CURRICULUM VITAE

FORMATO EUROPEO PER IL CURRICULUM VITAE

INFORMAZIONI PERSONALI

Nome e Cognome

Annalisa Vetro

Incarico attuale

Dirigente biologo, responsabile UOS Laboratorio di Genetica, UOC Genetica Medica AO Ospedali Riuniti Villa Sofia – Cervello, Palermo

ISTRUZIONE E FORMAZIONE

20/07/2005
Laurea in Scienze Biologiche (vecchio ordinamento)
Università degli Studi di Palermo, Palermo, voto 110/100 *cum laude*

26/10/2009
Diploma di Specializzazione in Genetica Medica
Università degli Studi di Pavia, Pavia, voto 50/50 *cum laude*,

08/02/2013
Dottorato di ricerca in Patologia Umana e Genetica Medica (XXV ciclo)
Università degli Studi di Pavia, Pavia

ESPERIENZA LAVORATIVA

Dal 01/07/2023 ad oggi: Dirigente Biologo a tempo indeterminato, UOC Genetica Medica, Azienda Ospedaliera Ospedali Riuniti Villa Sofia Cervello. Da settembre 2023 responsabile di UOS presso la stessa UOC.

Dal 01/04/2017 al 30/06/2023: Dirigente Biologo a tempo indeterminato, disciplina Laboratorio di Genetica Medica, Azienda Ospedaliero Universitaria Meyer IRCCS, Firenze.

Dal 29/12/2015 al 31/03/2017: Ricercatore a tempo determinato ai sensi dell'art. 24, comma 3, lettera a) della legge 240/10, Dipartimento di Medicina Molecolare, Università degli Studi di Pavia, Pavia.

Dal 19/03/2013 al 19/03/2015: Titolare di incarichi a termine per collaborare allo svolgimento di un progetto di ricerca, presso la Fondazione IRCCS Policlinico San Matteo, Pavia, L.S.R. Area Biotecnologie.

Dal 01/12/2012 al 19/03/2013: Titolare di Borsa di Studio per attività di ricerca Dipartimento di Medicina Molecolare, Università degli Studi di Pavia, Pavia.

Dal 27/10/2009 al 27/10/2012: Titolare di Borsa di Studio per lo svolgimento del Dottorato di Ricerca in Patologia e Genetica Medica (ciclo XXV) presso il Dipartimento di Patologia Umana ed Ereditaria Università degli Studi di Pavia, Pavia.

Dal 15/06/2006 al 01/11/2009: Titolare di Contratti di prestazione d'opera nella forma di collaborazione coordinata e continuativa per lo svolgimento di attività di ricerca presso il Dipartimento di Patologia Umana ed Ereditaria, Laboratorio di Citogenetica Prenatale e Molecolare Università degli Studi di Pavia, Pavia

MADRELINGUA

Italiano

ALTRE LINGUE

[Inglese]

- Capacità di lettura [eccellente]
- Capacità di scrittura [eccellente]
- Capacità di espressione orale [buono]

CAPACITÀ E COMPETENZE TECNICHE

Con computer, attrezzature specifiche, macchinari, ecc.

Citogenetica convenzionale e molecolare; biologia molecolare; bionformatica; genetica medica; analisi ed interpretazione dati.

ALTRO (PARTECIPAZIONE A CONVEGNI, SEMINARI, PUBBLICAZIONI, COLLABORAZIONI A RIVISTE,ECC. ED OGNI ALTRA INFORMAZIONE CHE IL COMPILANTE RITIENE DI DOVER PUBBLICARE)

Abilitazione alla professione di Biologo conseguita presso l'Università degli Studi di Palermo, Palermo, nella II sessione, anno 2005

Iscrizione albo Collegio/Ordine Professionale dei Biologi (regione Sicilia) dal 29/01/2015 n. posizione n. Sic_A4184

Abilitazione scientifica nazionale (art. 16 legge 240/2010) per il settore concorsuale 06/A1 conseguita in data 31/03/2017 per le funzioni di professore di seconda fascia;

Membro dell'Editorial Board della rivista scientifica European Journal of Human Genetics da Novembre 2021 ad oggi

PUBBLICAZIONI

Coautore di 85 articoli scientifici su riviste scientifiche indicizzate internazionali soggette a peer-review;
H-index (Scopus): 24
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Pubblicazioni degli ultimi 5 anni:

