EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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 318 original publications with a total IF >1,600 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 December 2020):

N publications in the last 10ys: 144

N total citations: 10,178

H-index (from Scopus - updated on January 2022): 53

average citations per item: 32.301

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

Over the last 5 years she won 15 grants from national and international competitive calls.Ten grants are currently active, 4 as PI/coordinator:

Source: Fondo Integrativo Speciale per la Ricerca (FISR) - Ministero dell'Universita e della Ricerca

Title: "Editing dell'RNA contro il Sars-CoV-2: hackerare il virus per identificare bersagli molecolari e attenuare l'infezione (HACKTHECOV)"

Duration: 2021-2022;

Role: Partner

Budget: 80.000,00 Euro (19.428,20 Euro to Unisi)

Source: IMI2-2020-23-05, H2020-JTI-IMI2-2020-23-two-stage (IMI2-RIA)

Title: "Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies"

Duration: 2021-2024;

Role: Partner

Budget: 1,.939,251.25 (168,750.00 Euro to Unisi)

Source: Travere Therapeutics, Inc.

Title: "From Benchtop to Preclinical Trials. CRISPR/Cas9 to Correct COL4A5 Mutations Causing Alport Syndrome in in-vitro and in-vivo Dog and Mouse Model."

Duration: 2021-2024;

Role: Coordinator

Budget: $1,061,891.00

Source: Istituto Buddista Italiano Soka Gakkai

Title: "Host genetics and pathogenetic mechanisms of COVID-19 (PAT-COVID)"

Duration: 2021-2022

Role: Partner

Cost of the entire project: 640.000,00 Euro (190.000 Euro to Unisi)

Source: Associazione ILA

Title: "La Biopsia Liquida Come Ricerca Genetica In Pazienti Affetti Da Malformazioni Vascolari"

Duration:2021-2022;

Role: Coordinator

Budget: 30.000 Euro

Source: Bando Ricerca COVID-19 Toscana

Title: "Identificazione delle basi genetiche determinanti la variabilita clinica di COVID-19 nella popolazione italiana (GEN-COVID)"

Duration: 2021-2022

Role: Coordinator

Budget: 500.000,00 Euro (190.000 Euro to coordinator)

Source: AstraZeneca

Title: WES Sequencing Agreement (Service Provider)

Duration: 2020-2021

Role: Coordinator

Budget: 150.000,00 Euro

Source: EU project H2020-SC1-FA-DTS-2018-2020;

Title: "International consortium for integrative genomics prediction (INTERVENE)" - Grant Agreement No. 101016775

Duration: 2021-2025

Role: Partner

Budget: (552,043.75 Euro to Unisi)

Source: Bando Ricerca Salute 2018 - Regione Toscana

Title: "Set-up of a platform for personalized diagnosis of rare kidney diseases (NIKE)"

Duration: 2020-2023

Role: Partner

Budget: 210.000,00 Euro

Source: 3rd EU Health Programme. HP-PJ-2019; Topic PJ-01-2019: Rare disease registries for the European Reference Networks

Title: "ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development".

Duration: 2020-2025

Role: Partner

Budget: 198.119 Euros (10.019 Euro to Alessandra Renieri)

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 Europeenne des Medecins Specialistes - 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 Europeenne des Medecins Specialistes - 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