Curriculum Vitae

Personal Information

Name	Tommaso Pippucci
Address	Via Albertoni 15, 40138 Bologna, Italia
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Nationality	Italian

Work Experience

2018- *01/12/2018 -*	U.O. Genetica Medica (Director: *Prof. Marco Seri*) IRCCS Azienda Ospedaliero-Universitaria di Bologna, Policlinico di Sant'Orsola Via Albertoni 15, 40138 Bologna, Italia **Dirigente Biologo di Laboratorio (Laboratory Chief Officer, Head of Computational Genomics)**
2017 *15/6/2017 – 15/9/2017*	Institute for Genomic Medicine (Director: *Prof. David B. Goldstein*) Columbia University, New York, NY, U.S. **Short-term Fellow of the European Molecular Biology Organization (EMBO)** Research Project : Identification of somatic mutations in malformations of cortical development Supervisor: *Dr. Erin Heinzen*
2015-2018 *26/01/2015 – 30/11/2018*	U.O. Genetica Medica (Director: *Prof. Marco Seri*) Azienda Ospedaliero-Universitaria di Bologna, Policlinico di Sant'Orsola Via Albertoni 15, 40138 Bologna, Italia **Contratto di Collaborazione Autonoma (Contract Researcher)** Research Project: Clinical Exome: generation and analysis of exome data in diagnostics for the clinico-molecular characterization of Mendelian diseases Supervisor: *Prof. Marco Seri*
2015-2016 *15/01/2015 – 14/01/2016*	Dipartimento di Scienze Ginecologiche, Ostetriche e Pediatriche Università di Bologna, Italia **Contratto di Collaborazione Coordinata e Continuativa (Research Consultant)** Research Project: Developing Medical Genetics Education Through Curriculum Reforms and Establishment of postgraduate Training Programs Art. 7, comma 6, D.LGS. 165/2001 Supervisor: *Prof. Marco Seri*
2012-2014 *01/06/2012– 30/11/2014*	U.O. Genetica Medica (Director: *Prof. Marco Seri*) Azienda Ospedaliero-Universitaria di Bologna, Policlinico di Sant'Orsola Via Albertoni 15, 40138 Bologna, Italia **Contratto di Collaborazione Autonoma come vincitore del programma Giovani Ricercatori del Ministero della Salute Italiano**

	(Principal Investigator for the Young Researchers program of the Italian Ministry of Health) Research Project: Classical and innovative strategies for the identification of genetic defects at the basis of rare forms of partial epilepsy
2010-2012 *06/11/2010– 05/11/2011* *09/11/2011 – 31/05/2012*	Dipartimento di Scienze Ginecologiche, Ostetriche e Pediatriche Università di Bologna, Italia **Assegnista di Ricerca "Senior" (Senior Post-Doctoral Fellow)** Art. 51, comma 6, L. 449/1997 Supervisor: *Prof. Marco Seri*
2008-2010 *10/09/2008 – 09/09/2009* *06/11/2009 – 05/11/2010*	Dipartimento di Scienze Ginecologiche, Ostetriche e Pediatriche Università di Bologna, Italia **Assegnista di Ricerca (Junior Post-Doctoral Fellow)** Art. 51, comma 6, L. 449/1997 Supervisor: *Prof. Marco Seri*
2007-2008 *01/09/2007 – 31/08/2008*	INSERM U535, Génétique epidémiologique et structure des Populations Humaines (Director: *Prof. Francoise Clerget-Darpoux)* Hôpital Paul Brousse, Villejuif, France **Visiting Post-Doctoral Fellow** Supervisor: *Drs. Emmanuelle Genin and Drs. Anne-Louise Leutenegger*
2007-2008 *01/09/2007 – 31/08/2008*	Dipartimento di Medicina Interna, Cardioangiologia ed Epatologia Università di Bologna, Italia **Assegnista di Ricerca (Junior Post-Doctoral Fellow)** Art. 24, comma 3, lettera a), L. 30.12.2010, n. 240 Supervisor: *Prof. Marco Seri*

