit of the General Hospital of Siena. Since 2001 she has coordinated, as director of the Medical Genetics Unit, more than 10,000 genetic counselling.

Her main research interest has always been the study of the genetic basis of of rare diseases, with a special focus on Rett syndrome, and other conditions with intellectual disabilities (ID), Alport syndrome, retinoblastoma and other rare cancers. She identified two new genes disease: FACL4 gene for X-linked ID and FOXG1 gene for Rett syndrome. Her laboratory was among the first in Italy to introduce the technology of array-CGH and of Next Generation Sequencing (NGS) for clinical diagnosis.

She has been involved in research on Rett syndrome for many years and she contributed to the identification of all 3 known genes presently associated to the disease, as well as to the definition of the associated clinical phenotype. Her group identified FOXG1 as the first autosomal gene involved in Rett syndrome. Her laboratory is a referral center for Rett in Italy and, since 1998, she directs the Genetic Biobank of Siena (GBS, http://www.biobank.unisi.it), one of the few in Italy certified SIGU-CERT and ISO9001, and funded by Telethon since 2002. GBS is the Italian Partner of BBMRI (Biobanking and Biomolecular Resources Research Infrastructure), member of EuroBioBank and RD-Connect. Since 2009, she coordinates the international Rett database network (http://www.rettdatabasenetwork.org). She also coordinates the Italian Registry of Alport disease, an Italian network for Alport disease, which aims to fund and support actions in favour of the management and treatment of ATS patients. In order to create a human cellular model for the study of the pathogenic mechanisms of Rett syndrome directly in human affected neurons, she set up the technique of genetic reprogramming in her laboratory (iPS).

She has been involved in cancer genetics for many years, including retinoblastoma, breast, colon and lung cancer. Her laboratory is among the first in Italy to introduce the use of NGS for “liquid biopsy” as an innovative diagnostic and prognostic technique in cancer for early detection and monitoring cancer growth and resistance to treatment for “personalized medicine”.

Prof. Renieri is HCP (Health Care Provider) representative/sub-representative for Azienda Ospedaliera Universitaria Senese (AOUS) of 5 European Reference Networks (ERNs): EuroBloodNet (on rare haematological diseases); ERKNET (on rare kidney diseases); ERN ITHACA (on ID and congenital anomalies); PaedCan-ERN (on paediatric cancers) and EURACAN (for rare adult solid cancers). She is coordinator of Registry WP with the ERN ITHACA and involved in the interoperability between Registries at European Level (coordinator of Rett Networked Database) and is leading for ERN ITHACA the project for H2020 HP-PJ-06-2016 "Support for New Registries" call.

She is an active member of the Telethon Network of Genetic Biobanks. She acts a medical advisor with several patient organizations including AIRETT, and supervises specialist clinics for rare disorders within AOUS.

Since 2017 her reasearch interest is focused on gene editing using CRISPR systems and its translation to clinical practice. Currently, she is running four gene editing projects. Three are using Crispr/Cas9 and AAV system and are related to Rett syndrome (FOXG1 variant), Parkinson (LRRK2 and GBA) and Alport syndrome (COL4A5). One is using Crispr/Cpf1 and lentiviral vector and it is related to Chronic Lymphocytic Leukemia and other TP53 mutared cancers. More recently, in Siena she was sorting out a sort of “factory” for producing plasmid & vectors for gene editing in vitro & animal models, preliminary for clinical trial for a number of diseases including Rett syndrome, Parkinson disease, Alport syndrome and Pompe diseases, among others.

I was among the first in Italy to introduce the use of Next Generation Sequencing (NGS) for “liquid biopsy” in cancer patients and to propose a novel approach of clinical trial in advanced cancer based on cell free DNA (cfDNA) sequencing results. Detection and analysis of cfDNA now offer the possibility to detect key mutations of cancer driver genes which may play a major role in the therapy escaping mechanism. Therefore, this clinical innovative approach is based on multi-biomarker genetic NGS screening and is capable of mirroring tumor heterogeneity enabling to identify the best personalized treatment targeting the expanding clones in different solid tumors.

