Curriculum Vitae Europass

Informazioni personali

Nome/ Cognome	**Carla Marini**
Indirizzo	
Telefoni	
E-mail	
Cittadinanza	Italiana
Data di nascita	
Sesso	F
CF	
Occupazione desiderata/Settore professionale	**Direttore SOD Neuropsichiatria Infantile e Centro Regionale per la Diagnosi e Cura Epilessia Infantile; Presidio Ospedaliero Materno Infantile G. Salesi; AOU 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 Dal 01/11/2017 al 01/01/2020: professore associato con incarichi assistenziali, dipartimento di Neurofarba; Università di Firenze e AUO Meyer.
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	Neuropsichiatria infantile; Ospedale Salesi; Presidio Ospedaliero G. Salesi; AOU Ancona; via Filippo Corridoni 11; 60123 Ancona
Tipo di attività o settore	Neurologia 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	MD, PhD

rilasciata	
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	**Comprensione**				**Parlato**		**Scritto**
Livello europeo (*)	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	Si allega 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 05/03/2024

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; 60123

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

1. D'Onofrio G, Accogli A, Severino M, Caliskan H, Kokotović T, Blazekovic A, Jercic KG, Markovic S, Zigman T, Goran K, Barišić N, Duranovic V, Ban A, Borovecki F, Ramadža DP, Barić I, Fazeli W, Herkenrath P, Marini C, Vittorini R, Gowda V, Bouman A, Rocca C, Alkhawaja IA, Murtaza BN, Rehman MMU, Al Alam C, Nader G, Mancardi MM, Giacomini T, Srivastava S, Alvi JR, Tomoum H, Matricardi S, Iacomino M, Riva A, Scala M, Madia F, Pistorio A, Salpietro V, Minetti C, Rivière JB, Srour M, Efthymiou S, Maroofian R, Houlden H, Vernes SC, Zara F, Striano P, Nagy V. Genotype-phenotype correlation in contactin-associated protein-like 2 (CNTNAP-2) developmental disorder. Hum Genet. 2023 Jul;142(7):909-925. doi: 10.1007/s00439-023-02552-2.

2. Balestrini S, Doccini V, Giometto S, Lucenteforte E, De Masi S, Giarola E, Brambilla I, Pieroni F, Perulli M, Battaglia D, Specchio N, Ragona F, Granata T, Pellacani S, Ferrari A, Marini C, Matricardi S, Cesaroni E, Giordano L, Accorsi P, Sciruicchio V, Tinuper P, Messana T, Russo A, Pruna D, Nosadini M, De Giorgis V, Caputo D; Residras Collaboration Group; Pellegrin S, Lo Barco T, Darra F, Dalla Bernardina B, Guerrini R. A registry for Dravet syndrome: The Italian experience. Epilepsia Open. 2023 Jun;8(2):517-534. doi: 10.1002/epi4.12730

3. Matricardi S, Cestèle S, Trivisano M, Kassabian B, Leroudier N, Vittorini R, Nosadini M, Cesaroni E, Siliquini S, Marinaccio C, Longaretti F, Podestà B, Operto FF, Luisi C, Sartori S, Boniver C, Specchio N, Vigevano F, Marini C, Mantegazza M. Gain of function SCN1A disease-causing variants: Expanding the phenotypic spectrum and functional studies guiding the choice of effective antiseizure medication. Epilepsia. 2023 May;64(5):1331-1347. doi: 10.1111/epi.17509.

4. Varesio C, De Giorgis V, Veggiotti P, Nardocci N, Granata T, Ragona F, Pasca L, Mensi MM, Borgatti R, Olivotto S, Previtali R, Riva A, Mancardi MM, Striano P, Cavallin M, Guerrini R, Operto FF, Pizzolato A, Di Maulo R, Martino F, Lodi A, Marini C. GLUT1-DS Italian registry: past, present, and future: a useful tool for rare disorders. Orphanet J Rare Dis. 2023 Mar 21;18(1):63. doi: 10.1186/s13023-023-02628-2.

5. Matricardi S, Cesaroni E, Bonanni P, Foschi N, D Aniello A, Di Gennaro G, Striano P, Cappanera S, Siliquini S, Freri E, Ragona F, Granata T, Deleo F, Villani F, Russo A, Messana T, Siri L, Bagnasco I, Vignoli A, Operto FF, Orsini A, Bonuccelli A, Papa A, Peruzzi C, Liguori C, Verrotti A, Chiarelli F, Marini C, Lattanzi S. Long-term effectiveness of add-on perampanel in patients

with Lennox-Gastaut syndrome: A multicenter retrospective study. Epilepsia. 2023 Jun;64(6):e98-e104. doi: 10.1111/epi.17601

6. Riva A, Roberti R, D'Onofrio G, Vari MS, Amadori E, De Giorgis V, Cerminara C, Specchio N, Pietrafusa N, Tombini M, Assenza G, Cappanera S, Marini C, Rasmini P, Veggiotti P, Zara F, Russo E, Striano P. A real-life pilot study of the clinical application of pharmacogenomics testing on saliva in epilepsy. Epilepsia Open. 2023 Feb 25. doi: 10.1002/epi4.12717.

7. Johannesen KM, Liu Y, Koko M, Gjerulfsen CE, Sonnenberg L, Schubert J, Fenger CD, Eltokhi A, Rannap M, Koch NA, Lauxmann S, Krüger J, Kegele J, Canafoglia L, Franceschetti S, Mayer T, Rebstock J, Zacher P, Ruf S, Alber M, Sterbova K, Lassuthová P, Vlckova M, Lemke JR, Platzer K, Krey I, Heine C, Wieczorek D, Kroell-Seger J, Lund C, Klein KM, Au PYB, Rho JM, Ho AW, Masnada S, Veggiotti P, Giordano L, Accorsi P, Hoei-Hansen CE, Striano P, Zara F, Verhelst H, Verhoeven JS, Braakman HMH, van der Zwaag B, Harder AVE, Brilstra E, Pendziwiat M, Lebon S, Vaccarezza M, Le NM, Christensen J, Grønborg S, Scherer SW, Howe J, Fazeli W, Howell KB, Leventer R, Stutterd C, Walsh S, Gerard M, Gerard B, Matricardi S, Bonardi CM, Sartori S, Berger A, Hoffman-Zacharska D, Mastrangelo M, Darra F, Vøllo A, Motazacker MM, Lakeman P, Nizon M, Betzler C, Altuzarra C, Caume R, Roubertie A, Gélisse P, Marini C, Guerrini R, Bilan F, Tibussek D, Koch-Hogrebe M, Perry MS, Ichikawa S, Dadali E, Sharkov A, Mishina I, Abramov M, Kanivets I, Korostelev S, Kutsev S, Wain KE, Eisenhauer N, Wagner M, Savatt JM, Müller-Schlüter K, Bassan H, Borovikov A, Nassogne MC, Destrée A, Schoonjans AS, Meuwissen M, Buzatu. Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain. 2022 Sep 14;145(9):2991-3009. doi: 10.1093/brain/awab321.

