

**EUROPEAN
CURRICULUM VITAE
FORMAT**



Dichiarazione sostitutiva di certificazione e Dichiarazione sostitutiva dell'atto di notorietà ai sensi del DPR 445/28.12.2000

La sottoscritta PELO ELISABETTA consapevole delle responsabilità penali cui può andare incontro in caso di dichiarazioni mendaci, ai sensi e per gli effetti di cui all'art. 76 del DPR 445/2000 e sotto la propria responsabilità

DICHIARA IL PROPRIO SEGUENTE CURRICULUM VITAE
Ai sensi dell'art. 46 e 47 del DPR 445/2000

PERSONAL INFORMATION

Name **ELISABETTA PELO**
E-mail **peloe@aou-careggi.toscana.it**
Nationality Italian
Date of birth 19/12/1968

WORK EXPERIENCES

- Dates (from – to) Since 1 February 2017 do date
- Name and address of employer Azienda Ospedaliero-Universitaria Careggi – SODc Genetic Diagnostic Unit
Largo Brambilla n.3 50134 Florence Italy
- Type of business or sector Public Hospital
- Occupation or position held Medical doctor, Director of SODc Genetic Diagnostic Unit
- Main activities and responsibilities Genetic counselling; Molecular analysis applied to genomic and cytogenomic disorders for prenatal e post natal diseases; Molecular and cytogenomic test applied to oncohematology disease; Histocompatibility and Immunogenetics laboratory, management of HLA test for transplantation

- Dates (from – to) Since 01 November 2014 to 31 January 2017
- Name and address of employer Azienda Sanitaria Toscana Centro – SOS Medical Genetic
Piazza Santa Maria Nuova Florence Italy
- Type of business or sector Public Hospital
- Occupation or position held Medical doctor Director of SOS
- Main activities and responsibilities Genetic visits for hereditary diseases, oncological diseases, pregnancies at risk for malformations Management of genetic cytogenetic, genomic and clinical exome tests,

- Dates (from – to) Since August 2008 to 30 October 2014
- Name and address of employer Azienda Ospedaliero-Universitaria Careggi – Genetic Diagnostic Unit
Largo Brambilla n.3 50134 Florence Italy
- Type of business or sector Public Hospital
- Occupation or position held Medical doctor
- Main activities and responsibilities Genetic counselling ; Molecular analysis applied to genomic and cytogenomic disorders, oncology disease

- Dates (from – to) December2000- August 2008
- Name and address of employer Azienda Ospedaliero-Universitaria Careggi – Genetic Diagnostic Unit
Largo Brambilla n.3 50134 Florence Italy
- Type of business or sector Public Hospital

- Occupation or position held *Medical doctor*
- Main activities and responsibilities Genetic counselling ; Molecular analysis applied to genetic and cytogenetic disorders

EDUCATION AND TRAINING

- *Dates (from – to)* March 2003- December 2006
- *Name and type of organisation providing education and training* Università degli Studi di Firenze
- *Title of qualification awarded* Scuola di Specializzazione in in Medicina Legale l'Università degli Studi di Firenze
Specialist in forensic pathologist
- *Dates (from – to)* October 1994- Novembre 1998
- *Name and type of organisation providing education and training* Università degli Studi di Firenze
- *Title of qualification awarded* Scuola di Specializzazione in Genetica Medica
Specialist in Medical Genetics
- *Dates (from – to)* November 1987- September 1994
- *Name and type of organisation providing education and training* Università degli Studi di Firenze
- *Title of qualification awarded* Corso di Laurea in Medicina e Chirurgia
Degree in Medical Doctor

PERSONAL SKILLS AND COMPETENCES

MOTHER TONGUE **ITALIAN**

OTHER LANGUAGES

- Reading skills **GOOD**
- Writing skills **SUFFICIENT**
- Verbal skills **SUFFICIENT**