To rapidly respond to the ongoing COVID-19 pandemic, she is focusing on developing the most informative diagnostic test and the most powerful therapy on the basis of host genome. She is leading the GEN-COVID Multicenter Study aimed at enrolling 2,000 COVID-19 patients for host genetic analysis and she is member e co-founder of the international Host Genetic Initiative (HGI). For these purposes, recently, a section dedicated to COVID-19 was included in the established and certified Biobank and Registry of the Medical Genetics Unit of the Hospital. The Genetic and COVID-19 Biobank of Siena, is member of BBMRI-IT, of Telethon Network of Genetic Biobanks (project no. GTB18001), of EuroBioBank, and of D-Connect, provided us with specimens.

Publications

Research activities of Prof. Alessandra Renieri are substantiated by 262 original publications with a total IF >1.000 and 3 patents. She is author of 6 book chapters, 9 reviews made by request, and one N&V in Nat Genet.

Citation parameters (Scopus– updated April 2020):

N publications in the last 10ys: 134

N total citations: 8904 Normalized citations: 318

H-index: 48

average citations per item: 30.89

Prof. Alessandra Renieri ranks above the median for full professors according to ANVUR scientific quality parameters (contemporary H –index, number of publications in the last ten years, normalized citations). Being also over the median for total H-index and total citations numbers, she has been recently selected in the committee for National Scientific Qualification (2012-2015).

Projects

Active Research Grants

Source: Alport Syndrome Foundation

Title: “AAV2-CRISPR/Cas9 preclinical trial on urine derived podocyte-lineage cells and on a naturally occurring ATS dog model” - Date and cost of entire project: 2018-2020 100.000 $

Source: Blackswan Foundation

Title: “FoxG1 gene therapy using CRISPR/Cas9 technology and AAV9 iPSCs-derived neurons” - Date and cost of entire project: 2017-2019 150.000 Euro

Source: AstraZeneca

Title: “Liquid biopsy in ovarian carcinoma”

Date and cost of entire project: 2017-2018 10.000 Euro

Source: Associazione ILA

Title: “Studio Genomico mutazionale e funzionale nelle Malformazioni Vascolari per delineare appropriate strategie terapeutiche (GeVaMa_2015)”

Date and cost of entire project: 2016-2018 Studio Genomico mutazionale e funzionale nelle Malformazioni Vascolari per delineare appropriate strategie terapeutiche (GeVaMa_2015)Euro

Source: Istituto Toscano Tumori

Title: “Identification of genetic bases of individual predisposition to lung cancer in non-smokers”

Date and cost of entire project: 2015-2017 118.000 Euro

Source: Telethon

Title: “Telethon Network of Genetic Biobanks.”

Date and cost of entire project: 2013-2017 140.000 Euro

Source: Regione Toscana

Title: “SMART: Messa a punto di una strategia innovative di medicina personalizzata per la diagnosi e la terapia delle malattie renali nei bambini”

Date and cost of entire project: 2016-2018 196.000 Euro

Expired Research Grants funded

Source: Ministero della Salute

Title: “Identification of common biological pathways in Rett patients mutated in MECP2, CDKL5 and FOXG1”

Date and cost of entire project: 2012-2015 248.440,00 Euro

Source: Alport Syndrome Foundation

Title: “Podocyte response to injury in Alport Syndrome: an answer from human amniotic fluid kidney progenitors”

Date and cost of entire project: 2014-2015 22.000,00 USD

Source: Telethon

Title: “Molecular bases and in vitro modeling of Cdkl5 dependent infantile neurological disorders”

Date and cost of entire project: 2011-2014 Euro 81.900,00

Source: Ministero della Salute

Title: “Induced pluripotent stem cells as in vitro models for the study of Rett syndrome pathogenesis and identification of therapeutic targets”

Date and cost of entire project: 2011-2014 Euro 171.000,00

Source: Novartis

Title: “Variabilità genetica della risposta al vaccino antiinfluenzale (VARGEN 2)”

Date and cost of entire project: 2013-2014 60.800 Euro

Source: Telethon:

