EUROPEAN CURRICULUM VITAE FORMAT

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Nationality	Italian
Date of birth	14 MARCH 1986
Birthplace	BOLOGNA
Registered in	ORDINE DEI MEDICI CHIRURGHI E DEGLI ODONTOIATRI DI BOLOGNA, REGISTRATION N. 17139
Affiliations	DIMEC - Department of Medical and Surgical Sciences Alma Mater Studiorum – University of Bologna Institute of Genomic Medicine Catholic University of the Sacred Heart, Rome
ORCID	0000-0001-8413-621X

EDUCATION AND TRAINING

September 2019 to date

PhD fellowship in Scienze Biomediche di Base e Sanità Pubblica

Catholic University of the Sacred Heart, Rome, Italy

Project title: “Somatic instability of the CAG strecth in *HTT* gene: explorative evaluation of his activity as a potential biomarker for HD patients”

Università Cattolica del Sacro Cuore-Istituto di Medicina Genomica, Roma, Italia

Supervisor: Prof. Francesco Danilo Tiziano

December 2014 – December 2018
Residency in Medical Genetics
Catholic University of the Sacred Heart, Rome, Italy
Specialization degree with 50/50 cum laude
Title of final dissertation:" Instabilita' somatica del tratto cag del gene *HTT* in sottopopolazioni linfocitarie di pazienti con malattia di Huntington"
Supervisor: Chiar.mo Prof Maurizio Genuardi
Co-supervisors: Prof. Anna Rita Bentivoglio, Prof. Pietro Chiurazzi

July 2013 – August 2013 and October 2013-November 2013
Post-graduate Internship in Medical Genetics
Institute of Medical Genetics
University of Bologna, Bologna, Italy

October 2005 – July 2013
Degree in Medicine and Surgery
University of Bologna, Bologna, Italy
Graduated with 110/110 cum laude
Dissertation in Medical Genetics, title: "Fattori genetici predisponenti al carcinoma mammario giovanile [Genetic factors predisposing to early-onset breast cancer]"
Mentor: Prof. Daniela Turchetti

September 2010– July 2011
Erasmus Scolarship
University of Murcia, Murcia, Spain

September 1999 – July 2004
High School Certification
Liceo Classico Luigi Galvani, Bologna, Italy

WORK EXPERIENCE

July 2022 to date
Junior full-time researcher
Research project entitled: "Genetic characterization and innovative therapies of neurological and neurodevelopmental diseases"
Department of Medical and Surgical Sciences – DIMEC
Alma Mater Studiorum – University of Bologna
Clinical practice at Unit of Medical Genetics
IRCCS Azienda Ospedaliero Universitaria di Bologna
Policlinico S.Orsola-Malpighi

September 2019 to date
Institute of Genomic Medicine
Catholic University of the Sacred Heart, Rome, Italy
PhD fellowship, with the following project: "Somatic instability of the CAG strecth in *HTT* gene: explorative evaluation of his activity as a potential biomarker for HD patients"

September 2021 to February 2022
Unit of Medical Genetics
IRCCS Azienda Ospedaliero Universitaria di Bologna
Policlinico S.Orsola-Malpighi

December 2020 September 2021
Unit of Medical Genetics
AUSL Romagna

Clinical geneticist

January 2019 to December 2020
Institute of Genomic Medicine
Catholic University of the Sacred Heart, Rome, Italy
Project Manager of "Screening neonatale per l'atrofia muscolare spinale nel Lazio ed in Toscana: un progetto pilota di due anni"

March 2019 to September 2019
Unit of Medical Genetics
Fondazione Policlinico Universitario A. Gemelli "IRCCS"
Genetic counselor within the XBioGem partnership

December 2014 to December 2018
Resident MD in Medical Genetics
Institute of Medical Genetics
Catholic University of the Sacred Heart, Rome, Italy
Training in:
Clinical evaluation of patients with intellectual disability with or without syndromic signs
Genetic counselling of patients and families for reproductive risk assessment, recurrence of rare disorders and risks of familial cancer conditions
Management of clinical and molecular data
Genetic counselling of patients and families with Huntington disease (under supervision of both Prof. Anna Rita Bentivoglio and Prof. Marina Frontali)

November 2017 – April 2018
Research Fellowship, under supervision of Prof. Bernhard Georg Landwehrmeyer
Department of Neurology
Ulm University Hospital, Ulm, Germany
Research projects on Huntington disease modifiers genes and prognostic biomarkers
Drafting of HD counseling gudelines

PUBLICATIONS

Simpson-Golabi-Behmel syndrome in a female : a case report and an unsolved issue
Vaisfeld A, Pomponi MG, Pietrobono R, Tabolacci E, Neri G. 2017.
Am J Med Genet Part A 173:285-288.

Guidelines reccomendations for diagnosis and clinical management of Ring14 syndrome-first report of an ad hoc task force
Rinaldi B, Vaisfeld A (co-authors), Amarri S, Baldo C, Gobbi G, Magini P, Melli E, Neri G, Novara F, Pippucci T, Rizzi R, Soresina A, Zampini L, Zuffardi O, Crimi M. 2017.
Orphanet J Rare Dis 11;12(1):69.

