Allegato B (for publication)

SIMONA PETRUCCI
Curriculum Vitae

Place: Rome
Date: 11th June 2023

Part I – General Information

Full Name	Simona Petrucci
Citizenship	Italian
Spoken Languages	Italian, English

Part II – Education

Type	Year	Institution	Notes (Degree, Experience,…)
University graduation	2006	Sapienza, University of Rome	Grades 110/110 "*cum laude*", degree thesis title: "Use of functional magnetic resonance imaging (fMRI) for the study of patients with epilepsy: localization cerebral areas involved in the genesis of critical and intercritical activity", tutor Prof. Anna Teresa Giallonardo.
Medical Licensure	2007	Order of Physicians of Ascoli Piceno, then transferred to the Order of Physicians of Rome	Medical License Number 62507
Specialty	2014	Sapienza, University of Rome	Specialty Degree in Medical Genetics, grades 70/70 "cum laude", thesis title: "Genetics of Parkinson's disease in Italy. Frequency and phenotypic characterization of Mendelian forms". Tutor Prof. Antonio Pizzuti; co-tutor Prof.sa Enza Maria Valente.
PhD	2018	Sapienza, University of Rome	Clinical and Experimental Neurosciences, thesis, evaluation optimus "cum laude",thesis title "Frequency,

			clinical and biochemical characteristics of Parkinson's disease associated with mutations *GBA* gene variants in the Italian population", tutor prof Alfredo Beradelli, co-tutor Enza Maria Valente.
National Scientific qualification as associate professor	2021	University and Research Ministry	Academic Recruitment Field 06/A1 – Medical Genetics MED03 (2021-2032)

Part III – Appointments

IIIA – Academic Appointments

Start	End	Institution	Position
2018	2023	Department of Clinical and Molecular Medicine, "Sapienza", University of Rome	Fixed-term research assistant, type A

IIIB – Other Appointments

Start	End	Institution	Position
2019	ongoing	AOU Sant'Andrea Hospital	Medical assistant at the UOD of Medical Genetics and Advanced Cellular Diagnostics (genetic counseling in postnatal clinical genetics, oncogenetics, cardiogenetics, neurogenetics; molecular diagnosis of hereditary forms of cancer; molecular diagnosis of hereditary forms of cardiomyopathy arrhythmias in adults; molecular diagnosis of CADASIL and Huntington disease)
2019	ongoing	AOU Sant'Andrea Hospital	Member of the Multidisciplinary Team (MDT) of the diagnostic and therapeutic pathway (PDTA) of breast cancer.
2021	ongoing	AOU Sant'Andrea Hospital	Member of the Multidisciplinary Team (MDT) of the diagnostic and therapeutic pathway (PDTA) of familial hereditary tumors.
2021	ongoing	Italian Huntington Disease Association (AICH) – Rome ONLUS	Medical genetics consultant: counselling, clinical and molecular

			diagnosis Huntington disease patients
2020	2022	Istituto Superiore di Sanità-Ministry of Health	Medical genetics consultant, project "Identification of germline and somatic molecular alterations that predispose to and participate in the pathogenesis of anaplastic large cell lymphoma associated with breast implants", PI prof.sa Arianna Di Napoli
2018	2020	IRCCS CSS-Mendel	Research consultant in studies aimed at the genotypic and phenotypic characterization of rare genetic syndromes.
2014	2018	IRCCS CSS-Mendel	Clinical geneticist (prenatal and postnatal clinical genetic counselling)

Part IV – Teaching experience

Year	Institution	Lecture/Course
2015	"Sapienza", University of Rome	Seminar "Movement disorders in developmental age: genetic components".
2018-2023	"Sapienza", University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course "Morphological and functional bases of the cell" (code 1047950) in the Nursing study course Rome - ASL ROMA 2 L/SNT1 (code 30014, Faculty of Medicine and Psychology).
2018-2023	"Sapienza", University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course of "Paediatrics" (code 1035902), in the course of Medicine and Surgery LM-41, (code 30033, Faculty of Medicine and Psychology).
2018-2023	"Sapienza", University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course "Genetic pathology and related techniques" (code 1041980) in the Biomedical Laboratory Techniques study course L/SNT3 (code 30020, Faculty of Medicine and Psychology).
2019-2023	"Sapienza", University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course "Morphological and functional bases of the cell" (code 1047950, 1 CFU) in the Nursing study course Rome- ASL Roma 3 - Ostia L /SNT1 (code 30013, Faculty of Medicine and Psychology).
2019-2022	"Sapienza", University of Rome	Teaching of Medical Genetics (SSD/MED03) in the integrated course "Professional relationship-obstetric counseling and prevention in obstetrics and gynecology"