Education

2012 *19/03/2012*	**Specializzazione in Genetica Medica,** 70/70 *cum laude* **(Postgraduate specialization in Medical Genetics)**, Università degli Studi di Firenze, Italia Supervisor: *Prof. Maurizio Genuardi* Dissertation topic: *NEXT GENERATION SEQUENCING IN SINDROMI ULTRARARE LEGATE AL CROMOSOMA X*
2007 *12/06/2007*	**Dottorato di Ricerca in Biochimica (PhD in Biochemistry**, Genomics and Post-Genomics Program**)** Università degli Studi di Bologna, Italia Supervisor: *Prof. Giovanni Romeo* Dissertation topic: *L'analisi di Linkage come metodo per la mappatura di malattie genetiche: studio di famiglie con paraparesi spastica ereditaria e anomalie del corpo calloso*
2003 *25/06/2003*	**Laurea in Scienze Biologiche,** 110/110 *cum laude* **(Master Degree in Biological Sciences)** Università degli Studi di Firenze, Italia

	Supervisor: *Dott.ssa Francesca Torricelli*
Research Experience	Bioinformatic analysis of Next Generation Sequencing data Development of tools for analysis of Exome/Genome data Identification of susceptibility genes in complex disorders Identification of mutations and genes in Mendelian disorders
Languages	
Italian	Mother Tongue
English	Reading: fluent Writing: fluent Speaking: fluent
French	Reading: fluent Writing: basic Speaking: basic

Research Projects

Horizon 2020 Framework Programme, H2020-SC1-PHE-CORONAVIRUS-2020-2 Pan-European COVID-19 cohorts	
Project ID	
Title	CONNECTING EUROPEAN COHORTS TO INCREASE COMMON AND EFFECTIVE RESPONSE TO SARS-COV-2 PANDEMIC: ORCHESTRA
Role	**WP6 Member and Member of the Data Analysis Coordination Unit**
Activity	Analysis of host genomics and organization and implementation of the molecular data flow
Ministero Italiano della Salute, Programma di Ricerca Finalizzata 2018	
Project ID	RF-2018-12366314
Title	Whole Genome Sequencing into the diagnostic workflow of rare diseases: a cost-effectiveness evaluation in a heterogeneous population of patients with inconclusive Whole Exome Sequencing
Role	**Task Coordinator**
Activity	Supervision, design and implementation of the bioinformatic workflow for Whole Genome Sequencing data analysis
Trans-European Mobility Programme for University Studies 2013	
Project ID	EACEA Tempus IV MedGen
Title	Developing Medical Genetics Education Through Curriculum Reforms and Establishment of postgraduate Training Programs
Role	**Task Coordinator**
Activity	Capacity-building activity through planning, desing and implementation of workshops on analysis of Next Generation Sequencing data organized at coordinator site (Università di Bologna) and partner sites (Yerevan University and Tel Aviv

	University)
Fondazione Telethon Call for Applications 2013	
Project ID	GP13200A
Title	In-depth clinical and genetic study of familial and sporadic patients with Nocturnal Frontal Lobe Epilepsy (NFLE): identification of new genes by WES in 192 cases negative for mutations in the neuronal nicotinic acetylcholine receptor subunits genes
Role	**Unit Coordinator**
Activity	Supervision of whole exome sequencing and targeted resequencing studies and statistical analysis of the data
Fondazione Telethon Call for Applications 2010	
Project ID	GP10089B
Title	Identification of new disease-causing genes in hereditary spastic paraplegia
Role	**Staff Member**
Activity	Analysis of whole exome sequencing data in families with recurrence of spastic paraplegia
Fondazione Telethon Call for Applications 2010	
Project ID	GP10121
Title	A new gene for inherited thrombocytopenias: clinical, pathogenetic and pharmacological studies
Role	**Staff Member**
Activity	Molecular and functional characterization of *ANKRD26* 5'UTR mutations in patients with inherited thrombocytopenia
Ministero Italiano della Salute, Programma Giovani Ricercatori 2009	
Project ID	GR-2009-1574072
Title	Classical and innovative strategies for the identification of genetic defects at the basis of rare forms of partial epilepsy
Role	**Principal Investigator, Project Coordinator**
Activity	Supervision of molecular genetic studies and statistical, bioinformatic analyses of genotyping, molecular karyotyping and next generation sequencing data in families with recurrence of focal epilepsy
Ministero Italiano della Salute, Programma Strategico 2009	
Project ID	RFPS-4-631972
Title	Genetic Bases of Birth Defects
Role	**Staff Member**
Activity	Candidate gene resequencing, homozygosity mapping, linkage analysis
Ministero Italiano dell'Università e Ricerca, Programmi di Ricerca Scientifica di Rilevante Interesse Nazionale 2006	
Project ID	2006063820_003