8. Stamberger H, Crosiers D, Balagura G, Bonardi CM, Basu A, Cantalupo G, Chiesa V, Christensen J, Dalla Bernardina B, Ellis CA, Furia F, Gardiner F, Giron C, Guerrini R, Klein KM, Korff C, Krijtova H, Leffler M, Lerche H, Lesca G, Lewis-Smith D, Marini C, Marjanovic D, Mazzola L, McKeown Ruggiero S, Mochel F, Ramond F, Reif PS, Richard-Mornas A, Rosenow F, Schropp C, Thomas RH, Vignoli A, Weber Y, Palmer E, Helbig I, Scheffer IE, Striano P, Møller RS, Gardella E, Weckhuysen S. Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood.Neurology. 2022 Jul 19;99(3):e221-e233. doi: 10.1212/WNL.0000000000200715

9. Matricardi S, Casciato S, Bozzetti S, Mariotto S, Stabile A, Freri E, Deleo F, Sartori S, Nosadini M, Pappalardo I, Meletti S, Giovannini G, Zucchi E, Di Bonaventura C, Di Gennaro G, Ferrari S, Zuliani L, Zoccarato M, Vogrig A, Lattanzi S, Michelucci R, Gambardella A, Ferlazzo E, Fusco L, Granata T, Villani F; Immune Epilepsies Study Group of the Italian League Against Epilepsy. Epileptic phenotypes in autoimmune encephalitis: from acute symptomatic seizures to autoimmune-associated epilepsy. J Neurol Neurosurg Psychiatry. 2022 Jul 25:jnnp-2022-329195. doi: 10.1136/jnnp-2022-329195.

10. Antonella Riva, Antonietta Coppola, Carlo DiBonaventura, Maurizio Elia, Edoardo Ferlazzo, Giuseppe Gobbi, Carla Marini, Stefano Meletti, Antonino Romeo, Katia Santoro, Alberto Verrotti, Giuseppe Capovilla, Pasquale Striano. An Italian consensus on the management of Lennox-Gastaut syndrome. Seizure; 2022,101; 134-140

11. Marini C, Giardino M. Novel treatments in epilepsy guided by genetic diagnosis. Br J Clin Pharmacol. 2022 Jun;88(6):2539-2551. doi: 10.1111/bcp.15139.

12. Balagura G, Xian J, Riva A, Marchese F, Ben Zeev B, Rios L, Sirsi D, Accorsi P, Amadori E, Astrea G, Baldassari S, Beccaria F, Boni A, Budetta M, Cantalupo G, Capovilla G, Cesaroni E, Chiesa V, Coppola A, Dilena R, Faggioli R, Ferrari A, Fiorini E, Madia F, Gennaro E, Giacomini T, Giordano L, Iacomino M, Lattanzi S, Marini C, Mancardi MM, Mastrangelo M, Messana T, Minetti C, Nobili L, Papa A, Parmeggiani A, Pisano T, Russo A, Salpietro V, Savasta S, Scala M, Accogli A, Scelsa B, Scudieri P, Spalice A, Specchio N, Trivisano M, Tzadok M, Valeriani M, Vari MS, Verrotti A, Vigevano F, Vignoli A, Toonen R, Zara F, Helbig I, Striano P. Epilepsy Course and Developmental Trajectories in STXBP1-DEE. Neurol Genet. 2022 May 31;8(3):e676. doi: 10.1212/NXG.0000000000000676.

13. Rosati A, L'Erario M, Bianchi R, Olivotto S, Battaglia DI, Darra F, Biban P, Biggeri A, Catelan D, Danieli G, Mondardini MC, Cordelli DM, Amigoni A, Cesaroni E, Conio A, Costa P, Lombardini M, Meleleo R, Pugi A, Tornaboni EE, Santarone ME, Vittorini R, Sartori S, Marini C, Vigevano F, Mastrangelo M, Pulitanò SM, Izzo F, Fusco L. KETASER01 protocol: What went right and what went wrong. Epilepsia Open. 2022 Jul 14. doi: 10.1002/epi4.12627.

14. Khan AQ, Coorg RK, Gill D, Marini C, Myers KA. Koolen-de Vries syndrome associated with continuous spike-wave in sleep. Epileptic Disord. 2022 Oct 1;24(5):1-6. doi: 10.1684/epd.2022.1452.

15. Stamberger H, Crosiers D, Balagura G, Bonardi CM, Basu A, Cantalupo G, Chiesa V, Christensen J, Dalla Bernardina B, Ellis CA, Furia F, Gardiner F, Giron C, Guerrini R, Klein KM, Korff C, Krijtova H, Leffner M, Lerche H, Lesca G, Lewis-Smith D, Marini C, Marjanovic D, Mazzola L, McKeown Ruggiero S, Mochel F, Ramond F, Reif PS, Richard-Mornas A, Rosenow F, Schropp C, Thomas RH, Vignoli A, Weber Y, Palmer E, Helbig I, Scheffer IE, Striano P, Møller RS, Gardella E, Weckhuysen S. Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology. 2022 Jul 19;99(3):e221-e233. doi: 10.1212/WNL.0000000000200715.

16. Monti Guarnieri N, Pompilio A, Marini C, Ortenzi GB, Andresciani E, Garzone AMF, Ieracitano MC, Polidori C. A pharmacovigilance study on antiepileptic medications in a paediatric hospital in Italy. Eur J Hosp Pharm. 2022 Apr 11:ejhpharm-2021-003053. doi: 10.1136/ejhpharm-2021-003053.

17. Manivannan SN, Roovers J, Smal N, Myers CT, Turkdogan D, Roelens F, Kanca O, Chung HL, Scholz T, Hermann K, Bierhals T, Caglayan HS, Stamberger H; MAE Working Group of EuroEPINOMICS RES Consortium, Mefford H, de Jonghe P, Yamamoto S, Weckhuysen S, Bellen HJ. De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain. 2022 Jun 3;145(5):1684-1697. doi: 10.1093/brain/awab409.

18. Matricardi S, Casciato S, Bozzetti S, Mariotto S, Stabile A, Freri E, Deleo F, Sartori S, Nosadini M, Pappalardo I, Meletti S, Giovannini G, Zucchi E, Di Bonaventura C, Di Gennaro G, Ferrari S, Zuliani L, Zoccarato M, Vogrig A, Lattanzi S, Michelucci R, Gambardella A, Ferlazzo E, Fusco L, Granata T, Villani F; Immune Epilepsies Study Group of the Italian League Against Epilepsy. Epileptic phenotypes in autoimmune encephalitis: from acute symptomatic seizures to

autoimmune-associated epilepsy. J Neurol Neurosurg Psychiatry. 2022 Jul 25:jnnp-2022-329195. doi: 10.1136/jnnp-2022-329195.

19. Cesaroni E, Matricardi S, Cappanera S, Marini C. First reported case of an inherited PACS2 pathogenic variant with variable expression. Epileptic Disord. 2022 Jun 1;24(3):572-576. doi: 10.1684/epd.2022.1417.

20. Costa AM, Lo Barco T, Spezia E, Conti V, Roli L, Marini L, Minghetti S, Caramaschi E, Pietrangelo L, Pecoraro L, D'Achille F, Accorsi P, Trenti T, Melani F, Marini C, Guerrini R, Darra F, Bergonzini P, Biagini G. Prospective Evaluation of Ghrelin and Des-Acyl Ghrelin Plasma Levels in Children with Newly Diagnosed Epilepsy: Evidence for Reduced Ghrelin-to-Des-Acyl Ghrelin Ratio in Generalized Epilepsies. J Pers Med. 2022 Mar 25;12(4):527. doi: 10.3390/jpm12040527.

21. 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

22. 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 Dec;60 S3:S2-S7.

23. 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.

24. 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.

25. 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.

26. 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.

27. 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.

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

29. 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.

30. 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.

31. 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.

32. 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.

33. 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.

34. 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.

35. 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.

36. 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

37. 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.

38. 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. 2019;266(1):92-101.

39. 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.

40. 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.

41. 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;141(6):1703-1718.