		(code 1036251, 1 CFU) in the course Obstetrics studies Rome-L/SNT1 (code 30016, Faculty of Medicine and Psychology).
2019-2023	“Sapienza”, University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course "Morphological and functional bases of the cell" (code 1047950) in the Nursing study course Rome- Celio L/SNT1 (code 30015, Faculty of Medicine and Psychology).
2019-2023	“Sapienza”, University of Rome	Medical Genetics teaching (SSD/MED03) in the integrated course "Pathology and general pathophysiology" (code 10592838) in the Medicine and Surgery course - Rome Sant'Andrea Hospital LM-41 (code 30033, Faculty of Medicine and Psychology).
2019-2023	“Sapienza”, University of Rome	Medical Genetics and Applied Biology teaching (SSD/MED03) in the integrated course "Morphological and functional bases of the cell" (code 1034829) in the course of Occupational Therapy L/SNT2 (code 29989, Faculty of Medicine and Dentistry).
2018-2019	“Sapienza”, University of Rome	Tutor of the degree thesis “Breast and ovarian hereditary cancer: frequency, clinical characteristics and management of subjects with pathogenic variants in the *BRCA1* and *BRCA2* genes" [LMCU - Ordin. 2010] Faculty of Medicine and Psychology.
2019-2020	“Sapienza”, University of Rome	Co-tutor of the degree thesis “"Electro-clinical phenotype and genetic correlation in GLUT1 deficiency epilepsies" [LMCU - Ordin. 2014]. Faculty of Medicine and Psychology.
2022-2023	“Sapienza”, University of Rome	External tutor of the degree thesis “The role of the geneticist nurse in the field of rare diseases in the era of genomics”, L/SNT1 (code 29865) Faculty of Pharmacy and Medicine, Medicine and Dentistry.

Part V - Society memberberships, Awards and Honors

Year Title	
2022-ongoing	Member of the Italian Society of Human Genetics (Società Italiana di Genetica Medica, SIGU)
2022-ongoing	Member of the European Huntington’s Disease Network (EHDN)
2015	Winner of the best poster at the "I CONGRESSO ACCADEMIA LIMPE-DISMOV" (Turin 2015) with the scientific contribution: "Frequency and clinical characteristics of mutations in the GBA gene in Italian patients with Parkinson's disease".
2020	Achievement of the recognition "Article of the month: the best articles published by Italian authors in the major Italian and foreign scientific journals" conferred

	by the LIMPE-DIS MOV Academy for the month of July 2020 for the publication of the article: "GBA-Related Parkinson's Disease: Dissection of Genotype–Phenotype Correlates in a Large Italian Cohort" in the international scientific journal Movement Disorders (2020 Jul 13. doi: 10.1002/mds.28195). (authors: Simona Petrucci, M. Ginevrino, I. Trezzi, *et al*. E) (viewable at the link: https:// www.accademialimpedismov.it/page/articolo-mese)
2022-ongoing	Member of the Scientific Commettee of The Italian Huntington Disease Association (AICH)-Rome

Part VI – Editorial and reviewing activity

2022-ongoing	Guest Editor of the Special Issue on Huntington's Disease "New Frontiers in Therapeutics"
2015-ongoing	Invited reviewer of the following international journals: "Journal of Affective Disorders"; "European Journal of Neurology", "Parkinson's Disease", BioMed Research International", "European Journal of Obstetrics & Gynecology and Reproductive Biology" "All Life", "Nutrition", "International Journal of Cardiology", "Annals of Human Genetics", "Frontiers in Genetics", "Frontiers Aging Neuroscience", "Plos ONE".
2020 and 2022	External Evaluation for the PhD thesis "Molecular and functional characterization of known and novel genes involved in pathogenesis of Hereditary Spastic Paraplegias" (Doctoral Course of Molecular, Cellular and Environmental Biology, XXXIII Cycle, Department of Science, "Roma Tre" University", 2020) and for the PhD thesis "Genetic Epilepsies and Epileptic/Developmental Encephalopathies with Movement Disorders" (Doctoral Course of Clincal- Experimetal Neuroscience and Psychiatry, XXXIV Cycle, "Sapienza" University of Rome, 2022).