Title: "Molecular Genetics of Alport syndrome"

Date and cost of entire project: 1994-1996 Lire 85.000.000

Source: Telethon:

Title: "Cloning and characterization of the genes responsible for a new X-linked contiguous gene syndrome including Alport phenotype, mental retardation and elliptocytosis"

Date and cost of entire project: 1996-1998 Lire 148.000.000

Source: Associazione Emma ed Ernesto Rulfo per la Genetica Medica:

Title: "Progressi verso la identificazione della base genetica della sindrome di Rett"

Date and cost of entire project: 1999-2001 Lire 30.000.000; anno 2003 Euro 13.000.

Source: PAR Università di Siena:

Title: “Uso delle sequenze della Celera Genomics nell’era post-genomica e esempio di applicazione alle malattie neuropsichiatriche: Autismo e Complesso della Sindrome di Rett”

Date and cost of entire project: 2001 Lire 35.000.000

Source: MURST cofin

Title: “Corrispondenza tra genotipo e fenotipo nell’eredità del glaucoma primario ad angolo aperto”.

Date and cost of entire project: 1999-2001 Lire 154.000.000

Source: Telethon

Title: "Molecular basis of AMME contiguous gene syndrome (A=Alport phenotype, M=mental retardation, M=midface hypoplasia and E=elliptocytosis) and single gene / phenotype correlation"

Date and cost of entire project: 2000-2003 Lire 306.000.000

Source: SOI:

Premio ricerca scientifica SOI 2002

Date and cost of entire project: 2002 Euro 20.000

Source: PAR University of Siena

Title: “Ruolo dei geni FACL4 e LAMA2 nella patogenesi della Sindrome di Rett e di disordini correlati”

Date and cost of entire project: 2002 Euro 24.000

Source: Telethon

Title: "Autism and Rett syndrome complex: molecular basis of complex disorders"

Date and cost of entire project: 2002-2004 Euro 76.253

Source: Italian Ministry of Health

Title: “Analisi di mutazione e di espressione con tecnologia microarray in malattie monogeniche rare

con ritardo mentale”

Date and cost of entire project: 2003-2005 Euro 30.900

Source: Telethon:

Title: “Cell Line and DNA bank of Rett syndrome and other X-linked mental retardation syndromes”

Date and cost of entire project: 2003-2004 Euro 52.000

Source: PAR University of Siena

Title: “Caratterizzazione del profilo di espressione genica in tessuti cerebrali di un feto Rett”

Date and cost of entire project: 2004 Euro 16.000

Source: Italian Ministry of Health

Title: “Diagnosi delle leucoencefalopatie genetiche”

Date and cost of entire project: 2004-2005 Euro 20.000

Source: Fondazione Mariani

Title: “X-linked mental retardation Italian Project”

Date and cost of entire project: 2004-2005 Euro 95.000

Source: MIUR (FIRB)

Title “Determinanti molecolari della sindrome di Rett e malattie causate dal gene MECP2”

Date and cost of entire project: 2004-2005 Euro 130.000

Source: Telethon:

Title: “FACL and mental retardation: cellular and mouse model”

Date and cost of entire project: 2004-2007 Euro 180 000

Source: MUR (PRIN2005)

Title: “Caratterizzazione funzionale delle proteine ACSL4 e MECP2” as part of the project “Strategie post-genomiche per lo studio e la prevenzione del ritardo mentale X-legato” PI G. Neri

Date and cost of the project assigned to AR: 2006-2007 Euro 51.510

Source: Telethon:

Title: “Cell Line and DNA bank of Rett syndrome and other X-linked mental retardation syndromes”

Date and cost of entire project: 2007 Euro 35.000

Source: ITT (Istituto Toscano Tumori):

Title: “Molecular determinants of variable expressivity in Retinoblastoma”

Date and cost of entire project: 2007 Euro 70.000

Source: Scuola Superiore S. Chiara:

Title: “Genetic bio-bank/databases”

Date and cost of entire project: 2007 Euro 25.000

Source: PAR 06 University of Siena

Title: " Allestimento e analisi di un modello Knock-out cellulare per il gene di ritardo mentale FACL4”

Date and cost of the project: 2007-2008 Euro 10.000

Source: Telethon:

Title: “Analysis of candidate genetic regions and peptides and metabolites profile in autism.”