42. Heyne HO, Singh T, Stamberger H, Abou Jamra R, Caglayan H, Craiu D, De Jonghe P, Guerrini R, Helbig KL, Koeleman BPC, Kosmicki JA, Linnankivi T, May P, Muhle H, Møller RS, Neubauer BA, Palotie A, Pendziwiat M, Striano P, Tang S, Wu S; EuroEPINOMICS RES Consortium, Poduri A, Weber YG, Weckhuysen S, Sisodiya SM, Daly MJ, Helbig I, Lal D, Lemke JR. De novo variants in neurodevelopmental disorders with epilepsy. Nat Genet. 2018;50(7):1048-1053.

43. 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;141(6):1703-1718.

44. Marini C, Romoli M, Parrini E, Costa C, Mei D, Mari F, Parmeggiani L, Procopio E, Metitieri T, Cellini E, Virdò S, De Vita D, Gentile M, Prontera P, Calabresi P, Guerrini R. Clinical features and outcome of 6 new patients carrying de novo KCNB1 gene mutations. Neurol Genet. 2017;3(6):e206.

45. Marini C, Hardies K, Pisano T, May P, Weckhuysen S, Cellini E, Suls A, Mei D, Balling R, Jonghe PD, Helbig I, Garozzo D; EuroEPINOMICS consortium AR working group, Guerrini R. Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects. Am J Med Genet A. 2017;173(4):1119-1123.

46. Mei D, Parrini E, Marini C, Guerrini R. The Impact of Next-Generation Sequencing on the Diagnosis and Treatment of Epilepsy in Paediatric Patients. Mol Diagn Ther. 2017;21(4):357-373

47. Parrini E, Marini C, Mei D, Galuppi A, Cellini E, Pucatti D, Chiti L, Rutigliano D, Bianchini C, Virdò S, De Vita D, Bigoni S, Barba C, Mari F, Montomoli M, Pisano T, Rosati A; Clinical Study Group, Guerrini R. Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. Hum Mutat. 2017;38(2):216-225.

48. EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium. De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. Am J Hum Genet. 2014 Oct 2;95(4):360-70. Epub 2014 Sep 25. Erratum in: Am J Hum Genet. 2017;100(1):179.

49. Møller RS, Wuttke TV, Helbig I, Marini C, Johannesen KM, Brilstra EH, Vaher U, Borggraefe I, Talvik I, Talvik T, Kluger G, Francois LL, Lesca G, de Bellescize J, Blichfeldt S, Chatron N, Holert N, Jacobs J, Swinkels M, Betzler C, Syrbe S, Nikanorova M, Myers CT, Larsen LH, Vejzovic S, Pendziwiat M, von Spiczak S, Hopkins S, Dubbs H, Mang Y, Mukhin K, Holthausen H, van Gassen KL, Dahl HA, Tommerup N, Mefford HC, Rubboli G, Guerrini R, Lemke JR, Lerche H, Muhle H, Maljevic S. Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. Neurology. 2017;88(5):483-492.

50. Cetica V, Chiari S, Mei D, Parrini E, Grisotto L, Marini C, Pucatti D, Ferrari A, Sicca F, Specchio N, Trivisano M, Battaglia D, Contaldo I, Zamponi N, Petrelli C, Granata T, Ragona F, Avanzini

G, Guerrini R. Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations. Neurology. 2017;88(11):1037-1044.

51. Sharma S, Goel S, Jain P, Marini C, Mei D. CDKL5 Encephalopathy in an Indian Girl: Partial Response to the Modified Atkins Diet. J Pediatr Neurosci. 2017;12(1):116-117

52. Epi4K Consortium; EuroEPINOMICS-RES Consortium; Epilepsy Phenome Genome Project. Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. Eur J Hum Genet. 2017;25(7):894-899.

53. Santiago-Sim T, Burrage LC, Ebstein F, Tokita MJ, Miller M, Bi W, Braxton AA, Rosenfeld JA, Shahrour M, Lehmann A, Cogné B, Küry S, Besnard T, Isidor B, Bézieau S, Hazart I, Nagakura H, Immken LL, Littlejohn RO, Roeder E; EuroEPINOMICS RES Consortium Autosomal Recessive working group, S. Hande Caglayan, Kara B, Hardies K, Weckhuysen S, May P, Lemke JR, Elpeleg O, Abu-Libdeh B, James KN, Silhavy JL, Issa MY, Zaki MS, Gleeson JG, Seavitt JR, Dickinson ME, Ljungberg MC, Wells S, Johnson SJ, Teboul L, Eng CM, Yang Y, Kloetzel PM, Heaney JD, Walkiewicz MA. Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. Am J Hum Genet. 2017;100(4):676-688.

54. Tang S, Hughes E, Lascelles K; EuroEPINOMICS RES myoclonic astatic epilepsy working group, Simpson MA, Pal DK. New SMARCA2 mutation in a patient with Nicolaides-Baraitser syndrome and myoclonic astatic epilepsy. Am J Med Genet A. 2017;173(1):195-199.

55. de Lange IM, Helbig KL, Weckhuysen S, Møller RS, Velinov M, Dolzhanskaya N, Marsh E, Helbig I, Devinsky O, Tang S, Mefford HC, Myers CT, van Paesschen W, Striano P, van Gassen K, van Kempen M, de Kovel CG, Piard J, Minassian BA, Nezarati MM, Pessoa A, Jacquette A, Maher B, Balestrini S, Sisodiya S, Warde MT, De St Martin A, Chelly J; EuroEPINOMICS-RES MAE working group, van 't Slot R, Van Maldergem L, Brilstra EH, Koeleman BP. De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. J Med Genet. 2016;53(12):850-858

56. Tessa A, Battini R, Rubegni A, Storti E, Marini C, Galatolo D, Pasquariello R, Santorelli FM. Identification of mutations in AP4S1/SPG52 through next generation sequencing in three families. Eur J Neurol. 2016;23(10):1580

57. Johannesen K, Marini C, Pfeffer S, Møller RS, Dorn T, Niturad CE, Gardella E, Weber Y, Søndergård M, Hjalgrim H, Nikanorova M, Becker F, Larsen LH, Dahl HA, Maier O, Mei D, Biskup S, Klein KM, Reif PS, Rosenow F, Elias AF, Hudson C, Helbig KL, Schubert-Bast S, Scordo MR, Craiu D, Djémié T, Hoffman-Zacharska D, Caglayan H, Helbig I, Serratosa J, Striano P, De Jonghe P, Weckhuysen S, Suls A, Muru K, Talvik I, Talvik T, Muhle H, Borggraefe I, Rost I, Guerrini R, Lerche H, Lemke JR, Rubboli G, Maljevic S. Phenotypic spectrum of GABRA1: From generalized epilepsies to severe epileptic encephalopathies. Neurology. 2016;87(11):1140-51.

58. Mignot C, von Stülpnagel C, Nava C, Ville D, Sanlaville D, Lesca G, Rastetter A, Gachet B, Marie Y, Korenke GC, Borggraefe I, Hoffmann-Zacharska D, Szczepanik E, Rudzka-Dybała M, Yiş U, Çağlayan H, Isapof A, Marey I, Panagiotakaki E, Korff C, Rossier E, Riess A, Beck-Woedl S, Rauch A, Zweier C, Hoyer J, Reis A, Mironov M, Bobylova M, Mukhin K, Hernandez-Hernandez L, Maher B, Sisodiya S, Kuhn M, Glaeser D, Weckhuysen S, Myers CT, Mefford HC, Hörtnagel K, Biskup S; EuroEPINOMICS-RES MAE working group, Lemke JR, Héron D, Kluger G, Depienne C. Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. J Med Genet. 2016 Aug;53(8):511-22. doi: 10.1136/jmedgenet-2015-103451. Epub 2016 Mar 17. Erratum in: J Med Genet. 2016 Oct;53(10):720.