Part VII - Funding Information [grants as PI-principal investigator or I-investigator]

Year	Title	Program	Grant value
2019-2023	Biobanks of primary cultures enriched with Lung Cancer Stem Cells (CSC) and adapted immune cells: targeting lipid metabolism and correlations patients risk factors	Sapienza University Grant (I)	29.000 €
2020-ongoing	Clinical and molecular study of GLUT1 related phenotypes: functional studies and new genotype-phenotype correlations	Sapienza University Grant (I)	10.000.00 €
2022-ongoing	NOTCH3 pathogenic variants in multiple sclerosis patients: frequency, clinical features and disease-associated cellular phenotypes	Sapienza University Grant (PI)	10.000.00 €

Part VIII- Conference Presentations

Date Title

19th, November 2010	“Parkinsonism plus of an undetermined nature in three affected brothers” (oral communication, “Young in Movement Disorders, II edition” congress, Naples).
20th, September 2012	Genetic basis of Parkinson's disease: clinical aspects and genetic counseling" (oral communication, “Update on Parkinson's disease and monogenic parkinsonisms” congress, Verona).
20th-21st, November 2012	"Paroxysmal dyskinesias. Genetics and pathogenesis" (oral communication, "Training course on the riots of the Mariani Foundation movement", Genoa).
8th-9th, March 2013	"Parkinsonism as a phenotype of genetic diseases" (oral communication, "DISMOV School. The school of movement disorders" congress, Ravello).
29th-30th, May 2013	"The genetics of Parkinson's disease" (oral communication, "Residential Course in Neurogenetics" congress, Pisa).
9th-11th, October 2013	"The genetics of dystonias" (oral communication, “Joint Congress LIMPE DISMOV/SIN” congress, Rome).
28th-30 th, September 2015	“Frequency and clinical features of GBA mutations in Italian patients with Parkinson disease” (poster, “I congress of the LIMPE/DISMOV Academia”, Turin)
4th-6th, May 2016	"Early onset parkinsonisms. Lesson of genetic forms" (oral comuncation) and “Genotype-phenotype paradoxes in an Italian family with a multiexonic duplication of the Parkin gene.” (poster), “National Congress LIMPE DISMOV Academy” congress, Bari.
24th, February 2017	"The role of genetics in the approach to Parkinson's disease" (oral communication, "New horizons in Parkinson's disease" congress, Sant'Eugenio Hospital, Rome).
25th-27th, October 2018	“De novo dominant TFG mutation causing complex spastic paraparesis” and ““Simultaneous analysis of Copy Number Variation (CNV) and point mutations in BRCA1/2 genes using a single workflow on the Ion PGM (Personal Genome Machine) platform.” (posters, “XXI National Congress SIGU” congress, Catania)
24th-30th, October 2018	"Neurogenetics" Poster session (moderator, “XLIX SIN National Congress”, Rome).
19th, April 2019	"Frequency, genotype and phenotypic features of variants in sarcomeric genes in Italian patients with hypertrophic cardiomyopathy" (oral communication, "Genetic tests in the diagnosis of hypertrophic cardiomyopathy" Scuola Medica Ospedaliera (SMO) course, Rome).
22th, May 2019	"Genetics in Dystonia" (oral communication, "Distonie" course in the V Limpe DISMOV National Congress, Catania).
19th-21th, September 2019	“De novo dominant TFG variant causes complex hereditary spastic parapaplegia type 57” (poster, “6th International Meeting on Spastic Paraparesis and Ataxia” congress, Nice)
13th-16 th, November 2019	"Results of the first year of the Italian Working Group activity on the Genetics of Cardio-arrhythmias" and “Molecular characterization of hereditary breast cancer: from the analysis of *BRCA1* and *BRCA2* genes to multi-gene panels". (posters, “XXII SIGU National Congress”, Rome)