A novel truncating variant within exon 7 of KAT6B associated with features of both Say-Barber-Bieseker-Young-Simpson syndrome and genitopatellar syndrome: Further evidence of a continuum in the clinical spectrum of KAT6B-related disorders
Marangi G, Di Giacomo MC, Lattante S, Orteschi D, Patrizi S, Doronzio PN, Riviello FN, Vaisfeld A, Frangella S, Zollino M. 2017.
Am J Med Genet A 176(2):455-459.

Lynch syndrome with exclusive skin involvement: time to consider a molecular definition?
Vaisfeld A, Calicchia M, Pomponi MG, Lucci-Cordisco E, Reggiani-Bonetti L, Genuardi M. 2019.
Fam Cancer. doi : 10.1007/s10689-019-00139-3.

Cromosome 14 deletions, rings and epilepsy genes: a riddle wrapped in a mystery inside an enigma
Vaisfeld A, Spartano S, Gobbi G, Vezzani A, Neri G.
Epilepsia. 2021 Jan;62(1):25-40. doi: 10.1111/epi.16754. Epub 2020 Nov 17

Neuroacanthocytosis in an Italian cohort: clinical spectrum, high genetic variability and muscle involvement
Vaisfeld A, Bruno G, Petracca M, Bentivoglio A, Servidei S, Vita MG, Bove F, Straccia G, Dato C, Di Iorio G, Sampaolo S, Peluso S, De Rosa A, De Michele G, Barghigiani M, Galatolo D, Tessa A, Santorelli FM, Chiurazzi P, Melone M.
Genes (Basel). 2021 Feb 26;12(3):344. doi: 10.3390/genes12030344

Neurological assessment of newborns with spinal muscular atrophy identified through neonatal screening
Pane M, Donati MA, Cutrona C, De Sanctis R, Pirinu M, Coratti G, Ricci M, Palermo C, Berti B, Leone D, Ticci C, Sacchini M, Cerboneschi M, Capasso A, Cicala G, Pera MC, Bravetti C, Abiusi E, Vaisfeld A, Vento G, Tiziano FD, Mercuri E.
Eur J Pediatr. 2022 May 6. doi: 10.1007/s00431-022-04470-3.

Phenotypic spectrum and molecular findings in 17 ATR-X syndrome Italian patients: some new insights
Vaisfeld A, Taormina S, Simonati A, Neri G.
Genes vol. 13,10 1792. 4 Oct. 2022, doi:10.3390/genes13101792

The changing scenario of the molecular diagnosis of spinal muscular atrophy in the era of newborn screening: the experience of a two-year, Italian pilot project
Emanuela Abiusi*, Alessandro Vaisfeld* (*co-autori), Stefania Fiori, Agnese Novelli, Serena Spartano, Maria Vittoria Faggiano, Teresa Giovanniello, Antonio Angeloni, Gianni Vento, Roberta Santoloci, Francesca Gigli, Adele D'Amico, Simonetta Costa, Chiara Ticci, Marta Daniotti, Michele Sacchini, Enrico Bertini, Antonio Lanzone, Giancarlo Lamarca, Maurizio Genuardi, Marika Pane, Maria Alice Donati, Eugenio Mercuri and Francesco Danilo Tiziano on behalf of the Italian SMA-NBS group
Accepted (Journal of Medical Genetics), in print

3'utr deletion of nono leads to corpus callosum anomaly, left ventricular non-compaction and ebstein's anomaly in a male fetus
Maria Grazia Giuffrida, Marina Goldoni, Maria Luce Genovesi,
Giovanna Carpentieri, Barbara Torres, Anca Daniela Deac, Serena Cecchetti,
Anna Martinelli, Alessandro Vaisfeld, Elisabetta Flex, Laura Bernardini
Diagnostics (Basel, Switzerland) vol. 12,10 2354. 28 Sep. 2022, doi:10.3390/diagnostics12102354

BOOK CHAPTERS

Atrofia muscolare spinale: aspetti genetici e patogenetici
Abiusi E, Vaisfeld A, Tiziano FD. 2019.
Giornale di Neuropsichiatria dell'età evolutiva 39 :79-85.

Gli screening neonatali e l'atrofia muscolare spinale: verso nuovi paradigmi diagnostico-assistenziali?
Tiziano FD, Pane M, Donati A, Abiusi E, Vaisfeld A, Mercuri E. Febbraio 2022.
Magazine della Società Italiana di Neonatologia, pag.3.