Title	Linkage studies in families with recurrence of a recessive form of hereditary spastic paraplegia and thin corpus callosum: analysis of the locus on chromosome 15q13-q15 and search for new disease loci
Role	**Staff Member**
Activity	Microsatellite genotyping, haplotype reconstruction, LOD score calculation, linkage analysis

Organization of Conferences, Courses and Workshops

Analisi e Interpretazione Dati nella Genomica Clinica	
Held by	Società Italiana di Genetica Umana
Location	Università Cattolica del Sacro Cuore, Roma, Italia (22/06/2018); Università di Genova, Genova, Italia (21/09/2018); Università di Bari, Bari, Italia (03/06/2019); XXII Congresso della Sociatà Italiana di Genetica Umana, Catania (27/10/2019)
Editions	-
Years	2018-2019
Role	**Director and Workshop Instructor**
European School of Genetic Medicine course in NGS and Clinical Genomics	
Held by	European Society of Human Genetics
Location	CEUB, Bertinoro, Italia/On-line
Editions	I-IV
Years	2017-2019, 2021
Role	**Co-director and Workshop Instructor**
EACEA Tempus IV MedGen Project	
Held by	Università di Bologna, Italia; Yerevan University, Armenia; Tel Aviv University, Israel
Location	Policlinico di Sant'Orsola, Bologna, Italia (9-13, 16-25/11/2015; Yerevan State Medical University, Armenia (28/03-03/04/2016); Beilinson Hospital, Rabin Medical Center, Petah-Tikva, Israel (04-07/04/2016)
Editions	-
Years	2015-2016
Role	**Coordinator of training activities on Next Generation Sequencing data analysis and Bioinformatics; Workshop Instructor**
L'NGS nella diagnostica (22-23/10/2020); NGS, varianti geniche e studi funzionali (24-25/10/2019); NGS nel laboratorio di genetica umana (04-05/10/2018); NGS e le sue applicazioni (9-10/06/2016); Patologia Mendeliana e NGS (28-29/05/2015); NGS, NIPT, Real Time PCR (09-10/06/2014)	
Held by	Scuola Medica Ospedaliera
Location	*Various Locations*, Roma, Italia/On-line

Editions	-
Years	2014, 2015, 2016, 2018, 2019, 2020
Role	**Faculty Member**

European School of Genetic Medicine course in Next Generation Sequencing

Held by	European Society of Human Genetics
Location	CEUB, Bertinoro, Italia
Editions	III-V
Years	2013-2016
Role	**Co-director and Workshop Instructor**

Corso di Alta Formazione in Elementi di Bioinformatica per l'Analisi di Dati NGS

Organization	Azienda Ospedaliero-Universitaria Careggi, Regione Toscana, Italia
Editions	I-IX
Years	2011-2015
Role	**Co-director, Seminar Speaker and Workshop Instructor**

Teaching Experience

Professore a contratto (Lecturer)

Institution	Università di Bologna
Course	**Medical Genomics** (Settore concorsuale MED/03) Ore annuali (Hours per year): 62; CFU: 6
Academic Years	2019-2020, 2020-2021, 2021-2022
Program	Laurea Triennale in Lingua Inglese in **Genomics** (BsC in **Genomics** held in English)

Professore a contratto (Lecturer)