ORGANISATIONAL SKILLS AND COMPETENCES **VERY GOOD SKILLS IN TEAM-WORKING, COORDINATING AND LEADING RESEARCHER PROJECTS**

TECHNICAL SKILLS AND COMPETENCES **GOOD SKILLS IN USING COMPUTERS (WINDOWS, MICROSOFT OFFICE).
VERY GOOD SKILLS IN GENETIC COUNSELLING AND IN USING SPECIFIC SOFTWARES FOR SEQUENCE AND MICROSATELLITE ANALYSIS AND GENOMIC ANALYSIS**

WORKS PAPER

- 1) "Methimazole Embriopathy: Delineation of the phenotype" Clementi M., Di Giannantonio E., Pelo E., Mammi I., Tenconi R. *Am. J. of Med. Gen.* 83:43-46 1999
- 2) Expression and function of gonadotropin-releasing hormone (GnRH) receptor in human olfactory GnRH-secreting neurons: an autocrine GnRH loop underlies neuronal migration" Romanelli RG, Barni T, Maggi M, Luconi M, Failli P, Pezzatini A, Pelo E, Torricelli F, Crescioli C, Ferruzzi P, Salerno R, Marini M, Rotella CM, Vannelli GB.; *J Biol Chem.* 2004 Jan 2;279(1):117-26. Epub 2003 Oct 16.
- 3) " A new ATTR Phe64Ile mutation with late-onset multiorgan involvement" Tarquini R, Perfetto F, Bergesio F, Miliani A, Pace SD, Frusconi S, Minuti B, Pelo E, Torricelli F. *Amyloid.* 2007 Dec;14(4):289-92.
- 4) The Italian National External quality assessment program in molecular genetic testing: results of the VII round (2010-2011). Censi F, Tosto F, Florida G, Marra M, Salvatore M, Baffico AM, Grasso M, Melis MA, Pelo E, Radice P, Ravani A, Rosatelli C, Resta N, Russo S, Seia M, Varesco L, Falbo V, Taruscio D. *Biomed Res Int.* 2013;2013:739010. doi: 10.1155/2013/739010. Epub 2013 Jan 29
- 5) 16p11.2 de novo microdeletion encompassing SRCAP gene in a patient with speech impairment, global developmental delay and behavioural problems. Gerundino F,