INVITED SPEAKER (CONFERENCES AND COURSES)

Malattia di Huntington: clinica, difetto genetico e terapia genica.
Vaisfeld A, Bentivoglio AR
Patologie neurodegenerative : clinica, genetica ed approcci diagnostici. Scuola Medica Ospedaliera : corso in videoconferenza su piattaforma web Zoom Cloud Meeting con assegnazione di 8 ECM, 11 Dicembre 2020

Complex contribution to epilepsy in 14q rearrangements
Vaisfeld A
Moving Mountains 2021 Conference
Westminster, Colorado, 7-9 Luglio 2021
Partecipazione da remoto

Sindrome ATR-X: introduzione agli aspetti molecolari
Vaisfeld A
I Convegno ATRX Italia
Lettomanoppello (PE), 9-11 Luglio 2021

Quando l'eccezione conferma la regola: alcuni casi inusuali e la loro soluzione.
Vaisfeld A
Lo screening pilota per la SMA – dal progetto pilota all'estensione nazionale.
In collaborazione con il Gruppo di Lavoro SIGU Sanità
Meeting con assegnazione di 6 ECM, 10 Marzo 2022

Screening genetico neonatale per la SMA: Razionale, stato dell'arte e discussione di alcuni casi inusuali
Vaisfeld A
Nell'ambito del Master in Malattie metaboliche ereditarie e screening neonatale, Alma Mater Studiorum-Università di Bologna,
Corso con partecipazione da remoto, 6 Maggio 2022

Sindrome ATR-X : metodo e razionale per la costruzione di un registro clinico-molecolare
Vaisfeld A, Neri G
II Convegno ATRX Italia
Lettomanoppello (PE), 4-5 Giugno 2021

Ricorrenza di anomalie fenotipiche multiple e ritardo psicomotorio grave in due fratelli [Recurrence of phenotypic abnormalities and severe cognitive impairment in two brothers]
Vaisfeld A, Zollino M.
XVIII Incontro Nazionale di Genetica Medica, 15 February 2016
[Oral communication]

Nuova variante *de novo* nell'esone 7 di *KAT6B* : ulteriore evidenza di un unico spettro fenotipico SBBYSS/GPS
Vaisfeld A, Zollino M, Di Giacomo M, Marangi G, Frangella S, Riviello F, Orteschi D.
XIX Incontro Nazionale di Genetica Clinica, 13-14 February 2017
[Oral communication]

Muir-Torre case report
Vaisfeld A, Genuardi M.
XXIV Incontro di Genetica Oncologica Clinica, 8 July 2016
[Oral communication]
Juvenile Huntington disease : a case presentation
Vaisfeld A, Landwehrmeyer GB.
ERN-RND Winter School 2018 : Diagnostics of Rare Movement Disorders
[Oral communication]

La nostra casistica di pazienti indirizzati ad analisi dei geni TP53, BAP1 e DICER1 negli ultimi 2 anni e mezzo [Our case series of patients addressed to the analysis of TP53, BAP1 and DICER1 genes in the last 2 and a half years].
Vaisfeld A, Brugnoletti F, LucciCordisco E, Genuardi M.
XXVI Incontro di Genetica Oncologica Clinica, 4 July 2018
[Oral communication]

Un caso di overgrowth segmentale senza diagnosi molecolare: le problematiche connesse con una presa in carico tardiva.
Vaisfeld A, Leoni C, Resta N, Zampino N, Onesimo R, Genuardi M.
Gruppo di lavoro SIGU « Patologie legate al pathway mTor : dalla Sclerosi Tuberosa al Cancro ». Bologna, 30 September 2019
[Oral communication]

MEMBERSHIP AND COLLABORATIONS

-Ring 14 International ONLUS (Clinical Collaboration)

-European Huntington's Disease Network (Member)

-ATRX Italia (Clinical Collaboration)

-International Journal for Research and Ethics (Member of Editorial Board)

CERTIFICATIONS

Certification of Good Clinical Practice (NIDA Clinical Trials Network)

UHDRS Motor Rater (Enroll-HD/EHDN)

PATENTS

Development of an all-in-one automatized compact device suitable for genetics/molecular biology and cellular procedures
Tiziano FD, Abiusi E, Vaisfeld A, Coletti S – *patent pending*

PROJECTS PARTICIPATION

Pilot newborn screening for SMA: the italian experience
PI: Tiziano FD
Supported by Biogen Pharmaceuticals

A randomized, double-blind, placebo-controlled, parallel group study to evaluate the efficacy and safety of CNP520 in participants at risk for the onset of clinical symptoms of Alzheimer's Disease (AD)
Local PI: Daniele A
Supported by Novartis

HLA and innate-immunity gene variants as severity predictors of COVID-19: an Italian multi-centric retrospective association study
PI: Cauda R
Sub-investigator: Tiziano FD
Supported by Emergex Vaccine Holding Ltd

Studio di correlazione fenotipo-instabilità somatica dell'allele *HTT* espanso in sottopopolazioni cellulari selezionate, in pazienti affetti da Malattia di Huntington
PI: Chiurazzi P
Sub-investigator: Vaisfeld A

I authorize the use of my personal data in the CV pursuant to italian Legislative Decree 30 June 2003, n. 196 " Codice in materia di protezione dei dati personali " and the GDPR (EU Regulation 2016/679).