Institution	Università di Bologna
Course	**Medical Genetics** (Settore concorsuale MED/03) Ore annuali (Hours per year): 32
Academic Years	2017-2018, 2018-2019, 2019-2020
Program	Diploma di Specializzazione in **Genetica Medica** (Postgraduate Specialization in **Medical Genetics**)

Professore a contratto (Lecturer)

Institution	Università di Firenze
Course	**Genomica ed epigenomica applicata alla clinica** (Settore concorsuale MED/03) Ore annuali (Hours per year): 18; CFU: 3
Academic Years	2020-2021
Program	Master di II livello in **Biomedicina Computazionale**

Supervising and Tutoring Experience

Tutoring of PhD students in the *Data analysis and interpretation in molecular medicine: from databases to artificial intelligence* Course – Module 1: Genomics (20-21/06/2019), Programs in "Medicina Traslazionale" and "Bioingegneria e Bioinformatica", University of Pavia, Pavia, Italia

Correlatore (**Co-advisor**) for:

- M.D. thesis entitled *Analisi di dati di Next Generation Sequencing nell'indagine diagnostica delle epilessie*, Corso di Laurea in Medicina e Chirurgia, A.A. 2019/2020, Università di Bologna, Italia
- B.Sc. thesis entitled *Epilessia parziale con aura uditiva: studio del coinvolgimento di mosaicismi somatici nel gene LGI1*, Corso di Laurea Triennale in Biotecnologie, A.A. 2015/2016, Università di Bologna, Italia
- M.Sc. thesis entitled *Comparazione della performace di variant caller per l'identificazione di SNV a mosaic da dati di Next Generation Sequencing in trio,* Corso di Laurea Magistrale in Biotecnologie Mediche, A.A. 2015/2016, Università di Bologna, Italia

Relatore (**Advisor**) for:

- B.Sc. thesis entitled *Accurate identification of low allelic frequency mosaicism in targeted sequencing experiments*, Corso di Laurea Triennale in Genomics, A.A. 2019/2020, University of Bologna, Italia
- B.Sc. thesis entitled *Calling copy number variations in a collection of 281 exomes from patients with epileptic disorders*, Corso di Laurea Triennale in Genomics, A.A. 2020/2021, University of Bologna, Italia

Talks at Conferences and Congresses

Selected by the Scientific Program Committees

SSBP1 mutations cause a complex optic atrophy spectrum disorder with mitochondrial depletion
European Society of Human Genetics Conference 2019
15-18/06/2019
Gothenburg, Sweden

A homozygous truncating NOTCH3 gene mutation is associated with arteriopathy and cavitating leukoencephalopathy
VIII International Congress on Vascular Dementia
17-20/10/2013
Athens, Greece

H3M2: an algorithm for the detection of biologically meaningful ROHs from NGS data
XVI Congresso della Società Italiana di Genetica Umana
25-28/09/2013
Roma, Italia

Mendelian inheritance as a filtering strategy
XV Congresso della Società Italiana di Genetica Umana
21-23/11/2012
Sorrento, Italia

X-chromosome exome in ultrarare disorders
XIV Congresso della Società Italiana di Genetica Umana:
13-16/11/2011
Milano, Italia

Mutations in the 5' UTR of ANKRD26, the Ankirin Repeat Domain 26 Gene, cause an Autosomal-Dominant form of inherited Thrombocytopenia, THC2
European Society of Human Genetics Conference 2011
28/05-01/06/2011
Amsterdam, the Netherlands

As Invited Speaker

Exome sequencing: from theory to practice with a focus on epilepsy and rare neurodevelopmental disorders
Catania International Summer School of NeuroScience in *Neurogenetics and Neurogenomics*
22-26/07/2019
Catania, Italia

Strategie d'analisi per le malattie autosomiche recessive e X-linked
XVIII Congresso della Società Italiana di Genetica Umana
21-24/10/2015
Rimini, italia

Sequenziamento esomico per la ricerca di nuovi geni-malattia: resa diagnostica e criteri di inclusione dei pazienti
XVII Congresso della Società Italiana di Genetica Umana
30-31/10/2014
Bologna, Italia

