Curriculum Vitae Europass

Informazioni personali

Nome(i) / Cognome(i)	**Diego Martinelli**
Telefono(i)	
Fax	
E-mail	
Cittadinanza	Italiana
Codice fiscale	
Data di nascita	

Occupazione desiderata/Settore professionale

Esperienza professionale

Date	Anno 2016-2021
Lavoro o posizione ricoperti	Dirigente Medico I livello
Principali attività e responsabilità	Medico Strutturato UOC Patologia Metabolica
Nome e indirizzo del datore di lavoro	Ospedale Pediatrico Bambino Gesù, Piazza S. Onofrio 4, 00165, Roma
Tipo di attività o settore	Pediatria – Malattie metaboliche

Istruzione e formazione

Date	Anno 2014 -2016
Titolo della qualifica rilasciata	Post-Doctoral Visiting Fellow
Nome e tipo di organizzazione Erogatrice dell'istruzione e formazione	National Institute of Child Health and Human Development, National Institutes of Health

Date	Anno 2012
Titolo della qualifica rilasciata	Dottorato di Ricerca in Neuroscienze dello Sviluppo
Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione	Facoltà di Medicina, Università Cattolica del Sacro Cuore, Roma, Italia

Date	Anno 2009
Titolo della qualifica rilasciata	Specializzazione in Neuropsichiatria Infantile, voto 50/50 e lode
Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione	Facoltà di Medicina, Università Cattolica del Sacro Cuore, Roma, Italia
	Facoltà di Medicina, Università Cattolica del Sacro Cuore, Roma, Italia
Date	Anno 2003
Titolo della qualifica rilasciata	Laurea in Medicina e Chirurgia, voto 110/110 e lode
Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione	Facoltà di Medicina, Università Cattolica del Sacro Cuore, Roma, Italia
	Facoltà di Medicina, Università Cattolica del Sacro Cuore, Roma, Italia

Capacità e competenze personali

Madrelingua(e) **Italiano**

Altra(e) lingua(e) **Inglese – Francese – Portoghese**

Autovalutazione	**Comprensione**				**Parlato**				**Scritto**	
	Ascolto		Lettura		Interazione orale		Produzione orale			
Livello europeo (*)	I	si	I	si	I	si	I	si	I	si
	F	si	F	si	F	si	F	si	F	si
	P	si	P	si	P	si	P	si	P	si

TRIAL CLINICI

Start Date (YYYY)	**Phase** (I,II,III,IV)	**Indicazione**
2019	Retrospective	Retrospective study of Vatiquinone efficacy and safety for the treatment of patients with pontocerebellar hypoplasia type 6 (rars2 syndrome). Sub-investigator
2019	III	A Randomised, Controlled , Open-Label Parallel Arm Study of the Safety, Pharmacokinetics and Ammonia Contrai of RAVICTl ® (Glycerol Phenylbutyrate [GPB]) Oral Liquid and Sodium Phenylbutyrate (NaPBA) in Phenylbutyrate Treatment Naive Patients with Urea Cycle Disorders (UCDs). HPN-100-021, Principal investigator
2013-2016	II	EPI-743 in Cobalamin C defect: effects on Visual and Neurological Impairment (NCT01793090). Principal investigator
2015	II/III	Studio in aperto, multinazionale, multicentrico di fase 2/3 per valutare l'efficacia e la sicurezza di ALXN1101 nei neonati con deficit del cofattore molibdeno (MOCD) di tipo A. protocollo ALXN1101-MCD-202.Sub-investigator
2013-ongoing	II	An open-label study of EPI-743 in Leigh Syndrome (OPBGC&RS 2011-004_CA2012). Sub-investigator
2011-2012	II	An open-label study of EPI-743 in Leigh Syndrome (EPI-2011-004) EudraCT Number: 2012-001294-84.Sub-investigator
2007-2008	II	Investigation of Sodium Valproate in Boys with Fragile X Syndrome and ADHD. NPL-005-1-FMR.Sub-investigator
2006-2008	II	An Open-Label Study of the Efficacy of Atomoxetine hydrocloride on Quality of Life of Children and Adolescents with Attention-Deficit/Hyperactivity Disorder with or without comorbid condition. Sub- investigator

Attività di ricerca

- La mia attività di ricerca è soprattutto rivolta all'elucidazione dei meccanismi patogenetici e all'identificazione di nuovi geni malattia e biomarcatori negli errori congeniti del metabolismo, in particolare malattie mitocondriali, disturbi del metabolismo del rame, malattie neurometaboliche, neurmuscolari e neurocutanee, difetti del ciclo dell'urea e acidurie organiche. Ho diretto numerosi progetti di medicina traslazionale, focalizzati sulla caratterizzazione clinica e biochimica di pazienti con errori congeniti del metabolismo e sullo sviluppo di nuovi protocolli di valutazione, linee guida e nuovi approcci terapeutici nelle malattie metaboliche e nei disordini del neurosviluppo. Tale attività di ricerca è stata svolta in collaborazione con molti centri in Europa (Francia, Svizzera, Paesi Bassi, Germania, Regno Unito) negli Stati Uniti e Australia.