59. Djémié T, Weckhuysen S, von Spiczak S, Carvill GL, Jaehn J, Anttonen AK, Brilstra E, Caglayan HS, de Kovel CG, Depienne C, Gaily E, Gennaro E, Giraldez BG, Gormley P, Guerrero-López R, Guerrini R, Hämäläinen E, Hartmann C, Hernandez-Hernandez L, Hjalgrim H, Koeleman BP, Leguern E, Lehesjoki AE, Lemke JR, Leu C, Marini C, McMahon JM, Mei D, Møller RS, Muhle H, Myers CT, Nava C, Serratosa JM, Sisodiya SM, Stephani U, Striano P, van Kempen MJ, Verbeek NE, Usluer S, Zara F, Palotie A, Mefford HC, Scheffer IE, De Jonghe P, Helbig I, Suls A; EuroEPINOMICS-RES Dravet working group. Pitfalls in genetic testing: the story of missed SCN1A mutations. Mol Genet Genomic Med. 2016;4(4):457-64.

60. de Kovel CG, Brilstra EH, van Kempen MJ, Van't Slot R, Nijman IJ, Afawi Z, De Jonghe P, Djémié T, Guerrini R, Hardies K, Helbig I, Hendrickx R, Kanaan M, Kramer U, Lehesjoki AE, Lemke JR, Marini C, Mei D, Møller RS, Pendziwiat M, Stamberger H, Suls A, Weckhuysen S; EuroEPINOMICS RES Consortium, Koeleman BP. Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Mol Genet Genomic Med. 2016;4(5):568-80.

61. Tinuper P, Bisulli F, Cross JH, Hesdorffer D, Kahane P, Nobili L, Provini F, Scheffer IE, Tassi L, Vignatelli L, Bassetti C, Cirignotta F, Derry C, Gambardella A, Guerrini R, Halasz P, Licchetta L, Mahowald M, Manni R, Marini C, Mostacci B, Naldi I, Parrino L, Picard F, Pugliatti M, Ryvlin P, Vigevano F,Zucconi M, Berkovic S, Ottman R. Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology. 2016;86(19):1834-42.

62. Ricos MG, Hodgson BL, Pippucci T, Saidin A, Ong YS, Heron SE, Licchetta L, Bisulli F, Bayly MA, Hughes J, Baldassari S, Palombo F, Epilepsy Electroclinical Study Group, Santucci M, Meletti S, Berkovic SF, Rubboli G, Thomas PQ, Scheffer IE, Tinuper P, Geoghegan J, Schreiber AW, Dibbens LM. Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Ann Neurol. 2016;79(1):120-31.

63. Mignot C, von Stülpnagel C, Nava C, Ville D, Sanlaville D, Lesca G, Rastetter A, Gachet B, Marie Y, Korenke GC, Borggraefe I, Hoffmann-Zacharska D, Szczepanik E, Rudzka-Dybała M, Yiş U, Çağlayan H, Isapof A, Marey I, Panagiotakaki E, Korff C, Rossier E, Riess A, Beck-Woedl S, Rauch A, Zweier C, Hoyer J, Reis A, Mironov M, Bobylova M, Mukhin K, Hernandez-Hernandez L, Maher B, Sisodiya S, Kuhn M, Glaeser D, Weckhuysen S, Myers CT, Mefford HC, Hörtnagel K, Biskup S; EuroEPINOMICS-RES MAE working group, Lemke JR, Héron D, Kluger G, Depienne C. Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. J Med Genet. 2016;53(8):511-22. Erratum in: J Med Genet. 2016;53(10):720.

64. Larsen J, Johannesen KM, Ek J, Tang S, Marini C, Blichfeldt S, Kibaek M, von Spiczak S, Weckhuysen S, Frangu M, Neubauer BA, Uldall P, Striano P, Zara F; MAE working group of EuroEPINOMICS RES Consortium, Kleiss R, Simpson M, Muhle H, Nikanorova M, Jepsen B, Tommerup N, Stephani U, Guerrini R, Duno M, Hjalgrim H, Pal D, Helbig I, Møller RS. The role of SLC2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of GLUT1 deficiency syndrome. Epilepsia. 2015;56(12):e203-8.

65. Hardies K, de Kovel CG, Weckhuysen S, Asselbergh B, Geuens T, Deconinck T, Azmi A, May P, Brilstra E, Becker F, Barisic N, Craiu D, Braun KP, Lal D, Thiele H, Schubert J, Weber Y, van 't Slot R, Nürnberg P, Balling R, Timmerman V, Lerche H, Maudsley S, Helbig I, Suls A, Koeleman BP, De Jonghe P; autosomal recessive working group of the EuroEPINOMICS RES Consortium. Recessive mutations in SLC13A5 result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain. 2015;138(Pt 11):3238-50.

66. Hildebrand MS, Tankard R, Gazina EV, Damiano JA, Lawrence KM, Dahl HH, Regan BM, Shearer AE, Smith RJ, Marini C, Guerrini R, Labate A, Gambardella A, Tinuper P, Lichetta L, Baldassari S, Bisulli F, Pippucci T, Scheffer IE, Reid CA, Petrou S, Bahlo M, Berkovic SF.

PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Ann Clin Transl Neurol. 2015;2(8):821-30.

67. Ilvento L, Rosati A, Marini C, L'Erario M, Mirabile L, Guerrini R. Ketamine in refractory convulsive status epilepticus in children avoids endotracheal intubation. Epilepsy Behav. 2015;49:343-6.

68. Tan C, Shard C, Ranieri E, Hynes K, Pham DH, Leach D, Buchanan G, Corbett M, Shoubridge C, Kumar R, Douglas E, Nguyen LS, Mcmahon J, Sadleir L, Specchio N, Marini C, Guerrini R, Moller RS, Depienne C, Haan E, Thomas PQ, Berkovic SF, Scheffer IE, Gecz J. Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Hum Mol Genet. 2015;24(18):5250-9.

69. Pisano T, Numis AL, Heavin SB, Weckhuysen S, Angriman M, Suls A, Podesta B, Thibert RL, Shapiro KA, Guerrini R, Scheffer IE, Marini C, Cilio MR. Early and effective treatment of KCNQ2 encephalopathy. Epilepsia. 2015;56(5):685-91.

70. Carvill GL, McMahon JM, Schneider A, Zemel M, Myers CT, Saykally J, Nguyen J, Robbiano A, Zara F, Specchio N, Mecarelli O, Smith RL, Leventer RJ, Møller RS, Nikanorova M, Dimova P, Jordanova A, Petrou S; EuroEPINOMICS Rare Epilepsy Syndrome Myoclonic-Astatic Epilepsy & Dravet working group, Helbig I, Striano P, Weckhuysen S, Berkovic SF, Scheffer IE, Mefford HC. Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. Am J Hum Genet. 2015;96(5):808-15.

71. Conti V, Aracri P, Chiti L, Brusco S, Mari F, Marini C, Albanese M, Marchi A, Liguori C, Placidi F, Romigi A, Becchetti A, Guerrini R. Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function. Neurology. 2015;84(15):1520-8.

