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Curriculum Vitae Europass

Informazioni personali

Nome(i) / Cognome(i)	**Carla Marini**
Indirizzo(i)	Via Podgora 57, 60124, Ancona
Telefono(i)	3468729086
E-mail	carla.marini@ospedaliriuniti.marche.it
Cittadinanza	Italiana
Data di nascita	02/06/1966
Sesso	F
Occupazione desiderata/	Direttore SOD Neuropsichiatria Infantile e Centro Regionale per la Diagnosi e Cura Epilessia Infantile
Settore professionale	Presidio Ospedaliero Materno Infantile G. Salesi; Ospedali Riuniti Ancona Ancona

Esperienza professionale

1992: laurea in Medicina e Chirurgia presso l'Università di Bologna; tesi di laurea con titolo: "Sindrome amnesica epilettica: osservazioni personali e contributi sperimentali', relatore professore Elio Lugaresi. Acquisizione di una discreta esperienza nella valutazione dei pazienti con epilessia del lobo temporale e nella valutazione del deficit neuropsicologico prodotto dalle crisi epilettiche.

1992-1994: attività di medicina generale come guardia medica nelle ASL di Cagli e Pergola (Pesaro-Urbino) con incarichi temporanei e rinnovabili per un totale di circa 500 ore lavorative, sostituzioni di medicina acquisizione di una buona esperienza della gestione dei problemi di medicina generale dei pazienti.

1994 –1998: scuola di specializzazione in Neurologia, clinica neurologica dell'Università di Bologna, partecipazione alle attività didattiche pratiche e teoriche previste dal corso di specializzazione. Le attività pratiche sono state svolte presso i reparti di Neurologia della Clinica Neurologica diretti dal Professore Pasquale Montagna e dal Professore Paolo Martinelli, presso il servizio di Neurologia dell'Ospedale Sant'Orsola diretto dal Professore Paolo Pazzaglia e presso il reparto di Neurologia dell'Ospedale Bellaria diretto dal Professore Carlo Alberto Tassinari. Frequentazione centro "G. M. Corsini" per lo studio ed il trattamento dell'epilessia ed il laboratorio di neurofisiologia diretto dal Professore Agostino Baruzzi e coordinato dal Dr Paolo Tinuper. Acquisizione di buona esperienza nella valutazione, diagnosi e trattamento dei pazienti con malattie neurologiche ma soprattutto dei pazienti con epilessia. Partecipazione agli ambulatori, all'esecuzione delle registrazioni video-EEG, polisonnografiche e per il monitoraggio delle funzioni del sistema nervoso autonomo ed alla acquisizione delle conoscenze necessarie per la refertazione di tali esami. Coinvolgimento nei programmi di ricerca che riguardavano: le registrazioni poligrafiche dei pazienti con crisi di caduta, l'epilessia frontale notturna e lo studio delle funzioni del sistema nervoso autonomo ed endocrinologico dei pazienti con crisi parziali

1998: stage di formazione specialistica all'estero, con una fellowship clinica presso il dipartimento di Neurologia dell'Austin & Repatriation Medical Center di Melbourne, Australia sotto la supervisione del Professor Samuel F Berkovic. Acquisizione delle basi per studi di genetica clinica che sono poi stati approfonditi negli anni successivi di PhD

1998: specializzazione in neurologia discutendo una tesi con titolo: 'La genetica delle epilessie con crisi ad assenza'. La tesi è stata realizzata con la supervisione del Professore Samuel F Berkovic, presso Epilepsy Research Institute, Austin & Repatriation Medical Centre, Melbourne, Australia.

1999: PhD presso l'Università di Melbourne, Australia, sotto la supervisione del professore Samuel F Berkovic. Interessi di ricerca focalizzati in particolare sulla genetica delle epilessie con studi clinici e neurofisiologici per la definizione del fenotipo e studi genealogici per la identificazione di famiglie con epilessia ereditaria per la valutazione del modello di ereditarietà in tali famiglie e della penetranza. Il corso di PhD comprendeva anche il lavoro clinico con la partecipazione al 'Comprehensive Epilepsy Program' diretto dal professore Samuel F Berkovic al Austin and Repatriation Medical Centre per la caratterizzazione e valutazione prechirurgica dei pazienti candidati alla chirurgia della epilessia, e agli ambulatori del 1st Seizure Clinic coordinato dal Dr Mark R Newton per pazienti con prima crisi, provenienti dai pronto soccorso degli Ospedali di Melbourne.