Ulteriori informazioni

- Autore di 132 pubblicazioni in extenso su riviste internazionali
- Autore di numerosi Capitoli su libri/testi italiani ed esteri
- Invitato come relatore/moderatore a numerosi congressi Nazionali ed Internazionali
- Membro del SSIEM (European Society for the Study of Inborn Errors of Metabolism)
- Vicepresidente della SIMMESN (Società italiana per lo studio delle Malattie Metaboliche Ereditarie e lo Screening Neonatale.)
- Membro del gruppo di lavoro per le linee guida dell' E-HOD (European Network and Registry for Homocystinurias and Methylation Defects)
- Membro del gruppo di lavoro per le linee guida dell'E-IMD (European Network and Registry for Intoxication Type Metabolic diseases)
- Membro del Board del Consorzio U-IMD (Unified registry for inborn error of metabolism)
- Membro dello Steering committee Biolectron Technology Corporation –OPBG
- Membro del METABERN-Subnetwork Mitochondrial Diseases –corresponsabile line guida
- Membro del METABERN- Co-cordinator Subnetwork Carbohydrate, fatty acid oxidation and ketone bodies disorders
- Founder Member of the DC and NIH chapter of ISSNAF (Italian Scientists and Scholars in North America Foundation)
- Former Representative of Italians at NIH scientific group
- SSIEM AWARD 2012 for the best late breaking new "MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy".
- Primo premio al Simposio Nazionale della società Italiana di malattia metaboliche 2012 (SIMMESN – SIMGePeD) per la presentazione orale "Sindrome MEDNIK: un nuodo difetto del metabolismo del rame trattabile con zinco acetato".
- Prize for best poster presentation - International Workshop "HUMAN DISORDERS OF COPPER METABOLISM: RECENT ADVANCES AND MAIN CHALLENGES". Johns Hopkins University, Baltimore, Maryland. April 8-9, 2013.
- NICHD Collaboration Award 2014. National Institutes of Child Health and Human Development, National Institutes of Health.
- Oral presenter at the Twelfth Annual NICHDFellows Retreat
- Vincitore nel 2019 di Finanzimento di 110.00 euro da parte della Bioelectron inc per lo studio retrospettivo: "Retrospective study of Vatiquinone efficacy and safety for the treatment of patients with pontocerebellar hypoplasia type 6 (rars2 syndrome)"
- Vincitore come PI della Ricerca Finalizzata 2018 –sezione Giovani Ricercatori con ricerca dal Titolo "Clinical-instrumental definition of the phenotypic spectrum, response to treatment and natural history in Pearson and Kearns-Sayre syndrome" - Finanziamento: 243.980 euro.
- Vincitore come PI del Grant Telethon 2020 con ricerca dal titolo: "Il ruolo dell'infiammazione nelle malattie legate all'integrità del DNA mitocondriale: nuovi potenziali biomarcatori e bersagli terapeutici". Finanziamento: 153.780 euro
- H Index: 31 (SCOPUS, WEB of SCIENCE)
- Scopus Author Id: 36163327600; ORCID ID: orcid.org/0000-0002-9324-2871 RESEARCH ID: K-9723-2016.