11th-13th, November 2020	"Very rare large duplication in *CHEK2* gene: an Italian founder effect mutation?"; "A very rare likely pathogenic variant in *ACVRL1* gene identified in a patient with atypical hereditary hemorrhagic telangiectasia" and o "Novel likely pathogenic variants in patients with complex *CACNA1A* related phenotypes." (posters, "XXIII SIGU National Congress", virtual edition).
19 th -20th, December 2020	"Evidence of genetic origin of QTc abnormalities in hypertrophic cardiomyopathy: a case report with co-inheritance of TNNI3 and KCNQ1 variants". (poster, "81st National Congress of the Italian Society of Cardiology", virtual edition).
17th-19th, November 2021	"A new SMAD4 splice site variant in a three generation Italian family with juvenile polyposis syndrome" and "A new GFAP missense variant causing a hereditary juvenile form of Alexander Disease in an Italian family." (posters, "XXIV SIGU National Congress", virtual edition).
27th, November 2021	"Emerging therapies in Huntington's disease: news from clinical trials" (oral communication, "Where were we?" congress, Associazione Italiana Corea di Huntington-AICH, Rome).
28th, May 2022	"Trials "WITHOUT" and "WITH" drugs at S.Andrea. The underestimated importance of the "WITHOUT" and the confirmation of the "WITH"" (oral communication, "Starting again from three: from our identity; by our group of doctors; from the way we treat patients and support families" congress, Associazione Italiana Corea di Huntington-AICH, Rome).
5th,July 2022	"Medical genetics of familial melanoma" (oral communication, "Dermo-Connect" congress, Rome).
7th-9th, September 2022	"Genotype-phenotype correlation in early and late-onset Hypertrophic Cardiomyopathy Italian patients" and "Long QT in HCM: a consequence of myocardial hypertrophy or a distinct genetic disease?" (posters, "XXV SIGU National Congress", Trieste)
3rd, December 2022	"Modifier genes in Huntington Disease: protagonists, role, clinical and therapeutic implications" (oral communication, "Between past and future: the new challenges of genetic counseling for people at risk" congress, Associazione Italiana Corea di Huntington-AICH, Rome).
12th, May 2023	"Familial melanoma and genetics" (oral communication, "Metropolitan Dermatology" congress, Viterbo).
13rd, May 2023	"Updates from UniQure: the first results of the Phase I/II investigational study with AMT-130" (oral communication, "Gene therapy in Huntington's disease: developments and perspectives" congress, Associazione Italiana Corea di Huntington-AICH, Rome).

Part XIX – Research Activities

Keywords	Brief Description
Parkinson Disease	2011-2016: Participation in research activities of the Neurogenetics Unit coordinated by Prof. E.M. Valente (IRCCS CSS-Mendel, Rome; IRRCS Fondazione Santa Lucia, Rome) in the context of projects financed by the

	Ministry of Health "Ricerca Corrente" with the following aims: i) clinical and molecular analysis of movement on a genetic basis (parkinsonisms and hereditary dystonic syndromes); ii) phenotypic and pathogenetic characterization of monogenic parkinsonisms with autosomal dominant and recessive transmission.
	2010-ongoing: participation in international multicenter studies: A) "European project on the Mendelian forms of Parkinson's disease. MEFOPA" (2010-2014): the aim of the study was the evaluation of correlations between Parkinson's disease, pathogenic variants responsible for the pathology and biomarkers/metabolites present in the plasma/or fibroblasts of affected and asymptomatic carriers of pathogenic variants, and in healthy controls; B) "European Project on the Global Assessment of genetic risk factors and environmental risk factors associated with Parkinson's disease ("COmprehensive Unbiased Risk factors Assessment for Genetics and Environment in Pakinson's Disease, COURAGE-PD) (2014-2017): the aim of the study was the evaluation of the genotypic characterization of large cohorts of Parkinson's disease patients belonging to different ethnicities, environmental exposures and the interactions between them, by an integrated approach, using new statistical methods and functional studies on innovative cellular models; C) International research project "MJFF Global Genetic PD project" coordinated by Prof. Christine Klein (2017-ongoing): the aim of the study is the genotypic and phenotypic characterization of a worldwide cohort of patients and families with monogenic forms of Parkinson's disease using a new and comprehensive approach based on the global collaboration of all the world's experts in movement disorders; D) Research projects promoted by the Genetic Epidemiology of Parkinson's Disease (GEoPD) Group (2012-ongoing), an international consortium among 60 sites on 6 continents, dedicated to the promotion, development and dissemination of scientific and translational research results in Parkinson's disease (projects and centers available on the website https://www.geopd.net/members); E) Global Parkinson's Genetics Program 2 (GP2) (2020-ongoing), a research program of the Aligning Science Across Parkinson's (ASAP) initiative focused on improving our understanding of the genetic architecture of Parkinson's disease (PD) and making this knowledge globally relevant.
GLUT1	2019-2022: Participation at the project "Clinical and molecular study of GLUT1-associated phenotypes: functional studies and genotype-phenotype correlations", aimed at identifying and functionally validate variants of the *SLC2A1* gene in patients with clinical characteristics attributable to the spectrum of phenotypes associated with GLUT1 deficiency, coordinated by Prof. Caputo V. (Department of Experimental Medicine, Sapienza University of Rome; CSS-Mendel Institute).
Hereditary cancers	2018-ongoing: Clinical geneticist involved in the genotypic and phenotypic characterization of hereditary cancers (Herediary Breast and Ovarian Cancer, Lynch Syndrome, and other syndromes with hereditary gastrointestinal tumours, Multiple Endocrine Neoplasia (research activity carried out at the Department of Clinical and Molecular Medicine, Sapienza University of Rome).