72. Mumoli L, Tarantino P, Michelucci R, Bianchi A, Labate A, Franceschetti S, Marini C, Striano P, Gagliardi M, Ferlazzo E, Sofia V, Pennese L, Annesi G, Aguglia U, Guerrini R, Zara F, Gambardella A; Genetic Commission, Italian League Against Epilepsy. No evidence of a role for cystatin B gene in juvenile myoclonic epilepsy. Epilepsia. 2015;56(4):e40-3

73. Syrbe S, Hedrich UB, Riesch E, Djémié T, Müller S, Møller RS, Maher B, Hernandez-Hernandez L, Synofzik M, Caglayan HS, Arslan M, Serratosa JM, Nothnagel M, May P, Krause R, Löffler H, Detert K, Dorn T, Vogt H, Krämer G, Schöls L, Mullis PE, Linnankivi T, Lehesjoki AE, Sterbova K, Craiu DC, Hoffman-Zacharska D, Korff CM, Weber YG, Steinlin M, Gallati S, Bertsche A, Bernhard MK, Merkenschlager A, Kiess W; EuroEPINOMICS RES, Gonzalez M, Züchner S, Palotie A, Suls A, De Jonghe P, Helbig I, Biskup S, Wolff M, Maljevic S, Schüle R, Sisodiya SM, Weckhuysen S, Lerche H, Lemke JR. De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nat Genet. 2015;47(4):393-9

74. Larsen J, Carvill GL, Gardella E, Kluger G, Schmiedel G, Barisic N, Depienne C, Brilstra E, Mang Y, Nielsen JE, Kirkpatrick M, Goudie D, Goldman R, Jähn JA, Jepsen B, Gill D, Döcker M, Biskup S, McMahon JM, Koeleman B, Harris M, Braun K, de Kovel CG, Marini C, Specchio N, Djémié T, Weckhuysen S, Tommerup N, Troncoso M, Troncoso L, Bevot A, Wolff M, Hjalgrim H, Guerrini R, Scheffer IE, Mefford HC,Møller RS; EuroEPINOMICS RES Consortium CRP. The phenotypic spectrum of SCN8A encephalopathy. Neurology. 2015 Feb 3;84(5):480-9.

75. EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium. De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. Am J Hum Genet. 2014 Oct 2;95(4):360-70.

76. Nava C, Dalle C, Rastetter A, Striano P, de Kovel CG, Nabbout R, Cancès C,Ville D, Brilstra EH, Gobbi G, Raffo E, Bouteiller D, Marie Y, Trouillard O, Robbiano A, Keren B, Agher D, Roze E, Lesage S, Nicolas A, Brice A, Baulac M, Vogt C, El Hajj N, Schneider E, Suls A, Weckhuysen S,

Gormley P, Lehesjoki AE, DeJonghe P, Helbig I, Baulac S, Zara F, Koeleman BP; EuroEPINOMICS RES Consortium, Haaf T, LeGuern E, Depienne C. De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nat Genet. 2014 Jun;46(6):640-5.

77. Guerrini R, Marini C, Mantegazza M. Genetic epilepsy syndromes without structural brain abnormalities: clinical features and experimental models. Neurotherapeutics. 2014 Apr;11(2):269-85

78. Mei D, Darra F, Barba C, Marini C, Fontana E, Chiti L, Parrini E, Dalla Bernardina B, Guerrini R. Optimizing the molecular diagnosis of CDKL5 gene-related epileptic encephalopathy in boys. Epilepsia. 2014;55(11):1748-53

79. Suls A, Jaehn JA, Kecskés A, Weber Y, Weckhuysen S, Craiu DC, Siekierska A, Djémié T, Afrikanova T, Gormley P, von Spiczak S, Kluger G, Iliescu CM, Talvik T, Talvik I, Meral C, Caglayan HS, Giraldez BG, Serratosa J, Lemke JR, Hoffman-Zacharska D, Szczepanik E, Barisic N, Komarek V, Hjalgrim H, Møller RS,Linnankivi T, Dimova P, Striano P, Zara F, Marini C, Guerrini R, Depienne C, Baulac S, Kuhlenbäumer G, Crawford AD, Lehesjoki AE, de Witte PA, Palotie A, Lerche H, Esguerra CV, De Jonghe P, Helbig I; EuroEPINOMICS RES Consortium. De novo loss-of-function mutations in CHD2 cause a fever-sensitive myoclonic epileptic encephalopathy sharing features with Dravet syndrome. Am J Hum Genet. 2013;93(5):967-75

80. Weckhuysen S, Ivanovic V, Hendrickx R, Van Coster R, Hjalgrim H, Møller RS, Grønborg S, Schoonjans AS, Ceulemans B, Heavin SB, Eltze C, Horvath R, Casara G, Pisano T, Giordano L, Rostasy K, Haberlandt E, Albrecht B, Bevot A, Benkel I, Syrbe S, Sheidley B, Guerrini R, Poduri A, Lemke JR, Mandelstam S, Scheffer I, Angriman M, Striano P, Marini C, Suls A, De Jonghe P; KCNQ2 Study Group. Extending the KCNQ2 encephalopathy spectrum: clinical and neuroimaging findings in 17 patients. Neurology. 2013;81(19):1697-703

81. Fabbri M, Marini C, Bisulli F, Di Vito L, Elia A, Guerrini R, Mei D, Tinuper P. Clinical and polygraphic study of familial paroxysmal kinesigenic dyskinesia with PRRT2 mutation. Epileptic Disord. 2013;15(2):123-7

82. Chen TT, Klassen TL, Goldman AM, Marini C, Guerrini R, Noebels JL. Novel brain expression of ClC-1 chloride channels and enrichment of CLCN1 variants in epilepsy. Neurology. 2013;80(12):1078-85.

83. Hirose S, Scheffer IE, Marini C, De Jonghe P, Andermann E, Goldman AM, Kauffman M, Tan NC, Lowenstein DH, Sisodiya SM, Ottman R, Berkovic SF; Genetics Commission of the International League Against Epilepsy. SCN1A testing for epilepsy: application in clinical practice. Epilepsia. 2013 ;54(5):946-52.

84. Kim YO, Dibbens L, Marini C, Suls A, Chemaly N, Mei D, McMahon JM, Iona X, Berkovic SF, De Jonghe P, Guerrini R, Nabbout R, Scheffer IE. Do mutations in SCN1B cause Dravet syndrome? Epilepsy Res. 2013;103(1):97-100.

85. Marini C, Conti V, Mei D, Battaglia D, Lettori D, Losito E, Bruccini G, Tortorella G, Guerrini R. PRRT2 mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. Neurology. 2012;79(21):2109-14.

86. Scheffer IE, Grinton BE, Heron SE, Kivity S, Afawi Z, Iona X, Goldberg-Stern H, Kinali M, Andrews I, Guerrini R, Marini C, Sadleir LG, Berkovic SF, Dibbens LM. PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology. 2012;79:2104-8.

