



# UNIVERSITÀ DEGLI STUDI DI MILANO

TO MAGNIFICO RETTORE OF UNIVERSITA' DEGLI STUDI DI MILANO

FRANCESCA CORTINI  
CURRICULUM VITAE

## PERSONAL INFORMATION

Surname	CORTINI
Name	FRANCESCA
Date of birth	[03/08/1977]

## PRESENT OCCUPATION

Appointment	Structure
Assegnista ricerca (Tipo B)	Università degli studi di Milano/Ospedale Maggiore Policlinico

## EDUCATION AND TRAINING

Degree	Course of studies	University	year of achievement of the degree
Degree	Degree in Biological Sciences	University of Milan	2004
Specialization			
PhD	Phd in Molecular Medicine	University of Milan	2009
Master			
Degree of medical specialization			
Degree of European specialization			
Other			



## REGISTRATION IN PROFESSIONAL ASSOCIATIONS

Date of registration	Association	City

## FOREIGN LANGUAGES

Languages	level of knowledge
English	B2 (spoken and written)

## AWARDS, ACKNOWLEDGEMENTS, SCHOLARSHIPS

Year	Description of award

## TRAINING OR RESEARCH ACTIVITY

I am a motivated biologist with vast experience in new molecular biologist techniques as Next Generation Sequencing and its applications. I have participated in a number of important collaborations with both Italian and US research groups in genetic screening projects of genes implicated in multifactorial diseases. I am co-author of 30 scientific papers published in international reviews and I participated in International meetings. Member of SFN (<http://sfn.org>).

## PROJECT ACTIVITY

Year	Project
2013-today	Genetic screening by Next Generation Sequencing (Target Enrichment Panel/MiSeq Illumina) of patients with Ehlers Danlos Syndrome
2009-2012	Genetic screening (Sanger Sequencing) and expression analysis (Real time PCR) of patients with AD or FTLD

## PATENTS

Patent



## CONGRESSES AND SEMINARS

Date	Title	Place
November 2016	NGS in Molecular Pathology	Netherlands Cancer Institute, Agilent Technologies and WUSTL
September 2016	Exome analysis using Galaxy	Università degli studi Milano-Bicocca
September 2016	Summer School on Big Data in Precision Medicine	University of Pavia Center for Health Technologies
November 2015	NGS for evolutionary biologists: from basic scripting to variant calling	ELIXIR-ITA in collaboration with CINECA Roma and Sapienza Università di Roma
November 2014	BASIC NEXT GENERATION SEQUENCING (NGS) PROCEDURES	Università dell'Insubria BustoArsizio, Varese, Italy
September 2014	CRS4 s.r.l - Bioinformatics - POLARIS Science and Technology Park Cagliari, Italy	CRS4 s.r.l - Bioinformatics - POLARIS Science and Technology Park Cagliari, Italy
May 2014	3rd COURSE IN NEXT GENERATION SEQUENCING	Accredited by the European Accreditation Council for Consulting Medical Education (EACCME) Bertinoro di Romagna, Italy

Articles in reviews
<b>EHLERS-DANLOS SYNDROME CLASSICAL TYPE: A NOVEL COL5A2 MISSENSE MUTATION WITH POSSIBLE ADDITIVE EFFECT OF A COL5A1 STOP-GAIN MUTATION IN A STRONGLY CORRELATED PHENOTYPE.</b> <b>Cortini F</b> , C Villa, Marinelli B, Franchetti S, M Seia, Pesatori AC, Montano N, Bassotti A. 2018 META GENE, ISSN: 2214-5400.
<b>EPILEPTIC SEIZURES IN AUTOSOMAL DOMINANT FORMS OF ALZHEIMER'S DISEASE.</b> <b>Cortini F</b> , Cantoni C, Villa C. Seizure. 2018 19; 61: 4-7.
<b>PICALM GENE METHYLATION IN BLOOD OF ALZHEIMER'S DISEASE PATIENTS IS ASSOCIATED WITH COGNITIVE DECLINE.</b> Mercurio R, Pergoli L, Galimberti D, Favero C, Carugno M, Dalla Valle E, Barretta F, <b>Cortini F</b> , Scarpini E, Valentina VB, Pesatori AC. J Alzheimers Dis. 2018; 65(1): 283-292.
<b>A RARE CAUSE OF JUVENILE STROKE: EXTRACRANIAL CAROTID ARTERY ANEURYSM WITH VENOUS COMPLETE RECONSTRUCTION OF THE CAROTID BIFURCATION.</b> Domanin M, Lanfranconi S, Romagnoli S, Runza L, <b>Cortini F</b> , Comi GP, Gabrielli L. Pediatr Neurosurg. 2018; 53(4): 275-279.
<b>EHLERS-DANLOS SYNDROMES AND EPILEPSY: AN UPDATE REVIEW.</b> <b>Cortini F</b> , Villa C. Seizure. 2018; 57:1-4.



**A NEW COL3A1 MUTATION IN EHLERS-DANLOS SYNDROME VASCULAR TYPE WITH DIFFERENT PHENOTYPES IN THE SAME FAMILY.**

**Cortini F**, Marinelli B, Romi S, Seresini A, Pesatori AC, Seia M, Montano N, Bassotti A. *Vascular and Endovascular surgery* 2017; 51(3): 141-145.