Date and cost of entire project: 2006-2008 Euro77.000

Source: PRIN

Title: “Postgenomic strategies for studying and preventing X-linked mental retardation”

Date and cost of entire project: 2006-2008 Euro 73.586

Source: Telethon

Title: "Analysis of candidate genetic region and peptides and metabolites profile in autism "PI F. Gurrieri GGP06170

Date and cost assigned to AR: 2007-2008 Euro 77.000,00

Source: MUR (FIRB2005)

Title: “Studio e trattamento dei tumori e delle malattie degenerative: sviluppo e produzione di una nuova piattaforma analitica in DHPLC completa di test diagnostici dedicati ai differenti settori applicative in oncologia e nelle malattie degenerative” PI G. Martinelli

Date and cost assigned to AR: 2007-2008 Euro 113.600,0

Source: Associazione Emma ed Ernesto Rulfo per la Genetica Medica 07

Title: "Basi molecolari della sindrome di Rett e dei disordini ad esso correlati"

Date and cost of the project: 2008 Euro 7.500

Source: Italian Ministry of Health

Title: “Genomic structural variation studies in mentally retarded and normal individuals in Italy”.

Date and cost of entire project: 2008-2010 Euro 90.200,00

Source: E-rare (07)

Title: "Performing genotype phenotype correlation on Rett syndrome" as part of the project “European network on Rett syndrome” PI L.Villard

Date and cost of the project assigned to AR: 2008-2010 Euro 13.500,00

Source: A.I.R. (Associazione Italiana Rett)

Title: “Produzione di cellule neuronali da fibroblasti riprogrammati di pazienti con Sindrome di Rett: una tecnologia innovative per testare strategie terapeutiche”.

Date and cost of entire project: 2009-2010 Euro 50.000,00

Source: Monte dei Paschi di Siena

Title: “Protocollo di studio per l'applicazione del test array-CGH in diagnosi prenatale: task force AOUS e territorio.”

Date and cost of entire project: 2010-2011 Euro 50.000,00

Source: Regione Toscana

Title: “Percorso diagnostico-assistenziale per pazienti affetti dalla sindrome di Rett”

Date and cost of entire project: 2010-2011 Euro 10.000,00

Source: Regione Toscana

Title: “Percorso diagnostico-terapeutico per pazienti affetti da Sindrome di Alport e nefropatie correlate”

Date and cost of entire project: 2010-2011 Euro 10.000,00

Source: Regione Toscana

Title: “Ottimizzazione del percorso diagnostico-assistenziale per pazienti affetti da malattie genetiche rare”

Date and cost of entire project: 2010-2011 Euro 10.000,00

Source: IRSF (International Rett syndrome Foundation)

Title: “International Rett syndrome database”

Date and cost of entire project: 2010-2011 $ 10,000

Source: Telethon:

Title: “Cell lines and DNA bank of Rett syndrome and other X-linked mental retardation”

Date and cost of the project: 2007-2012 Euro 213.600,00

Source: Telethon:

Title: "Network of Telethon Genetic Biobanks" GTB07001

Date and cost of the project: 2007-2012 Euro 216.300,00

Source: Telethon

Title: “Congenital Rett syndrome: cellular and mouse models for the study of FOXG1 impact on forebrain neurogenesis.”

Date and cost of entire project: 2009-2012 Euro 293.100,00

Source: MIUR

Title: “Metabolic and vascular characterization in animal models and patients with Rett syndrome”.

Date and cost of the project: 2010-2012 Euro: 29.000,00

Source: AIRett

Title: “Analisi dei geni HLA per valutare la suscettibilità genetica ai vaccini come componente della patogenesi della sindrome di Rett”.