1. Molecular and Phenotypic Characterization of the *RORB*-Related Disorder. Gokce-Samar Z*, Vetro A*, De Bellescize J*, Pisano T, Monteiro L, Penaud N, Korff CM, Fluss J, Marini C, Cesaroni E, Alvarez BM, Sanlaville D, Chatron N, Arzimanoglou AA, Labalme A, Cuddapah VA, Ruggiero SM, Lecoquierre F, Nicolas G, Marie GA, Lebas A, Testard HO, Helbig KL, Ruiz A, Ngoh A, Kurian MA, Reid K, Spaull R, Joset P, Ramantani G, Steindl K, Krenn M, Gerstl L, Vieker S, Craiu D, Pendziwiat M, Haldeman-Englert C, Kanivets I, Romanova I, Rajan DS, Rosenfeld JA, Au M, Grand K, Graham M Jr, Isapof A, Villeneuve N, Smol T, Caumes R, Zacher P, Neuser S, Tinschert S, Platzer K, Bartolomaeus T, Mohnke I, Radtke M, Jamra RA, Helbig I, Jansen FE, Koop K, Rudolf G, Küry S, Courchet J, Guerrini R, Lesca G. Neurology. 2024 Jan 23;102(2):e207945. doi: 10.1212/WNL.0000000000207945. (*equally contributing authors)

2. Bi-allelic *ACBD6* variants lead to a neurodevelopmental syndrome with progressive and complex movement disorders.Kaiyrzhanov R, Rad A, Lin SJ, Bertoli-Avella A, Kallemeijn WW, Godwin A, Zaki MS, Huang K, Lau T, Petree C, Efthymiou S, Karimiani EG, Hempel M, Normand EA, Rudnik-Schöneborn S, Schatz UA, Baggelaar MP, Ilyas M, Sultan T, Alvi JR, Ganieva M, Fowler B, Aanicai R, Tayfun GA, Al Saman A, Alswaid A, Amiri N, Asilova N, Shotelersuk V, Yeetong P, Azam M, Babaei M, Monajemi GB, Mohammadi P, Samie S, Banu SH, Pinto Basto J, Kortüm F, Bauer M, Bauer P, Beetz C, Garshasbi M, Issa AH, Eyaid W, Ahmed H, Hashemi N, Hassanpour K, Herman I, Ibrohimov S, Abdul-Majeed BA, Imdad M, Isrofilov M, Kaiyal Q, Khan S, Kirmse B, Koster J, Lourenço CM, Mitani T, Moldovan O, Murphy D, Najafi M, Pehlivan D, Rocha ME, Salpietro V, Schmidts M, Shalata A, Mahroum M, Talbeya JK, Taylor RW, Vazquez D, Vetro A, Waterham HR, Zaman M, Schrader TA, Chung WK, Guerrini R, Lupski JR, Gleeson J, Suri M, Jamshidi Y, Bhatia KP, Vona B, Schrader M, Severino M, Guille M, Tate EW, Varshney GK, Houlden H, Maroofian R.
Brain. 2024 Apr 4;147(4):1436-1456. doi: 10.1093/brain/awad380.

3. GWAS meta-analysis of over 29,000 people with epilepsy identifies 26 risk loci and subtype-specific genetic architecture. International League Against Epilepsy Consortium on Complex Epilepsies. Nat Genet. 2023 Sep;55(9):1471-1482. doi: 10.1038/s41588-023-01485-w.

4. Genome-wide identification and phenotypic characterization of seizure-associated copy number variations in 741,075 individuals. Montanucci L, Lewis-Smith D, Collins RL, Niestroj LM, Parthasarathy S, Xian J, Ganesan S, Macnee M, Brünger T, Thomas RH, Talkowski M; Epi25 Collaborative; Helbig I, Leu C, Lal D. Nat Commun. 2023 Jul 20;14(1):4392. doi: 10.1038/s41467-023-39539-6. PMID: 37474567

5. Stretch-activated ion channel *TMEM63B* associates with developmental and epileptic encephalopathies and progressive neurodegeneration. Vetro A*, Pelorosso C*, Balestrini S*, Masi A, Hambleton S, Argilli E,