Luglio 2001: corso di statistica per ricercatori presso l'Università di Melbourne, Australia
15.10 2003: completato il corso di PhD con la sottomissione di una tesi intitolata: FAMILY STUDIES OF EPILEPSY WITH SIMPLE AND COMPLEX INHERITANCE, sotto la supervisione del Professore Samuel F Berkovic, Università di Melbourne, Melbourne, Australia.

	03.06.2003- 23/03/07: dirigente medico di I livello a tempo determinato presso l' Unità Operativa di Epilettologia, Neurofisiologia e Neurogenetica dell'IRCCS Fondazione Stella Maris (Calambrone, Pisa, Italia) diretta dal Professore Renzo Guerrini con un contratto annuale rinnovabile e con fondi provenienti dal programma del Ministero della Istruzione, dell'Università e della Ricerca per il 'Rientro dei Cervelli'. Marzo 2007: vincitore di concorso per dirigente medico neurologo presso IRCCS Stella Maris, Calambrone, Pisa Dal 02/06/07 al 01/11/2017: trasferimento presso neurologia pediatrica Ospedale Meyer, Direttore Prof. Renzo Guerrini. Attività clinica e di ricerca nell'ambito della neurologia pediatrica con particolare interesse all'epilessia e soprattutto alle forme con eziologia genetica. Qualifica: Dirigente Medico I livello, Neurologia Pediatrica, Ospedale Pediatrico A. Meyer , Firenze Dal 01/11/2017 al 31/12/2019 professore associato con incarichi assistenziali, dipartimento di Neurofarba; Università di Firenze e AUO Meyer; Firenze
Lavoro o posizione ricoperti	Direttore SOD Neuropsichiatria Infantile e Centro Regionale per la Diagnosi e Cura Epilessia Infantile; Presidio Ospedaliero Materno Infantile G. Salesi; Ospedali Riuniti Ancona; Ancona
Principali attività e responsabilità	Dirigente medico specializzato nelle problematiche neurologiche dei bambini e che si svolge con attività di ambulatori, reparto, refertazione EEG ed emergenze neurologiche; direttore SOD di neuropsichiatria infantile e centro regionale per epilessia comprendente 19 posti letto compreso DH, per patologie neurologiche e psichiatriche del bambino; servizio diurno per valutazione bambini sotto i 6 anni con problemi di sviluppo psicomotorio, registrazioni video-EEG poligrafiche, centro regionale per ADHD.
Nome e indirizzo del datore di lavoro	Ospedali Riuniti Ancona, via Conca; Ancona
Tipo di attività o settore	Neurologia e Psichiatria Pediatrica
Istruzione e formazione	1985 Diploma di Maturità, Liceo Scientifico, Pergola, Pesaro, 1992 Laurea in Medicina e Chirurgia presso l'Università di Bologna, 1992 Tirocinio di abilitazione per l'esame di stato presso l'Ospedale Sant'Orsola, Bologna 1992 Esame di Stato per l'abilitazione all'esercizio della professione medico chirurgo 1998 Scuola di Specializzazione in Neurologia, Clinica Neurologica dell'Università di Bologna 1998 PhD presso l'Università di Melbourne, Melbourne, Australia 2003 Conferimento del titolo di Doctor of Philosophy
Titolo della qualifica rilasciata	MD, PhD
Principali tematiche/competenze professionali possedute	Attività clinica e di ricerca nelle malattie neurologiche con particolare interesse e competenze per le epilessie su base genetica

Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione	Università di Bologna, Clinica Neurologica dell'Università di Bologna, Università di Melbourne, Australia
Madrelingua(e)	**Italiano**

Altra(e) lingua(e)

Autovalutazione

Livello europeo (*)

	Comprensione				Parlato		Scritto
	Ascolto		Lettura		Interazione Orale	Produzione orale	
Lingua	**Inglese**	100%	Ottima	Ottima	Ottima	Ottima	Ottima
Lingua	**Francese**	70%	discreta	discreta	Sufficiente	Sufficiente	Sufficiente

(*) Quadro comune europeo di riferimento per le lingue

Capacità e competenze sociali	Buone capacità di collaborazione, di adattamento alle esigenze dell'ambiente o di altre persone, buona dialettica.
Capacità e competenze organizzative	Buone capacità organizzative
Capacità e competenze informatiche	Buone capacità informatiche per programmi di scrittura (word), database (file maker pro), excell, programmi per costruzione di alberi genealogici (apple work 6), programmi di grafica (adobe photoshop).
Patente	Patente di guida per automobile
Ulteriori informazioni	
Allegati	A seguire elenco delle pubblicazioni su riviste scientifiche internazionali

Autorizzo il trattamento dei miei dati personali ai sensi del Decreto Legislativo 30 giugno 2003, n. 196 "Codice in materia di protezione dei dati personali (facoltativo)".