- Idoneità a Professore di seconda fascia in Genetica Medica, abilitazione Scientifica nazionale 2016-2018 quarto quadrimestre
- In corso valutazione per idoneità a Professore di seconda fascia in Pediatria e Neuropsichiatria Infantile, abilitazione Scientifica nazionale 2021
- Patente: WIPO Patent Application WO/2013/006736, Kind Code:A1, titolo: "TREATMENT OF LEIGH SYNDROME AND LEIGH-LIKE SYNDROME, INCLUDING COMPLICATIONS OF SUCLA2 MUTATIONS, WITH TOCOTRIENOL QUINONE". Autori Guy Miller, Carlo Dionisi-Vici, Enrico Silvio Bertini, Diego Martinelli. 10/01/2013
- Patente: United States Patent Application 20150057363 Kind Code: A1, titolo "TREATMENT OF METHYLMALONIC ACIDURIA, ISOVALERIC ACIDURIA, AND OTHER ORGANIC ACIDURIAS WITH TOCOTRIENOL QUINONES". Autori Guy Miller, Carlo Dionisi-Vici, Enrco Silvio Bertini, Diego Martinelli. 26/02/2015
- Docenza nel Corso Alta Formazione " Disturbi del movimento nel bambino: nuove acquisizioni scientifiche e strategie diagnostiche", responsabile scientifico Prof. V. Leuzzi, DIPARTIMENTO DI PEDIATRIA E NEUROPSICHIATRIA INFANTILE " SAPIENZA" Università di Roma, Anno 2016-2017
- Docenza nel Corso Alta Formazione " Neurologia infantile: nuove acquisizioni scientifiche, nuove malattie, nuove strategie diagnostiche, nuovi approcci terapeutici", responsabile scientifico Prof. V. Leuzzi, DIPARTIMENTO DI PEDIATRIA E NEUROPSICHIATRIA INFANTILE " SAPIENZA" Università di Roma, Anno 2016-2017
- Docenza Master - La neuroriabilitazione e le neuroscienze dell'età evolutiva - consorzio Humanitas/Lumsa/2019-2020 con lezione dal titolo: " Il bambino con malattia neurodegenerativa"
- Docenza nell'ambito del corso di formazione "Novità sulla Patogenesi e il trattamento delle Malattie Metaboliche Ereditarie" organizzato dalla SIMMESN (societa' italiana malattie metaboliche ereditarie e screening neonatale).
- Docente nell'ambito del Master sulle Malattie Metaboliche Ereditarie e Screening Neonatale – Direttore Prof. Andrea Pession organizzato dalla Universita' di Bologna in collaborazione con la SIMMESN

Pubblicazioni

1) Baban, A., Lodato, V., Parlapiano, G.,Di Mambro C, Adorisio R, Bertini E, Dionisi-Vici C, .Drago, F., Martinelli, D.Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. Biomolecules, 2021, 11(11), 1578.
2) Brennenstuhl, H., Nashawi, M., Schröter, J., ...Kozich, V., Scarpa, M.Phenotypic diversity, disease progression, and pathogenicity of MVK missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44(5), pp. 1272–1287.
3) Laugwitz L, Seibt A, Herebian D, Peralta S, Kienzle I, Buchert R, Falb R, Gauck D, Müller A, Grimmel M, Beck-Woedel S, Kern J, Daliri K, Katibeh P, Danhauser K, Leiz S, Alesi V, Baertling F, Vasco G, Steinfeld R, Wagner M, Caglayan AO, Gumus H, Burmeister M, Mayatepek E, Martinelli D, Tamhankar PM, Tamhankar V, Joset P, Steindl K, Rauch A, Bonnen PE, Froukh T, Groeschel S, Krägeloh-Mann I, Haack TB, Distelmaier F. Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. J Med Genet 2021 Oct 16;jmedgenet-2021-107729.
4) Ardissone A, Bruno C, Diodato D, Donati A, Ghezzi D, Lamantea E, Lamperti C, Mancuso M, Martinelli D, Primiano G, Procopio E, Rubegni A, Santorelli F, Schiaffino MC, Servidei S, Tubili F, Bertini E, Moroni I. Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases Orphanet J Rare Dis. 2021 Oct 9;16(1):413.
5) Battaglia DI, Gambardella ML, Veltri S, Contaldo I, Chillemi G, Veredice C, Quintiliani M, Leoni C, Onesimo R, Verdolotti T, Radio FC, Martinelli D, Trivisano M, Specchio N, Dravet C, Tartaglia M, Zampino G. Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations Genes (Basel). 2021 Aug 26;12(9):1316.

6) Ferreira CR, Martinelli D, Blau N. Clinical and biochemical footprints of inherited metabolic diseases. VI. Metabolic dermatoses Mol Genet Metab. 2021 Sep-Oct;134(1-2):87-95.