Conti V, Giubbolini S, Barrick R, Bergant G, Writzl K, Bijlsma EK, Brunet T, Cacheiro P, Mei D, Devlin A, Hoffer MJV, Machol K, Mannaioni G, Sakamoto M, Menezes MP, Courtin T, Sherr E, Parra R, Richardson R, Roscioli T, Scala M, von Stülpnagel C, Smedley D; TMEM63B collaborators; Genomics England Research Consortium; Torella A, Tohyama J, Koichihara R, Hamada K, Ogata K, Suzuki T, Sugie A, van der Smagt JJ, van Gassen K, Valence S, Vittery E, Malone S, Kato M, Matsumoto N, Ratto GM, Guerrini R. Am J Hum Genet. 2023 Jun 28:S0002-9297(23)00209-4. doi: 10.1016/j.ajhg.2023.06.008. PMID: 37421948. (*equally contributing authors)

6. A novel *SLC5A6* homozygous variant in a family with multivitamin-dependent neurometabolic disorder: Phenotype expansion and long-term follow-up. Montomoli M, Vetro A, Tubili F, Donati MA, Daniotti M, Pochiero F, Rivieri F, Girlando S, Guerrini R. Eur J Med Genet. 2023 Aug;66(8):104808. doi: 10.1016/j.ejmg.2023.104808. PMID: 37391029

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9. Additive effect of *DNAJC30* and *NDUFA9* mutations causing Leigh syndrome. Nesti C, Ticci C, Rubegni A, Doccini S, Scaturro G, Vetro A, Guerrini R, Santorelli FM, Procopio E. J Neurol. 2023 Jun;270(6):3266-3269. doi: 10.1007/s00415-023-11673-7. PMID: 36939934

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13. Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Schwarz N, Seiffert S, Pendziwiat M, Rademacher AV, Brünger T, Hedrich UBS, Augustijn PB, Baier H, Bayat A, Bisulli F, Buono RJ, Bruria BZ, Doyle MG, Guerrini R, Heimer G, Iacomino M, Kearney H, Klein KM, Kousiappa I, Kunz WS, Lerche H, Licchetta L, Lohmann E, Minardi R, McDonald M, Montgomery S, Mulahasanovic L, Oegema R, Ortal B, Papacostas SS, Ragona F, Granata T, Reif PS, Rosenow F, Rothschild A, Scudieri P, Striano P, Tinuper P, Tanteles GA, Vetro A, Zahnert F, Goldberg EM, Zara F, Lal D, May P, Muhle H, Helbig I, Weber Y. Neurology. 2022 May 17;98(20):e2046-e2059. doi: 10.1212/WNL.0000000000200660. Epub 2022 Mar 21. PMID: 35314505

14. Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Marcogliese PC, Deal SL, Andrews J, Harnish JM, Bhavana VH, Graves HK, Jangam S, Luo X, Liu N, Bei D, Chao YH, Hull B, Lee PT, Pan H, Bhadane P, Huang MC, Longley CM, Chao HT, Chung HL, Haelterman NA, Kanca O, Manivannan SN, Rossetti LZ, German RJ, Gerard A, Schwaibold EMC, Fehr S, Guerrini R, Vetro A, England E, Murali CN, Barakat TS, van Dooren MF, Wilke M, van Slegtenhorst M, Lesca G, Sabatier I, Chatron N, Brownstein CA, Madden JA, Agrawal PB, Keren B, Courtin T, Perrin L, Brugger M, Roser T, Leiz S, Mau-Them FT, Delanne J, Sukarova-Angelovska E, Trajkova S, Rosenhahn E, Strehlow V, Platzer K, Keller R, Pavinato L, Brusco A, Rosenfeld JA, Marom R, Wangler MF, Yamamoto S. Cell Rep. 2022 Mar 15;38(11):110517. doi: 10.1016/j.celrep.2022.110517. PMID: 35294868

15. GM3 synthase deficiency in non-Amish patients. Heide S, Jacquemont ML, Cheillan D, Renouil M, Tallot M, Schwartz CE, Miquel J, Bintner M, Rodriguez D, Darcel F, Buratti J, Haye D, Passemard S, Gras D, Perrin L, Capri Y, Gérard B, Piton A, Keren B, Thauvin-Robinet C, Duffourd Y, Faivre L, Poe C, Pervillé A, Héron D, Thévenon J, Arnaud L, LeGuern E, La Selva L, Vetro A, Guerrini R, Nava C, Mignot C. Genet Med. 2022 Feb;24(2):492-498. doi: 10.1016/j.gim.2021.10.007. Epub 2021 Nov 30. PMID: 34906476

16. Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. Epi25 Collaborative. Am J Hum Genet. 2021 Oct 7;108(10):2024. doi: 10.1016/j.ajhg.2021.08.008.