Firma

Ancona 14/12/2020

DICHIARAZIONE SOSTITUTIVA DI CERTIFICAZIONE
(Art. 46 D.P.R. 28 dicembre 2000, n. 445 s.m.i.)

esente da bollo ai sensi dell'art. 37 D.P.R. 445/2000

La Sottoscritta Marini Carla

Nata a Frontone (PU) il 02/06/1966

residente ad Ancona; via Podgora 57; 60124

Dichiara di avere partecipato in qualità di co-autore alle seguenti pubblicazioni:

1. Matricardi S, De Liso P, Freri E, Costa P, Castellotti B, Magri S, Gellera C, Granata T, Musante L, Lesca G, Oertel J, Craiu D, Hammer TB, Møller RS, Barisic N, Abou Jamra R, Polster T, Vigevano F, Marini C. Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia. 2020;61(11):2474-2485.

2. Lenge M, Marini C, Canale E, Napolitano A, De Masi S, Trivisano M, Mei D, Longo D, Rossi Espagnet MC, Lucenteforte E; PCDH19 Clinical Study Group, Barba C, Specchio N, Guerrini R. Quantitative MRI-Based Analysis Identifies Developmental Limbic Abnormalities in PCDH19 Encephalopathy. Cereb Cortex. 2020;30(11):6039-6050.

3. Lal D, May P, Perez-Palma E, Samocha KE, Kosmicki JA, Robinson EB, Møller RS, Krause R, Nürnberg P, Weckhuysen S, De Jonghe P, Guerrini R, Niestroj LM, Du J, Marini C; EuroEPINOMICS-RES Consortium, Ware JS, Kurki M, Gormley P, Tang S, Wu S, Biskup S, Poduri A, Neubauer BA, Koeleman BPC, Helbig KL, Weber YG, Helbig I, Majithia AR, Palotie A, Daly MJ. Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Med. 2020;12(1):28

4. Kolc KL, Sadleir LG, Depienne C, Marini C, Scheffer IE, Møller RS, Trivisano M, Specchio N, Pham D, Kumar R, Roberts R, Gecz J. A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. Transl Psychiatry. 2020;10(1):127.

5. Tang S, Addis L, Smith A, Topp SD, Pendziwiat M, Mei D, Parker A, Agrawal S, Hughes E, Lascelles K, Williams RE, Fallon P, Robinson R, Cross HJ, Hedderly T, Eltze C, Kerr T, Desurkar A, Hussain N, Kinali M, Bagnasco I, Vassallo G, Whitehouse W, Goyal S, Absoud M; EuroEPINOMICS-RES Consortium, Møller RS, Helbig I, Weber YG, Marini C, Guerrini R, Simpson MA, Pal DK. Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia. 2020;61(5):995-1007

6. Chatron N, Becker F, Morsy H, Schmidts M, Hardies K, Tuysuz B, Roselli S, Najafi M, Alkaya DU, Ashrafzadeh F, Nabil A, Omar T, Maroofian R, Karimiani EG, Hussien H, Kok F, Ramos L, Gunes N, Bilguvar K, Labalme A, Alix E, Sanlaville D, de Bellescize J, Poulat AL; EuroEpinomics-RES consortium AR working group, Moslemi AR, Lerche H, May P, Lesca G, Weckhuysen S, Tajsharghi H. Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain. 2020 May 1;143(5):1447-1461.

7. Niestroj LM, Perez-Palma E, Howrigan DP, Zhou Y, Cheng F, Saarentaus E, Nürnberg P, Stevelink R, Daly MJ, Palotie A, Lal D; Epi25 Collaborative. Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. Brain. 2020 Jul 1;143(7):2106-2118

8. Kapoor D, Anand A, Sharma S, Mukherjee SB, Marini C, Mei D, Chopra SS, Chettali AM.

9. Dravet Syndrome: A Case Series. Indian J Pediatr. 2020 Jun 26

10. Mei D, Cetica V, Marini C, Guerrini R. Dravet syndrome as part of the clinical and genetic spectrum of sodium channel epilepsies and encephalopathies. Epilepsia. 2019;60 S3:S2-S7.