87. EPICURE Consortium; EMINet Consortium, Steffens M, Leu C, Ruppert AK, Zara F, Striano P, Robbiano A, Capovilla G, Tinuper P, Gambardella A, Bianchi A, La Neve A, Crichiutti G, de Kovel CG, Kasteleijn-Nolst Trenité D, de Haan GJ, Lindhout D, Gaus V, Schmitz B, Janz D,

Weber YG, Becker F, Lerche H, Steinhoff BJ, Kleefuß-Lie AA, Kunz WS, Surges R, Elger CE, Muhle H, von Spiczak S, Ostertag P, Helbig I, Stephani U, Møller RS, Hjalgrim H, Dibbens LM, Bellows S, Oliver K, Mullen S, Scheffer IE, Berkovic SF, Everett KV, Gardiner MR, Marini C, Guerrini R, Lehesjoki AE, Siren A, Guipponi M, Malafosse A, Thomas P, Nabbout R, Baulac S, Leguern E, Guerrero R, Serratosa JM, Reif PS, Rosenow F, Mörzinger M, Feucht M, Zimprich F, Kapser C, Schankin CJ, Suls A, Smets K, De Jonghe P, Jordanova A, Caglayan H, Yapici Z, Yalcin DA, Baykan B, Bebek N, Ozbek U, Gieger C, Wichmann HE, Balschun T, Ellinghaus D, Franke A, Meesters C, Becker T, Wienker TF, Hempelmann A, Schulz H, Rüschendorf F, Leber M, Pauck SM, Trucks H, Toliat MR, Nürnberg P, Avanzini G, Koeleman BP, Sander T. Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. Hum Mol Genet. 2012;21(24):5359-72.

88. Marini C, Darra F, Specchio N, Mei D, Terracciano A, Parmeggiani L, Ferrari A, Sicca F, Mastrangelo M, Spaccini L, Canopoli ML, Cesaroni E, Zamponi N, Caffi L, Ricciardelli P, Grosso S, Pisano T, Canevini MP, Granata T, Accorsi P, Battaglia D, Cusmai R, Vigevano F, Dalla Bernardina B, Guerrini R. Focal seizures with affective symptoms are a major feature of PCDH19 gene-related epilepsy. Epilepsia. 2012;53(12):2111-9.

89. Pisano T, Barkovich AJ, Leventer RJ, Squier W, Scheffer IE, Parrini E, Blaser S, Marini C, Robertson S, Tortorella G, Rosenow F, Thomas P, McGillivray G, Andermann E, Andermann F, Berkovic SF, Dobyns WB, Guerrini R. Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology. 2012;79(12):1244-51.

90. Striano P, Weber YG, Toliat MR, Schubert J, Leu C, Chaimana R, Baulac S, Guerrero R, LeGuern E, Lehesjoki AE, Polvi A, Robbiano A, Serratosa JM, Guerrini R, Nürnberg P, Sander T, Zara F, Lerche H, Marini C; EPICURE Consortium. GLUT1mutations are a rare cause of familial idiopathic generalized epilepsy. Neurology. 2012;78(8):557-62

91. EPICURE Consortium, Leu C, de Kovel CG, Zara F, Striano P, Pezzella M Robbiano A, Bianchi A, Bisulli F, Coppola A, Giallonardo AT, Beccaria F, Trenité DK, Lindhout D, Gaus V, Schmitz B, Janz D, Weber YG, Becker F, Lerche H, Kleefuss-Lie AA, Hallman K, Kunz WS, Elger CE, Muhle H, Stephani U, Møller RS, Hjalgrim H, Mullen S, Scheffer IE, Berkovic SF, Everett KV, Gardiner MR, Marini C, Guerrini R, Lehesjoki AE, Siren A, Nabbout R, Baulac S, Leguern E, Serratosa JM, Rosenow F, Feucht M, Unterberger I, Covanis A, Suls A, Weckhuysen S, Kaneva R, Caglayan H, Turkdogan D, Baykan B, Bebek N, Ozbek U, Hempelmann A, Schulz H, Rüschendorf F, Trucks H, Nürnberg P, Avanzini G, Koeleman BP, Sander T. Genome-wide linkage meta-analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia. 2012;53(2):308-18.

92. Paciorkowski AR, Thio LL, Rosenfeld JA, Gajecka M, Gurnett CA, Kulkarni S, Chung WK, Marsh ED, Gentile M, Reggin JD, Wheless JW, Balasubramanian S, Kumar R, Christian SL, Marini C, Guerrini R, Maltsev N, Shaffer LG, Dobyns WB. Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function Eur J Hum Genet. 2011;19(12):1238-45

93. Marini C, Scheffer IE, Nabbout R, Suls A, De Jonghe P, Zara F, Guerrini R. The genetics of Dravet syndrome. Epilepsia. 2011;52 Suppl 2:24-9.

94. Mullen SA, Marini C, Suls A, Mei D, Della Giustina E, Buti D, Arsov T, Damiano J, Lawrence K, De Jonghe P, Berkovic SF, Scheffer IE, Guerrini R. Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Arch Neurol. 2011;68(9):1152-5.

95. Specchio N, Marini C, Terracciano A, Mei D, Trivisano M, Sicca F, Fusco L, Cusmai R, Darra F, Bernardina BD, Bertini E, Guerrini R, Vigevano F..Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. Epilepsia. 2011;52(7):1251-7

96. Marini C, Mei D, Parmeggiani L, Norci V, Calado E, Ferrari A, Moreira A, Pisano T, Specchio N, Vigevano F, Battaglia D, Guerrini R.Protocadherin 19 mutations in girls with infantile-onset epilepsy. Neurology. 2010;75(7):646-53.

97. The genetics of Dravet syndrome. Marini C, Scheffer IE, Nabbout R, Suls A, De Jonghe P, Zara F, Guerrini R. Epilepsia. 2011;52 Suppl 2:24-9.

98. Conti V, Marini C, Gana S, Sudi J, Dobyns WB, Guerrini R. Corpus callosum agenesis, severe mental retardation, epilepsy, and dyskinetic quadriparesis due to a novel mutation in the homeodomain of ARX. Am J Med Genet A. 2011;155(4):892-7

99. Melani F, Mei D, Pisano T, Savasta S, Franzoni E, Ferrari AR, Marini C, Guerrini R. CDKL5 gene-related epileptic encephalopathy: electroclinical findings in the first year of life. Dev Med Child Neurol. 2011 Apr;53(4):354-60.

100. Conti V, Marini C, Mei D, Falchi M, Ferrari AR, Guerrini R.Contractions in the second polyA tract of ARX are rare, non-pathogenic polymorphisms. Am J Med Genet A. 2011 Jan;155A(1):164-7

101. Ragona F, Granata T, Dalla Bernardina B, Offredi F, Darra F, Morbi M, Battaglia D, Brazzo D, Cappelletti S, Chieffo D, De Giorgi I, Fontana E, Freri E, Marini C, Toraldo A, Specchio N, Veggiotti PA, Vigevano F, Guerrini R, Guzzetta , and Dravet C. Cognitive development in Dravet syndrome: a retrospective, multi center study of 26 patients. Epilepsia. 2011;52(2):386-92

102. Guerrini R, Cellini E, Mei D, Metitieri T, Petrelli C, Pucatti D, Marini C, and Zamponi N. Variable epilepsy phenotypes associated with a familialintragenic deletion of the SCN1A gene. Epilepsia. 2010;51(12):2474-7.

103. Muhle H, Steinich I, von Spiczak S, Franke A, Weber Y, Lerche H, Michael M, Heidemann S, Suls A, de Jonghe P, Marini C, Guerrini R, Scheffer IE, Berkovic SF, Stephani U, Siebert R, Sander T, Helbig I, Tönnies H. A duplication in 1q21.3 in a family with early-onset and childhood absence epilepsy. Epilepsia. 2010;51(12):2453-6

104. Marini C, Mei D, Parmeggiani L, Norci V, Calado E, Ferrari A, Moreira A, Pisano T, Specchio N, Vigevano F, Battaglia D, Guerrini R.Protocadherin 19 mutations in girls with infantile-onset epilepsy. Neurology. 2010;75(7):646-53.