BI-ALCL	2020-2022: Participation in the project "Identification of germinal and somatic molecular alterations that predispose and participate in the pathogenesis of anaplastic large cell lymphoma associated with breast implants (BI-ALCL)", a national multicenter study that aims to identify genetic and non-genetic risk factors predisposing to BI-ALCL in patient with breast implants.
Hereditary Cardiomyopathy	2018-ongoing: Clinical geneticist involved in the genotypic and phenotypic evaluation of monogenic forms of cardiomyopathy and arrythmias (research activity carried out at the Department of Clinical and Molecular Medicine, Sapienza University of Rome).
	2019-ongoing: Participation in the Italian Working Group on the Genetics of Cardio-Arrhythmias, created from the need of the 23 participating centers to standardize and harmonize the laboratory activity and the diagnostic pathways that lead to the molecular diagnosis of hereditary cardiomyopathy and arrythmias, nationwide, considering ethical aspects and implications of references in clinical practice.
CADASIL	2019-ongoing: Clinical geneticist involved in the genotypic and phenotypic evaluation of patients with hereditary vascular multifocal leukoencephalopathy (research activity carried out at the Department of Clinical and Molecular Medicine, Sapienza University of Rome): PI of the project “NOTCH3 pathogenic variants in multiple sclerosis patients: frequency, clinical features and disease-associated cellular phenotypes”, a project that aims at detecting the *NOTCH3* variants frequency in patients with MS and the clinical, MRI and CSF features able to discern MS-CADASIL co-occurrence from “inflammatory CADASIL.

Part X – Summary of Scientific Achievements

Product type	Number	Data Base	Start	End
Papers [international]	56	Scopus	2008	2023
Books [scientific]	1	Scopus	2015	2015

Total Impact factor (IF)	282 (WOS)
Average IF per Publication	4,944
Average of IF Product	4,77
Total Citations	1085 (Scopus)
Average Citations per Product	19 (Scopus)
Hirsch (H) index	20 (Scopus)
Normalized H index*	1,33 (Scopus)
Contemporary H index	16 (Scopus)

*****H index divided by the academic seniority.

Part XI– All publications

1. “Prenatal CFAP53-related laterality defect: case report and review of the literature “. Mastromoro G, Guadagnolo D, Novelli A, Torres B, Piane M, Magliozzi M, Bernardini L, Ventriglia F, Pizzuti A, **Petrucci S**.. J Matern Fetal Neonatal Med. 2023 Dec;36(1):2201653. doi: 10.1080/14767058.2023.2201653. PMID: 37041101. IF: n.d. (WOS); Cit.: 0 (Scopus).