7) Ticci C, Orsucci D, Ardissone A, Bello L, Bertini E, Bonato I, Bruno C, Carelli V, Diodato D, Doccini S, Donati MA, Dosi C, Filosto M, Fiorillo C, La Morgia C, Lamperti C, Marchet S, Martinelli D, Minetti C, Moggio M, Mongini TE, Montano V, Moroni I, Musumeci O, Pancheri E, Pegoraro E, Primiano G, Procopio E, Rubegni A, Scalise R, Sciacco M, Servidei S, Siciliano G, Simoncini C, Tolomeo D, Tonin P, Toscano A, Tubili F, Mancuso M, Battini R, Santorelli FM. Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. J Clin Med 2021 May 12;10(10):2063.

8) Olivieri G, Martinelli D, Longo D, Grimaldi C, Liccardo D, Di Meo I, Pietrobattista A, Sidorina A, Semeraro M, Dionisi-Vici C. Ethylmalonic encephalopathy and liver transplantation: long-term outcome of the first treated patient. Orphanet J Rare Dis. 2021 May 19;16(1):229. doi: 10.1186/s13023-021-01867-5.

9) Di Mambro C, Martinelli D, Tamborrino PP, Drago F.Involvement of the cardiac conduction system in Kearns-Sayre syndrome is progressive: Authors' reply. Europace. 2021 Jun 7;23(6):980. doi: 10.1093/europace/euab056

10) Maines E, Franceschi R, Martinelli D, Soli F, Lepri FR, Piccoli G, Soffiati M. Hypoglycemia due to PI3K/AKT/mTOR signaling pathway defects: two novel cases and review of the literature. Hormones (Athens). 2021 Apr 20. doi: 10.1007/s42000-021-00287-1.

11) Rossi A, Hoogeveen IJ, Lubout CMA, de Boer F, Fokkert-Wilts MJ, Rodenburg IL, van Dam E, Grünert SC, Martinelli D, Scarpa M; CONNECT MetabERN collaboration group, Dekker H, Te Boekhorst ST, van Spronsen FJ, Derks TGJ. A generic emergency protocol for patients with inborn errors of metabolism causing fasting intolerance: a retrospective, single-center study and the generation of www.emergencyprotocol.net. J Inherit Metab Dis. 2021 Apr 12. doi: 10.1002/jimd.12386.

12) Torraco A, Nasca A, Verrigni D, Pennisi A, Zaki MS, Olivieri G, Assouline Z, Martinelli D, Maroofian R, Rizza T, Di Nottia M, Invernizzi F, Lamantea E, Longo D, Houlden H, Prokisch H, Rötig A, Dionisi-Vici C, Bertini E, Ghezzi D, Carrozzo R, Diodato D. Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Hum Mutat. 2021 Mar 14.

13) Diamanti A, Calvitti G, Martinelli D, Santariga E, Capriati T, Bolasco G, Iughetti L, Pujia A, Knafelz D, Maggiore G. Etiology and Management of Pediatric Intestinal Failure: Focus on the Non-Digestive Causes. Nutrients. 2021 Feb 27;13(3):786

14) Diociaiuti A, Martinelli D, Nicita F, Cesario C, Pisaneschi E, Macchiaiolo M, Rossi S, Condorelli AG, Zambruno G, El Hachem M. Two Italian Patients with ELOVL4-Related Neuro-Ichthyosis: Expanding the Genotypic and Phenotypic Spectrum and Ultrastructural Characterization. Genes (Basel). 2021 Feb 26;12(3):343.

15) Pettinato F, Mostile G, Battini R, Martinelli D, Madeo A, Biamino E, Frattini D, Garozzo D, Gasperini S, Parini R, Sirchia F, Sortino G, Sturiale L, Matthijs G, Morrone A, Di Rocco M, Rizzo R, Jaeken J, Fiumara A, Barone R. Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). Cerebellum. 2021 Feb 22

16) Masnada S, Martinelli D, Correa-Vela M, Agolini E, Baide-Mairena H, Marcé-Grau A, Parazzini C, Veggiotti P, Perez-Duenas B, Tonduti D. PRKRA-Related Disorders: Bilateral Striatal Degeneration in Addition to DYT16 Spectrum. Mov Disord. 2021 Apr;36(4):1038-1040. doi: 10.1002/mds.28492.

17) Opladen T, Gleich F, Kozich V, Scarpa M, Martinelli D, Schaefer F, Jeltsch K, Juliá-Palacios N, García-Cazorla Á, Dionisi-Vici C, Kölker S. U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet J Rare Dis. 2021 Feb 18;16(1):95. doi: 10.1186/s13023-021-01726-3.