- Marseglia G, Pescucci C, Pelo E, Benelli M, Giachini C, Federighi B, Antonelli C, Torricelli F. Eur J Med Genet. 2014 Nov-Dec;57(11-12):649-53.
- 6) Validation of a method for noninvasive prenatal testing for fetal aneuploidies risk and considerations for its introduction in the Public Health System. Gerundino F, Giachini C, Contini E, Benelli M, Marseglia G, Giuliani C, Marin F, Nannetti G, Lisi E, Sberini F, Periti E, Cordisco A, Colosi E, D'ambrosio V, Mazzi M, Rossi M, Staderini L, Minuti B, Pelo E, Cicatiello R, Maruotti GM, Sglavo G, Conti A, Frusconi S, Pescucci C, Torricelli F. J Matern Fetal Neonatal Med. 2017 Mar;30(6):710-716. doi: 10.1080/14767058.2016.1183633. Epub 2016 May 26.
 - 7) Defining the diagnostic effectiveness of genes for inclusion in panels: the experience of two decades of genetic testing for hypertrophic cardiomyopathy at a single center. Mazzarotto F, Girolami F, Boschi B, Barlocco F, Tomberli A, Baldini K, Coppini R, Tanini I, Bardi S, Contini E, Cecchi F, Pelo E, Cook SA, Cerbai E, Poggesi C, Torricelli F, Walsh R, Olivetto I. Genet Med. 2018 Jun 6. doi: 10.1038/s41436-018-0046-0
 - 8) Fundus phenotype in retinitis pigmentosa associated with EYS mutations. Mucciolo DP, Sodi A, Passerini I, Murro V, Cipollini F, Borg I, Pelo E, Contini E, Virgili G, Rizzo S. Ophthalmic Genet. 2018 Oct;39(5):589-602. doi: 10.1080/13816810.2018.1509351. Epub 2018 Aug 28
 - 9) Clin Exp Med 2020 May;20(2):313-320. Review .Epub 2020 Feb 17 Extramedullary blastic transformation of primary myelofibrosis in the form of disseminated myeloid sarcoma: a case report and review of the literature Giacomo Coltro 1 2 3, Francesco Mannelli 1, Federica Vergoni 4, Raffaella Santi 4, Daniela Massi 4 5, Luisa Margherita Siliani 6, Antonella Marzullo 7, Stefania Bonifacio 7, Elisabetta Pelo 7, Annalisa Pacilli 1 2, Chiara Paoli 1 2, Annalisa Franci 1 2, Laura Calabresi 1 2, Alberto Bosi 3, Alessandro Maria Vannucchi 1 2, Paola Guglielmelli 8
 - 10) Ophthalmic Genet 2019 Dec;40(6):514-520. En face OCT in choroideremia Vittoria Murro 1, Dario Pasquale Mucciolo 1, Andrea Sodi 1, Dario Giorgio 1, Ilaria Passerini 2, Elisabetta Pelo 2, Gianni Virgili 1, Stanislao Rizzo 1
 - 11) Long Reads, Short Time: Feasibility of Prenatal Sample Karyotyping by Nanopore Genome Sequencing Niccolò Bartalucci 1, Simone Romagnoli 1 2, Elisa Contini 1, Giuseppina Marseglia 3, Alberto Magi 4, Paola Guglielmelli 1, Elisabetta Pelo 3, Alessandro M Vannucchi Clin Chem 2019 Dec;65(12):1605-1608.
 - 12) Nano-GLADIATOR: real-time detection of copy number alterations from nanopore sequencing data Alberto Magi 1, Davide Bolognini 2, Niccolò Bartalucci 3, Alessandra Mingrino 2, Roberto Semeraro 2, Luna Giovannini 2, Stefania Bonifacio 4, Daniela Parrini 4, Elisabetta Pelo 4, Francesco Mannelli 3, Paola Guglielmelli 3, Alessandro Maria Vannucchi Bioinformatics 2019 Nov 1;35(21):4213-4221.
 - 13) Defining the diagnostic effectiveness of genes for inclusion in panels: the experience of two decades of genetic testing for hypertrophic cardiomyopathy at a single center Francesco Mazzarotto # 1 2 3, Francesca Girolami # 4 5, Beatrice Boschi 4 5, Fausto Barlocco 4, Alessia Tomberli 4, Katia Baldini 4, Raffaele Coppini 6, Ilaria Tanini 4, Sara Bardi 5, Elisa Contini 5, Franco Cecchi 7, Elisabetta Pelo 5, Stuart A Cook 8 9 10, Elisabetta Cerbai 6, Corrado Poggesi 7, Francesca Torricelli 5, Roddy Walsh # 8, Iacopo Olivetto # 4 Genet Med 2019 Feb;21(2):284-292.
 - 14) RPE65-associated inherited retinal diseases: consensus recommendations for eligibility to gene therapy Andrea Sodi # 1, Sandro Banfi # 2 3, Francesco Testa # 4, Michele Della Corte, Ilaria Passerini 6, Elisabetta Pelo 6, Settimio Rossi 5, Francesca Simonelli 5, Italian IRD Working Group Orphanet J Rare Dis 2021 Jun 4;16(1):257

La sottoscritta è a conoscenza che, ai sensi dell'art. 26 della legge 15/68, le dichiarazioni mendaci, la falsità negli atti e l'uso di atti falsi sono puniti ai sensi del codice penale e delle leggi speciali.

Inoltre, la sottoscritta ai sensi e per gli effetti del Regolamento UE 2016/679 sul trattamento dei dati personali e del precedente D. Lgs. n. 196/2003, autorizza il trattamento dei dati personali.

Florence, 2023 December 15th

Firma