11. Licchetta L, Pippucci T, Baldassari S, Minardi R, Provini F, Mostacci B, Plazzi G, Tinuper P, Bisulli F; Collaborative Group of Italian League Against Epilepsy (LICE) Genetic Study Group on SHE. Sleep-related hypermotor epilepsy (SHE): Contribution of known genes in 103 patients. Seizure. 2019 Nov 23;74:60-64.

12. Esposito A, Falace A, Wagner M, Gal M, Mei D, Conti V, Pisano T, Aprile D, Cerullo MS, De Fusco A, Giovedì S, Seibt A, Magen D, Polster T, Eran A, Stenton SL, Fiorillo C, Ravid S, Mayatepek E, Hafner H, Wortmann S, Levanon EY, Marini C, Mandel H, Benfenati F, Distelmaier F, Fassio A, Guerrini R. Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain. 2019 Dec 1;142(12):3876-3891.

13. Burgess R, Wang S, McTague A, Boysen KE, Yang X, Zeng Q, Myers KA, Rochtus A, Trivisano M, Gill D; EIMFS Consortium, Sadleir LG, Specchio N, Guerrini R, Marini C, Zhang YH, Mefford HC, Kurian MA, Poduri AH, Scheffer IE. The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Ann Neurol. 2019 Dec;86(6):821-831.

14. Bar C, Barcia G, Jennesson M, Le Guyader G, Schneider A, Mignot C, Lesca G, Breuillard D, Montomoli M, Keren B, Doummar D, Billette de Villemeur T, Afenjar A, Marey I, Gerard M, Isnard H, Poisson A, Dupont S, Berquin P, Meyer P, Genevieve D, De Saint Martin A, El Chehadeh S, Chelly J, Guët A, Scalais E, Dorison N, Myers CT, Mefford HC, Howell KB, Marini C, Freeman JL, Nica A, Terrone G, Sekhara T, Lebre AS, Odent S, Sadleir LG, Munnich A, Guerrini R, Scheffer IE, Kabashi E, Nabbout R. Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Hum Mutat. 2020 Jan;41(1):69-80.

15. Muir AM, Myers CT, Nguyen NT, Saykally J, Craiu D, De Jonghe P, Helbig I, Hoffman-Zacharska D, Guerrini R, Lehesjoki AE, Marini C, Møller RS, Serratosa J, Štěrbová K, Striano P, von Spiczak S, Weckhuysen S, Mefford HC; EuroEPINOMICS-RES NLES working group, Sarah Weckhuysen. Genetic heterogeneity in infantile spasms. Epilepsy Res. 2019 Oct;156:106181.

16. Guerrini R, Marini C, Barba C. Generalized epilepsies. Handb Clin Neurol.2019;161:3-15.

17. Guerrini R, Parrini E, Marini C, Mei D. What is the role of next generation sequencing in status epilepticus? Epilepsy Behav. 2019 Dec;101(Pt B):106373. Helbig I, Lopez-Hernandez T, Shor O, Galer P, Ganesan S, Pendziwiat M, Rademacher A, Ellis CA, Hümpfer N, Schwarz N, Seiffert S, Peeden J, Shen J, Štěrbová K, Hammer TB, Møller RS, Shinde DN, Tang S, Smith L, Poduri A, Krause R, Benninger F, Helbig KL, Haucke V, Weber YG; EuroEPINOMICS-RES Consortium; GRIN Consortium. A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. Am J Hum Genet. 2019 Jun 6;104(6):1060-1072.

18. Bisulli F, Licchetta L, Baldassari S, Muccioli L, Marconi C, Cantalupo G, Myers C, Menghi V, Minardi R, Caporali L, Marini C, Guerrini R, Mefford HC, Tinuper P, Pippucci T. SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disord. 2019 Apr 1;21(2):185-191.

19. Pippucci T, Licchetta L, Baldassari S, Marconi C, De Luise M, Myers C, Nardi E, Provini F, Cameli C, Minardi R, Bacchelli E, Giordano L, Crichiutti G, d'Orsi G, Seri M, Gasparre G, Mefford HC, Tinuper P, Bisulli F; Collaborative Group of Italian League Against Epilepsy (LICE) Genetic Commission.

Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. Ann Clin Transl Neurol. 2019 Feb 25;6(3):475-485.