**NEXT-GENERATION SEQUENCING AND A NOVEL COL3A1 MUTATION ASSOCIATED WITH VASCULAR EHLERS-DANLOS SYNDROME WITH SEVERE INTESTINAL INVOLVEMENT: A CASE REPORT.**

**Cortini F**, Marinelli B, Seia M, De Giorgio B, Pesatori AC, Montano N, Bassotti A. *J Med Case Rep.* 2016 Oct 31;10(1):303.

**CEREBROSPINAL FLUID BIOMARKERS IN PROGRANULIN MUTATIONS CARRIERS**

Carecchio M, Fenoglio C, **Cortini F**, Comi C, Benussi L, Ghidoni R, Borroni B, De Riz M, Serpente M, Cantoni C, Franceschi M, Albertini V, Monaco F, Rainero I, Binetti G, Padovani A, Bresolin N, Scarpini E, Galimberti D. *J Alzheimers Dis.* 2011 Jan 1.

**DNA METHYLATION IN REPETITIVE ELEMENTS AND ALZHEIMER DISEASE**

Bollati V, Galimberti D, Pergoli L, Dalla Valle E, Barretta F, **Cortini F**, Scarpini E, Bertazzi PA, Baccarelli A. *Brain Behav Immun.* 2011 Feb 3.

**CELL-DEPENDENT KINASE INHIBITOR (CDKN) 2A AND 2B GENETIC VARIABILITY IN PATIENTS WITH ALZHEIMER'S DISEASE<sup>[1]</sup><sub>[SEP]</sub>**

**Cortini F**, Fenoglio C, Venturelli E, Villa C, Clerici F, Serpente M, Cantoni C, Fumagalli G, Mariani C, Bresolin N, Scarpini E, Galimberti D. *Journal of Neurology* 2010 Nov 11.

**GRN VARIABILITY CONTRIBUTES TO SPORADIC FRONTOTEMPORAL LOBAR DE-GENERATION<sup>[1]</sup><sub>[SEP]</sub>**

Galimberti D, Fenoglio C, **Cortini F.**, Serpente M, Venturelli E, Villa C, Clerici F, Marcone A, Benussi L, Ghidoni R, Gallone S, Scalabrini D, Restelli I, Boneschi FM, Cappa S, Binetti G, Mariani C, Rainero I, Giordana MT, Bresolin N, Scarpini. *J Alzheimers Dis.* 2010 Jan; 19(1):171-7.

**NOVEL EXON 1 GENE VARIANT IN ALZHEIMER'S DISEASE**

**Cortini F.**, Fenoglio C, Guidi I, Venturelli E, Pomati S, Marcone A, Scalabrini D, Villa C, Clerici F, Dalla Valle E, Mariani C, Cappa S, Bresolin N, Scarpini E, Galimberti D. *Eur J Neurol.* 2008 Oct; 15(10): 1111-7.

TECHNICAL KNOWLEDGE



**MOLECULAR BIOLOGY**

DNA analysis and cloning  
Digestion with restriction enzymes  
Preparation of bacterial culture  
Extraction and purification of plasmidic DNA  
Transcription and mutagenesis site-specific in vitro  
Genomic DNA extraction with specific kit and RNA extraction from total blood with Trizol  
Ficoll Techniques for blood cells isolation  
Employment of Tecan liquid handling (EVO 100, EVO 150) for PCR and NGS  
MLPA (Multiplex Ligation dependent Probe Amplification)  
Direct Sequencing, fragment analysis, mini-sequencing techniques (SnPshot) with ABI310 and ABI3130  
Next Generation Sequencing (MiSeq and NextSeq – Illumina Platform), True Seq Custom Amplicon (Illumina kit, Haloplex (Agilent Technologies), Clinical Exome (Sure Select Focus Exome-Agilent Technologies)  
PCR techniques: Touchdown PCR, Nested PCR, Allelic Specific PCR  
Real time PCR: allelic discrimination, gene expression by ABI7000 and ABI7500  
HRM (High Resolution Melting) and its applications

**CELLULAR BIOLOGY**

Fibroblasts cell culture and its application

**EPIGENETIC**

Methylation studies based on sodium bisulfite treatment of DNA, Pyrosequencing

**BIOINFORMATIC TOOLS**

Haploview, Pawe, Spliceview, Sift, Polyphen, Sequencing Analysis, SeqScape, Gene Mapper, BioEdit, Peak Scanner, Galaxy for analysis Next Generation Sequencing data

**SOFTWARE OS**

Microsoft Office (MS Word, MS Access, MS Excel, Powerpoint), Adobe Photoshop, Windows&Mac

Declarations given in the present curriculum must be considered released according to art. 46 and 47 of DPR n. 445/2000.

The present curriculum does not contain confidential and legal information according to art. 4, paragraph 1, points d) and e) of D.Lgs. 30.06.2003 n. 196.

Place and date: VERCELLI, 18/11/2018

SIGNATURE



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