105. Marini C and Mantegazza M. Sodium channelopathies and epilepsy: recent advances and new perspectives. Expert Rev Clin Pharmacol. 2010;3(3):371-84

106. Leventer RJ, Jansen A, Pilz DT, Stoodley N, Marini C, Dubeau F, Malone J, Mitchell LA, Mandelstam S, Scheffer IE, Berkovic SF, Andermann F, Andermann E, Guerrini R, Dobyns WB.Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain. 2010;133(Pt 5):1415-27.

107. Dibbens LM, Mullen S, Helbig I, Mefford HC, Bayly MA, Bellows S, Leu C, Trucks H, Obermeier T, Wittig M, Franke A, Caglayan H, Yapici Z; EPICURE Consortium, Sander T, Eichler EE, Scheffer IE, Mulley JC, Berkovic SF. Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Hum Mol Genet. 2009;18(19):3626-31.

108. Mei D, Marini C, Novara F, Dalla Bernardina B, Fontana E, Parrini E, ferrari AR, Murgia A, Zuffardi O, Guerrini R. Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia. 2010;51(4):647-54.

109. Sisodiya SM, Marini C. Genetics of antiepileptic drug resistance. Curr Opin Neurol. 2009;22(2):150-6.

110. Gambardella A, Marini C. Clinical spectrum of SCN1A mutations. Epilepsia. 2009 May;50 Suppl 5:20-3.

111. Marini C, Scheffer IE, Nabbout R, Mei D, Cox K, Dibbens LM, McMahon JM, Iona X, Carpintero RS, Elia M, Cilio MR, Specchio N, Giordano L, Striano P, Gennaro E, Cross JH, Kivity S, Neufeld MY, Afawi Z, Andermann E, Keene D, Dulac O, Zara F, Berkovic SF, Guerrini R, Mulley JC. SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia. 2009;50(7):1670-8.

112. Fedi M, Berkovic SF, Scheffer IE, O'Keefe G, Marini C, Mulligan R, Gong S, Tochon-Danguy H, Reutens DC. Reduced striatal D1 receptor binding in autosomal dominant nocturnal frontal lobe epilepsy. Neurology. 2008;71(11):795-8.

113. Hoda JC, Gu W, Friedli M, Phillips H, Bertrand S, Antonarakis SE, Goudie D, Roberts R, Scheffer I, Marini C, Patel J, Berkovic SF, Mulley JC, Steinlein OK, Bertrand DC. Human nocturnal frontal lobe epilepsy: pharmocogenomic profiles of pathogenic nAChR {beta}-subunit mutations outside the ion channel pore. Mol Pharmacol. 2008;74(2):379-91.

114. Mei D, Lewis R, Parrini E, Lazarou LP, Marini C, Pilz DT, Guerrini R. High frequency of genomic deletions and duplication in the LIS1 gene in lissencephaly: implications for molecular diagnosis. J Med Genet. 2008;45(6):355-61.

115. Fedi M, Berkovic SF, Macdonell RA, Curatolo JM, Marini C, Reutens DC. Intracortical Hyperexcitability in Humans with a GABAA Receptor Mutation. Cereb Cortex. 2008;18(3):664-9.

116. Marini C, Guerrini R. The role of the nicotinic acetylcholine receptors in sleep-related epilepsy. Biochem Pharmacol. Biochem Pharmacol. 2007;74(8):1308-14

117. Annesi F, Gambardella A, Michelucci R, Bianchi A, Marini C, Canevini MP, Capovilla G, Elia M, Buti D, Chifari R, Striano P, Rocca FE, Castellotti B, Cali F, Labate A, Lepiane E, Besana D, Sofia V, Tabiadon G, Tortorella G, Vigliano P, Vignoli A, Beccaria F, Annesi G, Striano S, Aguglia U, Guerrini R, Quattrone A. Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia. 2007;48(9):1686-90.

118. Sisodiya S, Cross JH, Blumcke I, Chadwick D, Craig J, Crino PB, Debenham P, Delanty N, Elmslie F, Gardiner M, Golden J, Goldstein D, Greenberg DA, Guerrini R, Hanna M, Harris J, Harrison P, Johnson MR, Kirov G, Kullman DM, Makoff A, Marini C, Nabbout R, Nashef L, Noebels JL, Ottman R, Pirmohamed M, Pitkanen A, Scheffer I, Shorvon S, Sills G, Wood N, Zuberi S. Genetics of epilepsy: epilepsy research foundation workshop report. Epileptic Disord. 2007;9(2):194-236.

119. Marini C, Mei D, Temudo T, Ferrari AR, Buti D, Dravet C, Dias AI, Moreira A, Calado E, Seri S, Neville B, Narbona J, Reid E, Michelucci R, Sicca F, Cross HJ, Guerrini R. Idiopathic Epilepsies with Seizures Precipitated by Fever and SCN1A Abnormalities. Epilepsia. 2007;48(9):1678-85.

120. Striano P, Mancardi MM, Biancheri R, Madia F, Gennaro E, Paravidino R, Beccaria F, Capovilla G, Dalla Bernardina B, Darra F, Elia M, Giordano L, Gobbi G, Granata T, Ragona F, Guerrini R, Marini C, Mei D, Longaretti F, Romeo A, Siri L, Specchio N, Vigevano F, Striano S, Tortora F, Rossi A, Minetti C, Dravet C, Gaggero R, Zara F. Brain MRI findings in severe myoclonic epilepsy in infancy and genotype-phenotype correlations. Epilepsia. 2007;48(6):1092-6.

121. Mei D, Parrini E, Pasqualetti M, Tortorella G, Franzoni E, Giussani U, Marini C, Migliarini S, Guerrini R. Multiplex ligation-dependent probe amplification detects DCX gene deletions in band heterotopia. Neurology. 2007;68(6):446-50.

122. Marini C, Mei D, Helen Cross J, Guerrini R. Mosaic SCN1A mutation in familial severe myoclonic epilepsy of infancy. Epilepsia. 2006;47(10):1737-40.

123. Gardella E, Tinuper P, Marini C, Guerrini R, Parrini E, Bisulli F, Liguori R, Montagna P, Lugaresi E.Autosomal dominant early-onset cortical myoclonus, photic-induced myoclonus, and epilepsy in a large pedigree. Epilepsia. 2006;47(10):1643-9.

124. Durner M, Gorroochurn P, Marini C, Guerrini R. Can we increase the likelihood of success for future association studies in epilepsy? Epilepsia. 2006;47(10):1617-21;

125. Fedi M, Berkovic SF, Marini C, Mulligan R, Tochon-Danguy H, Reutens DC. A GABAA receptor mutation causing generalized epilepsy reduces benzodiazepine receptor binding. Neuroimage. 2006;32(3):995-1000

126. Aridon P, Marini C, Di Resta C, Brilli E, De Fusco M, Politi F, Parrini E, Manfredi I, Pisano T, Pruna D, Curia G, Cianchetti C, Pasqualetti M, Becchetti A, Guerrini R, Casari G. Increased sensitivity of the neuronal nicotinic receptor alpha 2 subunit causes familial epilepsy with nocturnal wandering and ictal fear. Am J Hum Genet. 2006;79(2):342-50.

127. Guerrini R, Marini C. Genetic malformations of cortical development. Exp Brain Res. 2006;173(2):322-33.

128. Parrini E, Ramazzotti A, Dobyns WB, Mei D, Moro F, Veggiotti P, Marini C, Brilstra EH, Dalla Bernardina B, Goodwin L, Bodell A, Jones MC, Nangeroni M, Palmeri S, Said E, Sander JW, Striano P, Takahashi Y, Van Maldergem L, Leonardi G, Wright M, Walsh CA, Guerrini R. Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain. 2006;129(Pt 7):1892-906.