20. Coppola A, Cellini E, Stamberger H, Saarentaus E, Cetica V, Lal D, Djémié T, Bartnik-Glaska M, Ceulemans B, Helen Cross J, Deconinck T, Masi S, Dorn T, Guerrini R, Hoffman-Zacharska D, Kooy F, Lagae L, Lench N, Lemke JR, Lucenteforte E, Madia F, Mefford HC, Morrogh D, Nuernberg P, Palotie A, Schoonjans AS, Striano P, Szczepanik E, Tostevin A, Vermeesch JR, Van Esch H, Van Paesschen W, Waters JJ, Weckhuysen S, Zara F, De Jonghe P, Sisodiya SM, Marini C; EuroEPINOMICS-RES Consortium; EpiCNV Consortium. Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia. 2019 Apr;60(4):689-706.

21. Siekierska A, Stamberger H, Deconinck T, Oprescu SN, Partoens M, Zhang Y,Sourbron J, Adriaenssens E, Mullen P, Wiencek P, Hardies K, Lee JS, Giong HK,Distelmaier F, Elpeleg O, Helbig KL, Hersh J, Isikay S, Jordan E, Karaca E,Kecskes A, Lupski JR, Kovacs-Nagy R, May P, Narayanan V, Pendziwiat M, Ramsey K, Rangasamy S, Shinde DN, Spiegel R, Timmerman V, von Spiczak S, Helbig I; C4RCDResearch Group; AR working group of the EuroEPINOMICS RES Consortium, Weckhuysen S, Francklyn C, Antonellis A, de Witte P, De Jonghe P. Biallelic VARS variantscause developmental encephalopathy with microcephaly that is recapitulated invars knockout zebrafish. Nat Commun. 2019 Feb 12;10(1):708.

22. Wolking S, May P, Mei D, Møller RS, Balestrini S, Helbig KL, Altuzarra CD, Chatron N, Kaiwar C, Stöhr K, Widdess-Walsh P, Mendelsohn BA, Numis A, Cilio MR, Van Paesschen W, Svendsen LL, Oates S, Hughes E, Goyal S, Brown K, Sifuentes Saenz M, Dorn T, Muhle H, Pagnamenta AT, Vavoulis DV, Knight SJL, Taylor JC, Canevini MP, Darra F, Gavrilova RH, Powis Z, Tang S, Marquetand J, Armstrong M, McHale D, Klee EW, Kluger GJ, Lowenstein DH, Weckhuysen S, Pal DK, Helbig I, Guerrini R, Thomas RH, Rees MI, Lesca G, Sisodiya SM, Weber YG, Lal D, Marini C, Lerche H, Schubert J. Clinical spectrum of STX1B-related epileptic disorders. Neurology. 2019 Mar 12;92(11):e1238-e1249.

23. Cellini E, Vetro A, Conti V, Marini C, Doccini V, Clementella C, Parrini E, Giglio S, Della Monica M, Fichera M, Musumeci SA, Guerrini R. Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. Eur J Hum Genet. 2019 Jun;27(6):909-918.

24. Trivisano M, Pietrafusa N, Terracciano A, Marini C, Mei D, Darra F, Accorsi P, Battaglia D, Caffi L, Canevini MP, Cappelletti S, Cesaroni E, de Palma L, Costa P, Cusmai R, Giordano L, Ferrari A, Freri E, Fusco L, Granata T, Martino T, Mastrangelo M, Bova SM, Parmeggiani L, Ragona F, Sicca F, Striano P, Specchio LM, Tondo I, Zambrelli E, Zamponi N, Zanus C, Boniver C, Vecchi M, Avolio C, Dalla Bernardina B, Bertini E, Guerrini R, Vigevano F, Specchio N. Defining the electroclinical phenotype and outcome of PCDH19-related epilepsy: A multicenter study. Epilepsia. 2018;59(12):2260-2271

25. Marini C, Porro A, Rastetter A, Dalle C, Rivolta I, Bauer D, Oegema R, Nava C, Parrini E, Mei D, Mercer C, Dhamija R, Chambers C, Coubes C, Thévenon J, Kuentz P, Julia S, Pasquier L, Dubourg C, Carré W, Rosati A, Melani F, Pisano T, Giardino M, Innes AM, Alembik Y, Scheidecker S, Santos M, Figueiroa S, Garrido C, Fusco C, Frattini D, Spagnoli C, Binda A, Granata T, Ragona F, Freri E, Franceschetti S, Canafoglia L, Castellotti B, Gellera C, Milanesi R, Mancardi MM, Clark DR, Kok F, Helbig KL, Ichikawa S, Sadler L, Neupauerová J, Laššuthova P, Šterbová K, Laridon A, Brilstra E, Koeleman B, Lemke JR, Zara F, Striano P, Soblet J, Smits G, Deconinck N, Barbuti A, DiFrancesco D, LeGuern E, Guerrini R, Santoro B, Hamacher K, Thiel G, Moroni A, DiFrancesco JC, Depienne C. HCN1 mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain. 2018;141(11):3160-3178.