18) Forny P, Hörster F, Ballhausen D, Chakrapani A, Chapman KA, Dionisi-Vici C, Dixon M, Grünert SC, Grunewald S, Haliloglu G, Hochuli M, Honzik T, Karall D, Martinelli D, Molema F, Sass JO, Scholl-Bürgi S, Tal G, Williams M, Huemer M, Baumgartner MR. Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: First revision. J Inherit Metab Dis. 2021 Feb 17. doi: 10.1002/jimd.12370.

19) Di Mambro C, Tamborrino PP, Silvetti MS, Yammine ML, Marcolin C, Righi D, Baban A, Martinelli D, Vici CD, Drago F. Progressive involvement of cardiac conduction system in paediatric patients with Kearns-Sayre syndrome: how to predict occurrence of complete heart block and sudden cardiac death? Europace. 2020 Dec 18:euaa335.

20) Polovitskaya MM, Barbini C, Martinelli D, Harms FL, Cole FS, Calligari P, Bocchinfuso G, Stella L, Ciolfi A, Niceta M, Rizza T, Shinawi M, Sisco K, Johannsen J, Denecke J, Carrozzo R, Wegner DJ, Kutsche K, Tartaglia M, Jentsch TJ. A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl-/H+-Exchanger, Causes Early-Onset Neurodegeneration. Am J Hum Genet. 2020 Dec 3;107(6):1062-1077. doi: 10.1016/j.ajhg.2020.11.004.

21) Pasquini L, Guarnera A, Rossi-Espagnet MC, Napolitano A, Diodato D, Martinelli D, Longo D. Reply to: "Is the spinal cord truly affected in half of the patients with Kearns-Sayre syndrome?" and "Spinal cord and heart involvement in Kearns Sayre Syndrome: which link?". Neuroradiology. 2021 Jan;63(1):9-10. doi: 10.1007/s00234-020-02593-8. Epub 2020 Nov 3.

22) Pasquini L, Guarnera A, Rossi-Espagnet MC, Napolitano A, Martinelli D, Deodato F, Diodato D, Carrozzo R, Dionisi-Vici C, Longo D. Correction to: Spinal cord involvement in Kearns-Sayre syndrome: a neuroimaging study. Neuroradiology. 2020 Dec;62(12):1725. doi: 10.1007/s00234-020-02573-y. Erratum for: Neuroradiology. 2020 Oct;62(10):1315-1321.

23) Molema F, Martinelli D, Hörster F, Kölker S, Tangeraas T, de Koning B, Dionisi-Vici C, Williams M; additional individual contributors of MetabERN. Liver and/or kidney transplantation in amino and organic acid-related inborn errors of metabolism: An overview on European data. J Inherit Metab Dis. 2020 Sep 30.

24) Bösch F, Landolt MA, Baumgartner MR, Zeltner N, Kölker S, Gleich F, Burlina A, Cazzorla C, Packman W, V D Schwartz I, Vieira Neto E, Ribeiro MG, Martinelli D, Olivieri G, Huemer M. Health-related quality of life in paediatric patients with intoxication-type inborn errors of metabolism: Analysis of an international data set. J Inherit Metab Dis. 2021 Jan;44(1):215-225.

25) Casetta B, Malvagia S, Funghini S, Martinelli D, Dionisi-Vici C, Barone R, Fiumara A, Donati MA, Guerrini R, la Marca G. A new strategy implementing mass spectrometry in the diagnosis of congenital disorders of N-glycosylation (CDG). Clin Chem Lab Med. 2020 Aug 10;59(1):165-171

26) Maines E, Catesini G, Boenzi S, Mosca A, Candusso M, Dello Strologo L, Martinelli D, Maiorana A, Liguori A, Olivieri G, Taurisano R, Piemonte F, Rizzo C, Spada M, Dionisi-Vici C. Plasma methylcitric acid and its correlations with other disease biomarkers: The impact in the follow up of patients with propionic and methylmalonic acidemia. J Inherit Metab Dis. 2020 Nov;43(6):1173-1185.

27) Marti-Sanchez L, Baide-Mairena H, Marcé-Grau A, Pons R, Skouma A, López-Laso E, Sigatullina M, Rizzo C, Semeraro M, Martinelli D, Carrozzo R, Dionisi-Vici C, González-Gutiérrez-Solana L, Correa-Vela M, Ortigoza-Escobar JD, Sánchez-Montañez Á, Vazquez É, Delgado I, Aguilera-Albesa S, Yoldi ME, Ribes A, Tort F, Pollini L, Galosi S, Leuzzi V, Tolve M, Pérez-Gay L, Aldamiz-Echevarría L, Del Toro M, Arranz A, Roelens F, Urreizti R, Artuch R, Macaya A, Pérez-Dueñas B. Delineating the neurological phenotype in children with defects in the ECHS1 or HIBCH gene. J Inherit Metab Dis. 2021 Mar;44(2):401-414.