Whole exome sequencing, homozygosity, autozygosity
European Biotechnology Thematic Network Association Congress
15-18/05/2014
Lecce, Italia

EX-HOM: exome sequencing in small consanguineous pedigrees
European Biotechnology Thematic Network Association Congress
28/09-01/10/2011
Istanbul, Turkey

Awards

Fellowships

European Molecular Biology Organization **Short Term Fellowship** for a 3-months visit (June-September 2017) to Dr. Erin Heinzen Laboratory, Institute for Genomic Medicine, Columbia University, New York, N.Y., USA with a project on the *Identification of brain-specific mosaic mutations in malformations of cortical development and epilepsy*

Grants

Ministero Italiano della Salute, **Programma Giovani Ricercatori** (**Young Investigator Award**) 2009, Project GR-2009-1574072 entitled "Classical and innovative strategies for the identification of genetic defects at the basis of rare forms of partial epilepsy"

Fondazione Telethon Call for Applications 2013, Project GP13200A entitled "In-depth clinical and genetic study of familial and sporadic patients with Nocturnal Frontal Lobe Epilepsy (NFLE): identification of new genes by WES in 192 cases negative for mutations in the neuronal nicotinic acetylcholine receptor subunits genes"

Honors

Abilitazione Scientifica Nazionale a Professore di II Fascia (Scientific Qualification as

Associate Professor in **Genetica Medica** (06/A1) (**Medical Genetics**), **Genetica** (05/I1) (**Genetics**), **Biologia Applicata** (05/F1) (**Applied Biology**)

Memberships

2016-	Lega Italiana Contro l'Epilessia
27/01/2016-	Albo dell'Ordine Nazionale dei Biologi
2017-	Società Italiana di Genetica Umana

Engagement in Public Understanding of Genetics

- **Annual Lecture** on topics related to the use of DNA sequencing technologies and analysis in medicine to the students of the Istituto Tecnico Tecnologico Statale "Guglielmo Marconi", Forlì (FC), 2018, 2019, 2020, 2021
- **Article** entitled *Anche i virus sbagliano*, published in Focus Junior, n°3 March 2021
- **Lecture** entitled *Gli studi di Genetica*, 3rd European Workshop on Pallister-Killian syndrome organized by "Associazione Italiana Sindrome di Pallister-Killian", 6-8/09/2019, Sassuolo (MO), Italia
- **Lecture** entitled *Dalle medicine per tutti alla Medicina Personalizzata - Come la scienza sta cambiando l'utopia della salute,* invited by Associazione "Amici della Primo Levi Valle del Reno", 23/02/2018, Casalecchio di Reno (BO), Italia
- **Cycle of Conferences** entitled *Si nasce o si diventa? Fino a che punto siamo condizionati dai nostril geni?*, invited by Università Primo Levi, 08-29/03/2017, Castiglione dei Pepoli (BO), Italia
- **Lecture** entitled *Geni in vendita. Quanto vale il nostro DNA?,* organized by Associazione "Amici della Primo Levi Valle del Reno", 07/10/2016, Casalecchio di Reno (BO), Italia

Service

Editorial activities	
2019-2020	**Review Editor** for *Frontiers in Genetics* and *Frontiers in Pediatrics,* Section *Genetics of Common and Rare Diseases*
2020-	**Associate Editor** for *Frontiers in Genetics* and *Frontiers in Pediatrics,* Section *Genetics of Common and Rare Diseases*
	Reviewer for *Bioinformatics, BMC Medical Genetics, Brain, Clinical Genetics, Epilepsia, Epilepsy Research, European Journal of Human Genetics, Gene, Human Heredity, Human Genomics, Human Mutation, Genes and Frontiers in Pediatrics*
Scientific Societies	
2019	European Society of Human GeneticsConference**, Abstract Evaluator**
2018	European Society of Human Genetics Conference, **Member** of the **Poster Jury**
2017	XX Congresso Nazionale di Genetica Umana, **Member** of the **Scientific Program Committee**
2016-	Lega italiana Contro l'Epilessia**, Member** of the **Commissione**