26. Tonin R, Catarzi S, Caciotti A, Procopio E, Marini C, Guerrini R, Morrone A. Progressive myoclonus epilepsy in Gaucher Disease due to a new Gly-Gly mutation causing loss of an Exonic Splicing Enhancer. J Neurol. 2018 Oct 31. (Epub ahead of print)

27. Gardella E, Marini C, Trivisano M, Fitzgerald MP, Alber M, Howell KB, Darra F, Siliquini S, Bölsterli BK, Masnada S, Pichiecchio A, Johannesen KM, Jepsen B, Fontana E, Anibaldi G, Russo S, Cogliati F, Montomoli M, Specchio N, Rubboli G, Veggiotti P, Beniczky S, Wolff M, Helbig I, Vigevano F, Scheffer IE, Guerrini R, Møller RS. The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology. 2018;91(12):e1112-e1124.

28. May P, Girard S, Harrer M, Bobbili DR, Schubert J, Wolking S, Becker F, Lachance-Touchette P, Meloche C, Gravel M, Niturad CE, Knaus J, De Kovel C, Toliat M, Polvi A, Iacomino M, Guerrero-López R, Baulac S, Marini C, Thiele H, Altmüller J, Jabbari K, Ruppert AK, Jurkowski W, Lal D, Rusconi R, Cestèle S, Terragni B, Coombs ID, Reid CA, Striano P, Caglayan H, Siren A, Everett K, Møller RS, Hjalgrim H, Muhle H, Helbig I, Kunz WS, Weber YG, Weckhuysen S, Jonghe P, Sisodiya SM, Nabbout R, Franceschetti S, Coppola A, Vari MS, Kasteleijn-Nolst Trenité D, Baykan B, Ozbek U, Bebek N, Klein KM, Rosenow F, Nguyen DK, Dubeau F, Carmant L, Lortie A, Desbiens R, Clément JF, Cieuta-Walti C, Sills GJ, Auce P, Francis B, Johnson MR, Marson AG, Berghuis B, Sander JW, Avbersek A, McCormack M, Cavalleri GL, Delanty N, Depondt C, Krenn M, Zimprich F, Peter S, Nikanorova M, Kraaij R, van Rooij J, Balling R, Ikram MA, Uitterlinden AG, Avanzini G, Schorge S, Petrou S, Mantegazza M, Sander T, LeGuern E, Serratosa JM, Koeleman BPC, Palotie A, Lehesjoki AE, Nothnagel M, Nürnberg P, Maljevic S, Zara F, Cossette P, Krause R, Lerche H; Epicure Consortium; EuroEPINOMICS CoGIE Consortium; EpiPGX Consortium. Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. De novo variants in neurodevelopmental disorders with epilepsy. Lancet Neurol. 2018;17(8):699-708.

29. Fassio A, Esposito A, Kato M, Saitsu H, Mei D, Marini C, Conti V, Nakashima M, Okamoto N, Olmez Turker A, Albuz B, Semerci Gündüz CN, Yanagihara K, Belmonte E, Maragliano L, Ramsey K, Balak C, Siniard A, Narayanan V; C4RCD Research Group, Ohba C, Shiina M, Ogata K, Matsumoto N, Benfenati F, Guerrini R. De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain. 2018 Jun 1;141(6):1703-1718.

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dichiara inoltre di:

essere consapevole delle sanzioni penali, previste in caso di dichiarazioni non veritiere e di falsità negli atti e della conseguente decadenza dai benefici di cui agli artt. 75 e 76 del D.P.R. 445/2000;

essere informato che i dati personali raccolti saranno trattati, anche con mezzi informatici, esclusivamente per il procedimento per il quale la dichiarazione viene resa (art. 13 Dlgs 196/2003).

LUOGO e DATA **FIRMA DEL DICHIARANTE***

ANCONA 14/12/2020