17. Delineating the molecular and phenotypic spectrum of the *SETD1B*-related syndrome. Weerts MJA, Lanko K, Guzmán-Vega FJ, Jackson A, Ramakrishnan R, Cardona-Londoño KJ, Peña-Guerra KA, van Bever Y, van Paassen BW, Kievit A, van Slegtenhorst M, Allen NM, Kehoe CM, Robinson HK, Pang L, Banu SH, Zaman M, Efthymiou S, Houlden H, Järvelä I, Lauronen L, Määttä T, Schrauwen I, Leal SM, Ruivenkamp CAL, Barge-Schaapveld DQCM, Peeters-Scholte CMPCD, Galehdari H, Mazaheri N, Sisodiya SM, Harrison V, Sun A, Thies J, Pedroza LA, Lara-Taranchenko Y, Chinn IK, Lupski JR, Garza-Flores A, McGlothlin J, Yang L, Huang S, Wang X, Jewett T, Rosso G, Lin X, Mohammed S, Merritt JL 2nd, Mirzaa GM, Timms AE, Scheck J, Elting MW, Polstra AM, Schenck L, Ruzhnikov MRZ, Vetro A, Montomoli M, Guerrini R, Koboldt DC, Mosher TM, Pastore MT, McBride KL, Peng J, Pan Z, Willemsen M, Koning S, Turnpenny PD, de Vries BBA, Gilissen C, Pfundt R, Lees M, Braddock SR, Klemp KC, Vansenne F, van Gijn ME, Quindipan C, Deardorff MA, Hamm JA, Putnam AM, Baud R, Walsh L, Lynch SA, Baptista J, Person RE, Monaghan KG, Crunk A, Keller-Ramey J, Reich A, Elloumi HZ, Alders M, Kerkhof J, McConkey H, Haghshenas S; Genomics England Research Consortium, Maroofian R, Sadikovic B, Banka S, Arold ST, Barakat TS. Genet Med. 2021 Nov;23(11):2122-2137. doi: 10.1038/s41436-021-01246-2.

18. Pathogenic *MAST3* Variants in the STK Domain Are Associated with Epilepsy. Spinelli E, Christensen KR, Bryant E, Schneider A, Rakotomamonjy J, Muir AM, Giannelli J, Littlejohn RO, Roeder ER, Schmidt B, Wilson WG, Marco EJ, Iwama K, Kumada S, Pisano T, Barba C, Vetro A, Brilstra EH, van Jaarsveld RH, Matsumoto N, Goldberg-Stern H, Carney PW, Andrews PI, El Achkar CM, Berkovic S, Rodan LH; Undiagnosed Diseases Network (UDN), McWalter K, Guerrini R, Scheffer IE, Mefford HC, Mandelstam S, Laux L, Millichap JJ, Guemez-Gamboa A, Nairn AC, Carvill GL. Ann Neurol. 2021 Aug;90(2):274-284. doi: 10.1002/ana.26147.

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League Against Epilepsy Consortium on Complex Epilepsies; Epi25 Collaborative. Epilepsia. 2021 Jul;62(7):1518-1527. doi: 10.1111/epi.16922.

21. *ATP1A2*- and *ATP1A3*-associated early profound epileptic encephalopathy and polymicrogyria. Vetro A, Nielsen HN, Holm R, Hevner RF, Parrini E, Powis Z, Møller RS, Bellan C, Simonati A, Lesca G, Helbig KL, Palmer EE, Mei D, Ballardini E, Van Haeringen A, Syrbe S, Leuzzi V, Cioni G, Curry CJ, Costain G, Santucci M, Chong K, Mancini GMS, Clayton-Smith J, ATP1A2/A3-collaborators, Bigoni S, Scheffer IE, Dobyns WB, Vilsen B, Guerrini R; Brain. 2021 Apr 21:awab052. doi: 10.1093/brain/awab052.

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