Date and cost of the project: 2011-2012 Euro 25.000,00

Source: Associazione Emma ed Ernesto Rulfo per la Genetica Medica

Title: " Produzione di neuroni da fibroblasti riprogrammati di pazienti con sindrome di Rett: un modello innovativo per testare strategie terapeutiche.”

Date and cost of the project: 2011-2012 Euro 10.000,00

Source: AIRett

Title: “Studio dell’azione moderatrice del complesso ENA/VASP sul difetto sinaptico indotto da mutazioni di MeCP2 in neuroni di topo e ottenuti da cellule staminali pluripotenti indotte (iPSC) da pazienti.”

Date and cost of the project: 2011-2013 Euro 53.200,00

Patents:

- Alessandra Renieri e Ilaria Meloni “Diagnostic and therapeutic tools for X-linked mental retardation syndrome “ International application N° PCT/IT03/00134, issued March 2003.

- Renieri A, Conticello S, Pinto AM, Meloni I, Daga S, Donati F, Croci S, Lopergolo D. “CRISPR-Cas system for gene therapy” (Patent application N° 102018000020230) for the use of CRISPR/Cas9 technology in rare genetic diseases. December 19, 2018.

- Renieri A, Conticello S, Donati F, Niccheri F, Mari F, Papa FT, Lorenzetti FC. “Sistema CRISPRCas per l’editing genomico” (Patent application N° 102018000009431) for the employment of CRISPR/Cpf1 technology for specific delivery of suicide gene in cancer cells mutated in TP53. October 15, 2018.

Memberships

AFFILIATIONS TO SCIENTIFIC SOCIETIES and COMMITEE:

-Societa' Italiana di Genetica Umana (SIGU)

-American Society of Human Genetics (ASHG)

-European Society of Human Genetics (ESHG): board member from 2004 to 2009

-Societa' Italiana di Pedagogia Medica (SIPEM)

-EBMG (European Board of Medical Genetics), a professional organism of ESHG (from 2014)

-SIGU representative within UEMS (Union Européenne des Médecins Spécialistes - European Union of Medical Specialists) from 2013 to end of 2016

- European Society of Human Reproduction and Embryology (ESHRE) since 2016

- Secretary of Clinical Genetics Section within UEMS (Union Européenne des Médecins Spécialistes - European Union of Medical Specialists) from 1st Jauary, 2017

-Coordinator of the Working Group of Clinical Genetics of the Italian Society of Human Genetics (SIGU) from 2014

-Member of Ethical Commitee of Azienda Ospedaliera Universitaria Senese, Siena, Italy from 2016

- Coordinator of Network for Italian Genomes (NIG) from 2017

Other Relevant Information

I coordinate a PhD Course in Genetics Oncology and Clinical Medicine as well as Director of Medical Genetics Specialty School at the University of Siena. I am an expert clinical geneticist (MD, PhD), director of the Medical Genetics Unit at AOUS in Siena with long experience in syndromic and non-syndromic intellectual disabilities (ID). Since 2016 I am member of Ethical Commitee of AOUS.

REVIEWER FOR INTERNATIONAL JOURNALS

- American Journal of Human Genetics

- American Journal of Medical Genetics

- BMC Medical Genetics

- Clinical Chemistry

- Clinical Genetics

- European Journal of Human Genetics

- Journal of Endocrinology
- Journal of Medical Genetics
-Journal of Nephrology
- Nature Genetics
- Nephron
- Pediatric Nephrology

GRANT REVIEWER AND NATIONAL INITIATIVES

- Projects for Wellcome Trust 2002 and 2004
- CV revision “Rientro cervelli” for CRUI 2003 and 2004
- University of Padova 2006
- Evaluator for National Assessment of Research Quality (VQR) 2012 for research products 2004-2010
- Component of Scientific Commitee for National Academic Qualification (ASN) 2012 and 2013
- OTKA proposal for National Scientific Research Fund in Hungary 2013
- Evaluator for National Assessment of Research Quality (VQR) 2016 for research products 2011-2014

EDITORIAL BOARD

- Giornale Italiano di Nefrologia
- Section Editor di European Journal of Human Genetics (EJHG) from 2014