2. “Investigation of Shared Genetic Risk Factors Between Parkinson's Disease and Cancers”. Sugier PE, Lucotte EA, Domenighetti C, Law MH, Iles MM, Brown K, Amos C, McKay JD, Hung RJ, Karimi M, Bacq-Daian D, Boland-Augé A, Olaso R, Deleuze JF, Lesueur F, Ostroumova E, Kesminiene A, de Vathaire F, Guénel P; EPITHYR consortium; Sreelatha AAK, Schulte C, Grover S, May P, Bobbili DR, Radivojkov-Blagojevic M, Lichtner P, Singleton AB, Hernandez DG, Edsall C, Mellick GD, Zimprich A, Pirker W, Rogaeva E, Lang AE, Koks S, Taba P, Lesage S, Brice A, Corvol JC, Chartier-Harlin MC, Mutez E, Brockmann K, Deutschländer AB, Hadjigeorgiou GM, Dardiotis E, Stefanis L, Simitsi AM, Valente EM, **Petrucci S**, Straniero L, Zecchinelli A, Pezzoli G, Brighina L, Ferrarese C, Annesi G, Quattrone A, Gagliardi M, Matsuo H, Nakayama A, Hattori N, Nishioka K, Chung SJ, Kim YJ, Kolber P, van de Warrenburg BPC, Bloem BR, Aasly J, Toft M, Pihlstrøm L, Guedes LC, Ferreira JJ, Bardien S, Carr J, Tolosa E, Ezquerra M, Pastor P, Diez-Fairen M, Wirdefeldt K, Pedersen N, Ran C, Belin AC, Puschmann A, Rödström EY, Clarke CE, Morrison KE, Tan M, Krainc D, Burbulla LF, Farrer MJ, Kruger R, Gasser T, Sharma M; Comprehensive Unbiased Risk Factor Assessment for Genetics and Environment in Parkinson's Disease (Courage-PD) consortium, Truong T, Elbaz A. Mov Disord. 2023 Apr;38(4):604-615. doi: 10.1002/mds.29337. Epub 2023 Feb 14. PMID: 36788297. IF: 9,689 (WOS), Cit.:2 (Scopus)

3. “Long QTc in hypertrophic cardiomyopathy: A consequence of structural myocardial damage or a distinct genetic disease?”. Cava F, Micolonghi C, Musumeci MB, **Petrucci S**, Savio C, Fabiani M, Tini G, Germani A, Libi F, Rossi C, Visco V, Pizzuti A, Volpe M, Autore C, Rubattu S, Piane M. Front Cardiovasc Med. 2023 Apr 5;10:1112759. doi: 10.3389/fcvm.2023.1112759. PMID: 37089884. IF: 5,848 (WOS), Cit.: 0 (Scopus).

4. “A Novel Nonsense Pathogenic TTN Variant Identified in a Patient with Severe Dilated Cardiomyopathy”. Micolonghi C, Fabiani M, Pagannone E, Savio C, Ricci M, Caroselli S, Gambioli V, Musumeci B, Germani A, Tini G, Autore C, Pizzuti A, Visco V, Rubattu S, **Petrucci S**, Piane M. Curr Issues Mol Biol. 2023 Mar 15;45(3):2422-2430. doi: 10.3390/cimb45030157. PMID: 3697552. IF: 2,976 (WOS), Cit.: 0 (Scopus).

5. “Embracing Monogenic Parkinson's Disease: The MJFF Global Genetic PD Cohort”. Vollstedt EJ, Schaake S, Lohmann K, Padmanabhan S, Brice A, Lesage S, Tesson C, Vidailhet M, Wurster I, Hentati F, Mirelman A, Giladi N, Marder K, Waters C, Fahn S, Kasten M, Brüggemann N, Borsche M, Foroud T, Tolosa E, Garrido A, Annesi G, Gagliardi M, Bozi M, Stefanis L, Ferreira JJ, Correia Guedes L, Avenali M, **Petrucci S**, Clark L, Fedotova EY, Abramycheva NY, Alvarez V, Menéndez-González M, Jesús Maestre S, Gómez-Garre P, Mir P, Belin AC, Ran C, Lin CH, Kuo MC, Crosiers D, Wszolek ZK, Ross OA, Jankovic J, Nishioka K, Funayama M, Clarimon J, Williams-Gray CH, Camacho M, Cornejo-Olivas M, Torres-Ramirez L, Wu YR, Lee-Chen GJ, Morgadinho A, Pulkes T, Termsarasab P, Berg D, Kuhlenbäumer G, Kühn AA, Borngräber F, de Michele G, De Rosa A, Zimprich A, Puschmann A, Mellick GD, Dorszewska J, Carr J, Ferese R, Gambardella S, Chase B, Markopoulou K, Satake W, Toda T, Rossi M, Merello M, Lynch T, Olszewska DA, Lim SY, Ahmad-Annuar A, Tan AH, Al-Mubarak B, Hanagasi H, Koziorowski D, Ertan S, Genç G, de Carvalho Aguiar P, Barkhuizen M, Pimentel MMG, Saunders-Pullman R, van de Warrenburg B, Bressman S, Toft M, Appel-Cresswell S, Lang AE, Skorvanek M, Boon AJW, Krüger R, Sammler

EM, …, Klein C. Mov Disord. 2023 Feb;38(2):286-303. doi: 10.1002/mds.29288. PMID: 36692014. IF: 9,689 (WOS), Cit.: 2 (Scopus).