28) Martinelli D, Schiff M, Semeraro M, Agolini E, Novelli A, Dionisi-Vici C. CUGC for lysinuric protein intolerance (LPI). Eur J Hum Genet. 2020 Aug;28(8):1129-1134.

29) Martinelli D, Fiermonte G, Häberle J, Boenzi S, Goffredo BM, Travaglini L, Agolini E, Porcelli V, Dionisi-Vici C. CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. Eur J Hum Genet. 2020 Jul;28(7):982-987.

30) Summa S, Schirinzi T, Favetta M, Romano A, Minosse S, Diodato D, Olivieri G, Martinelli D, Sancesario A, Zanni G, Castelli E, Bertini E, Petrarca M, Vasco G. A wearable video-oculography based evaluation of saccades and respective clinical correlates in patients with early onset ataxia. J Neurosci Methods. 2020 May 15;338:108697. doi: 10.1016/j.jneumeth.2020.108697. Epub 2020 Mar 20. PMID: 32205159.

31) Hengel H, Bosso-Lefèvre C, Grady G, Szenker-Ravi E, Li H, Pierce S, Lebigot É, Tan TT, Eio MY, Narayanan G, Utami KH, Yau M, Handal N, Deigendesch W, Keimer R, Marzouqa HM, Gunay-Aygun M, Muriello MJ, Verhelst H, Weckhuysen S, Mahida S, Naidu S, Thomas TG, Lim JY, Tan ES, Haye D, Willemsen MAAP,

Oegema R, Mitchell WG, Pierson TM, Andrews MV, Willing MC, Rodan LH, Barakat TS, van Slegtenhorst M, Gavrilova RH, Martinelli D, Gilboa T, Tamim AM, Hashem MO, AlSayed MD, Abdulrahim MM, Al-Owain M, Awaji A, Mahmoud AAH, Faqeih EA, Asmari AA, Algain SM, Jad LA, Aldhalaan HM, Helbig I, Koolen DA, Riess A, Kraegeloh-Mann I, Bauer P, Gulsuner S, Stamberger H, Ng AYJ, Tang S, Tohari S, Keren B, Schultz-Rogers LE, Klee EW, Barresi S, Tartaglia M, Mor-Shaked H, Maddirevula S, Begtrup A, Telegrafi A, Pfundt R, Schüle R, Ciruna B, Bonnard C, Pouladi MA, Stewart JC, Claridge-Chang A, Lefeber DJ, Alkuraya FS, Mathuru AS, Venkatesh B, Barycki JJ, Simpson MA, Jamuar SS, Schöls L, Reversade B. Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nat Commun. 2020 Jan 30;11(1):595. doi: 10.1038/s41467-020-14360-7. PubMed PMID: 32001716; PubMed Central PMCID: PMC6992768.

32) Ticci C, Sicca F, Ardissone A, Bertini E, Carelli V, Diodato D, Di Vito L, Filosto La Morgia C, Lamperti C, Martinelli D, Moroni I, Musumeci O, Orsucci D, Pancheri E, Peverelli L, Primiano G, Rubegni A, Servidei S, Siciliano G, Simoncini C, Tonin P, Toscano A, Mancuso M, Santorelli FM. Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics. 2020 Jan 3. doi: 10.1007/s10048-019-00601-5. [Epub ahead of print] PubMed PMID: 31900734.

33) Agolini E, Cherchi C, Bellacchio E, Martinelli D, Cocciadiferro D, Cutrera R, Chiarini Testa MB, Barone C, Bianca S, Novelli A. Expanding the clinical and molecular spectrum of Lethal Congenital Contracture Syndrome 8 associated with biallelic variants of ADCY6. Clin Genet. 2019 Dec 17. doi: 10.1111/cge.13691. [Epub ahead of print] PubMed PMID: 31846058.

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Data 15 novembre 2021

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".

Il/La Sottoscritto/a, ai sensi degli artt. 46 e 47 D.P.R. n. 445/2000, consapevole delle sanzioni penali previste dall'art. 76 D.P.R. n. 445/2000 nel caso di mendaci dichiarazioni, falsità negli atti, uso o esibizione di atti falsi o contenenti dati non più corrispondenti a verità, dichiara che quanto sopra riportato corrisponde a verità.
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