129. Derry CP, Davey M, Johns M, Kron K, Glencross D, Marini C, Scheffer IE, Berkovic SF. Distinguishing sleep disorders from seizures: diagnosing bumps in the night. Arch Neurol. 2006;63(5):705-9

130. Marini C, Parmeggiani L, Masi G, D'Arcangelo G, Guerrini R. Nonconvulsive status epilepticus precipitated by carbamazepine presenting as dissociative and affective disorders in adolescents. J Child Neurol. 2005;20(8):693-6.

131. Winawer MR, Marini C, Grinton BE, Rabinowitz D, Berkovic SF, Scheffer IE, Ottman R. Familial clustering of seizure types within the idiopathic generalized epilepsies. Neurology. 2005;65(4):523-8.

132. Martinelli Boneschi F, Aridon P, Zara F, Guerrini R, Marini C, De Fusco M, Comi G, Casari G. No evidence of ATP1A2 involvement in 12 multiplex Italian families with benign familial infantile seizures. Neurosci Lett. 2005;388(2):71-4.

133. Pisano T, Marini C, Brovedani P, Brizzolara D, Pruna D, Mei D, Moro F, Cianchetti C, Guerrini R. Abnormal phonologic processing in familial lateral temporal lobe epilepsy due to a new LGI1 mutation. Epilepsia. 2005;46(1):118-23.

134. Guerrini R, Parmeggiani L, Marini C, Brovedani P, Bonanni P. Autosomal dominant cortical myoclonus and epilepsy (ADCME) with linkage to chromosome 2p11.1-q12.2. Adv Neurol. 2005;95:273-9.

135. Taylor I, Marini C, Johnson MR, Turner S, Berkovic SF, Scheffer IE. Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap? Brain. 2004;127(Pt 8):1878-86.

136. Marini C, Scheffer IE, Crossland KM, Grinton BE, Phillips FL, McMahon JM, Turner SJ, Dean JT, Kivity S, Mazarib A, Neufeld MY, Korczyn AD, Harkin LA, Dibbens LM, Wallace RH, Mulley JC, Berkovic SF. Genetic architecture of idiopathic generalized epilepsy: clinical genetic analysis of 55 multiplex families. Epilepsia. 2004;45(5):467-78.

137. Berkovic SF, Heron SE, Giordano L, Marini C, Guerrini R, Kaplan RE, Gambardella A, Steinlein OK, Grinton BE, Dean JT, Bordo L, Hodgson BL, Yamamoto T, Mulley JC, Zara F, Scheffer IE. Benign familial neonatal-infantile seizures: characterization of a new sodium channelopathy. Ann Neurol. 2004;55(4):550-7.

138. Bonanni P, Malcarne M, Moro F, Veggiotti P, Buti D, Ferrari AR, Parrini E, Mei D, Volzone A, Zara F, Heron SE, Bordo L, Marini C, Guerrini R. Generalized epilepsy with febrile seizures plus (GEFS+): clinical spectrum in seven Italian families unrelated to SCN1A, SCN1B, and GABRG2 gene mutations. Epilepsia. 2004;45(2):149-58.

139. Marini C, Harvey AS, Pelekanos JT, Berkovic SF. Epilepsy in offspring of whom both parents have idiopathic generalized epilepsy: biparental inheritance. Epilepsia. 2003;44(9):1250-4.

140. Guerrini R, Casari G, Marini C. The genetic and molecular basis of epilepsy. Trends Mol Med. 2003;9(7):300-6.

141. Yenjun S, Harvey AS, Marini C, Newton MR, King MA, Berkovic SF. EEG in adult-onset idiopathic generalized epilepsy. Epilepsia. 2003;44(2):252-6.

142. Marini C, King MA, Archer JS, Newton MR, Berkovic SF. Idiopathic generalised epilepsy of adult onset: clinical syndromes and genetics. J Neurol Neurosurg Psychiatry. 2003;74:192-6.

143. Marini C, Harkin LA, Wallace RH, Mulley JC, Scheffer IE, Berkovic SF. Childhood absence epilepsy and febrile seizures: a family with a GABA(A) receptor mutation. Brain. 2003;126(Pt 1):230-40.

144. Bisulli F, Tinuper P, Marini C, Avoni P, Carraro G, Nobile C. Partial epilepsy with prominent auditory symptoms not linked to chromosome 10q. Epileptic Disord. 2002;4(3):183-7.

145. Bowser DN, Wagner DA, Czajkowski C, Cromer BA, Parker MW, Wallace RH, Harkin LA, Mulley JC, Marini C, Berkovic SF, Williams DA, Jones MV, Petrou S. Altered kinetics and benzodiazepine sensitivity of a GABAA receptor subunit mutation [gamma 2(R43Q)] found in human epilepsy. Proc Natl Acad Sci U S A. 2002;99(23):15170-5.

146. Tinuper P, Bisulli F, Cerullo A, Carcangiu R, Marini C, Pierangeli G, Cortelli P. Ictal bradycardia in partial epileptic seizures: Autonomic investigation in three cases and literature review. Brain. 2001;124(Pt 12):2361-71.

147. Wallace RH, Marini C, Petrou S, Harkin LA, Bowser DN, Panchal RG, Williams DA, Sutherland GR, Mulley JC, Scheffer IE, Berkovic SF. Mutant GABA(A) receptor gamma2-subunit in childhood absence epilepsy and febrile seizures. Nat Genet. 2001;28(1):49-52.

148. Phillips HA, Marini C, Scheffer IE, Sutherland GR, Mulley JC, Berkovic SF. A de novo mutation in sporadic nocturnal frontal lobe epilepsy. Ann Neurol. 2000;48(2):264-7.

149. Valenti MP, Tinuper P, Cerullo A, Carcangiu R, Marini C. Reading epilepsy in a patient with previous idiopathic focal epilepsy with centrotemporal spikes. Epileptic Disord. 1999;1(3):167-71.

150. Cerullo A, Marini C, Carcangiu R, Baruzzi A, Tinuper P. Clinical and video-polygraphic features of epileptic spasms in adults with cortical migration disorder. Epileptic Disord. 1999;1(1):27-33.

151. Cerullo A, Marini C, Cevoli S, Carelli V, Montagna P, Tinuper P. Colpocephaly in two siblings: further evidence of a genetic transmission. Dev Med Child Neurol. 2000;42:280-2.

152. Cerullo A, Tinuper P, Provini F, Contin M, Rosati A, Marini C, Cortelli P. Autonomic and hormonal ictal changes in gelastic seizures from hypothalamic hamartomas. Electroencephalogr Clin Neurophysiol. 1998;107(5):317-22.

153. Tinuper P, Cerullo A, Marini C, Avoni P, Rosati A, Riva R, Baruzzi A, Lugaresi E. Epileptic drop attacks in partial epilepsy: clinical features, evolution, and prognosis. J Neurol Neurosurg Psychiatry. 1998;64(2):231-7.

154. Tinuper P, Provini F, Marini C, Cerullo A, Plazzi G, Avoni P, Baruzzi A. Partial epilepsy of long duration: changing semiology with age. Epilepsia. 1996; 37:162-4.

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 02/08/2023_____ _____

_____

(per esteso e leggibile)

*La dichiarazione é sottoscritta dall'interessato in presenza del dipendente addetto, **oppure sottoscritta e inviata assieme alla fotocopia del documento di identità via fax, a mezzo posta ordinaria o elettronica o tramite un incaricato** (art. 38 D.P.R. 445/2000).