6. “Diagnostic and therapeutic recommendations in adult dystonia: a joint document by the Italian Society of Neurology, the Italian Academy for the Study of Parkinson's Disease and Movement Disorders, and the Italian Network on Botulinum Toxin”. Romano M, Bagnato S, Altavista MC, Avanzino L, Belvisi D, Bologna M, Bono F, Carecchio M, Castagna A, Ceravolo R, Conte A, Cosentino G, Eleopra R, Ercoli T, Esposito M, Fabbrini G, Ferrazzano G, Lalli S, Mascia MM, Osio M, Pellicciari R, **Petrucci S,** Valente EM, Valentino F, Zappia M, Zibetti M, Girlanda P, Tinazzi M, Defazio G, Berardelli ANeurol Sci. 2022 Dec;43(12):6929-6945. doi: 10.1007/s10072-022-06424-x. Epub 2022 Oct 3. PMID: 36190683. IF: 3,83 (WOS), Cit.: 6 (Scopus).

7. “Complete Pseudo-Anodontia in an Adult Woman with Pseudo-Hypoparathyroidism Type 1a: A New Additional Nonclassical Feature?”. Sciacchitano S, De Francesco GP, Piane M, Savio C, De Vitis C, **Petrucci S**, Salvati V, Goldoni M, Fabiani M, Mesoraca A, Micolonghi C, Torres B, Piccinetti A, Pippi R, Mancini R. Diagnostics (Basel). 2022 Nov 30;12(12):2997. doi: 10.3390/diagnostics12122997. PMID: 36553004. IF: 3,992 (WOS), Cit.: 0 (Scopus).

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Part XII– Selected Publications for evaluation

1. A New SMAD4 Splice Site Variant in a Three-Generation Italian Family with Juvenile Polyposis Syndrome". Micolonghi C, Piane M, Germani A, Sadeghi S, Libi F, Savio C, Fabiani M, Mancini R, Ranieri D, Pizzuti A, Corleto VD, Parisi P, Visco V, Di Nardo G, **Petrucci S**. Diagnostics (Basel). 2022 Nov 4;12(11):2684. PMID: 36359527. PMCID: PMC9689379. DOI: 10.3390/diagnostics12112684. IF: 3,992 (WOS), Cit.: 0 (Scopus).
2. "Genotype-Phenotype Correlations in Monogenic Parkinson Disease: A Review on Clinical and Molecular Findings". Guadagnolo D, Piane M, Torrisi MR, Pizzuti A, **Petrucci S**. Front Neurol. 2021 Sep 22; eCollection 2021. PMID: 34630269. PMCID: PMC8494251. DOI: 10.3389/fneur.2021.648588. IF: 4,086 (WOS), Cit.: 17 (Scopus).
3. "Beyond BRCA1 and BRCA2: Deleterious Variants in DNA Repair Pathway Genes in Italian Families with Breast/Ovarian and Pancreatic Cancers". Germani A, **Petrucci S**, De Marchis L, Libi F, Savio C, Amanti C, Bonifacino A, Campanella B, Capalbo C, Lombardi A, Maggi S, Mattei M, Osti MF, Pellegrini P, Speranza A, Stanzani G, Vitale V, Pizzuti A, Torrisi MR, Piane M. J Clin Med. 2020 Sep 17;9(9):3003. PMID: 32957588. PMCID: PMC7563793. DOI: 10.3390/jcm9093003. IF: 4,242 (WOS), Cit.: 5 (Scopus).
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12. "Phenotypic variability of PINK1 expression: 12 Years' clinical follow-up of two Italian families ". Ricciardi L, **Petrucci S**, Guidubaldi A, Ialongo T, Serra L, Ferraris A, Spanò B, Bozzali M, Valente EM, Bentivoglio AR. Mov Disord. 2014 Oct;29(12):1561-6. Epub 2014 Aug 27. PMID: 25164310. doi: 10.1002/mds.25994. IF: 5,68 (WOS). Cit.: 42 (Scopus).