	Genetica (**Genetic Committee**)
Scientific Impact	*ORCID ID*: 0000-0001-7737-7963 Scientific Communications published as Articles, Reports, Letters, Reviews on international scientific peer-reviewed journals: 94 Published as 1st Author: **11** Published as Corresponding/Last Author: **12** Global Impact Factor (IF): **574,571** Mean IF: **6,11** H-Index (Source: SCOPUS): **28** Total Citations (Source: SCOPUS): **2615**
Selected publications	**Pippucci T**, Savoia A, Perrotta S, Pujol-Moix N, Noris P, Castegnaro G, Pecci A, Gnan C, Punzo F, Marconi C, Gherardi S, Loffredo G, De Rocco D, Scianguetta S, Barozzi S, Magini P, Bozzi V, Dezzani L, Di Stazio M, Ferraro M, Perini G, Seri M, Balduini CL. *Mutations in the 5'UTR of the ankirin repeat domain 26 gene (ANKRD26) cause an autosomal dominant form of inherited thrombocytopenia (THC2)*. American Journal of Human Genetics, 2011 Jan 7;88(1):115-20. **Pippucci T***, Maresca A*, Magini P, Cenacchi G, Donadio V, Palombo F, Papa V, Incensi A, Gasparre G, Valentino ML, Preziuso C, Pisano A, Ragno M, Liguori R, Giordano C, Tonon C, Lodi R, Parmeggiani A, Carelli V, Seri M. *Homozygous NOTCH3 null mutation and impaired NOTCH3 signaling in recessive early-onset arteriopathy and cavitating leukoencephalopathy.* EMBO Mol Med. 2015 Apr 13;7(6):848-58 Magi A*, Tattini L*, Palombo F, Benelli M, Gialluisi A, Giusti B, Abbate R, Seri M, Gensini GF, Romeo G, **Pippucci T**. *H3M2: detection of runs of homozygosity from whole-exome sequencing data.* Bioinformatics. 2014 Oct 15;30(20):2852-9. Magini P, Smits DJ, Vandervore L, Schot R, Columbaro M, Kasteleijn E, van der Ent M, Palombo F, Lequin MH, Dremmen M, de Wit MCY, Severino M, Divizia MT, Striano P, Ordonez-Herrera N, Alhashem A, Al Fares A, Al Ghamdi M, Rolfs A, Bauer P, Demmers J, Verheijen FW, Wilke M, van Slegtenhorst M, van der Spek PJ, Seri M, Jansen AC, Stottmann RW, Hufnagel RB, Hopkin RJ, Aljeaid D, Wiszniewski W, Gawlinski P, Laure-Kamionowska M, Alkuraya FS, Akleh H, Stanley V, Musaev D, Gleeson JG, Zaki MS, Brunetti-Pierri N, Cappuccio G,

Davidov B, Basel-Salmon L, Bazak L, Shahar NR, Bertoli-Avella A, Mirzaa GM, Dobyns WB, **Pippucci T**$, Fornerod M, Mancini GMS$. *Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis.* Am J Hum Genet. 2019 Sep 5. pii:S0002-9297(19)30309-X.

Del Dotto V, Ullah F, Di Meo I, Magini P, Gusic M, Maresca A, Caporali L, Palombo F, Tagliavini F, Baugh EH, Macao B, Szilagyi Z, Péron C, Gustafson MA, Khan K, La Morgia C, Barboni P, Carbonelli M, Valentino ML, Liguori R, Shashi V, Sullivan JA, Nagaraj S, El-Dairi M, Iannaccone A, Cutcutache I, Bertini E, Carrozzo R, Emma F, Diomedi-Camassei F, Zanna C, Armstrong M, Page MJ, Boesch S, Wortmann SB, Kopajtich R, Stong N, Sperl W, Davis E, Copeland WC, Seri M, Falkenberg M, Prokisch H*, Katsanis N*, Tiranti V*, **Pippucci T**$*, Carelli V$*. *SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder*. J Clin Invest. 2019 Sep 24. pii: 128514.

* Equal contribution
§ Corresponding Author