**UOC GENETICA MEDICA**

**Pubblicazioni 2022**

1. Accogli A, Geraldo AF, Piccolo G, Riva A, Scala M, Balagura G, Salpietro V, Madia F, Maghnie M, Zara F, Striano P, Tortora D, Severino M, Capra V. Diagnostic Approach to Macrocephaly in Children. Front Pediatr. 2022 Jan 14;9:794069. doi: 10.3389/fped.2021.794069.
2. Accogli A, Lu S, Musante I, Scudieri P, Rosenfeld JA, Severino M, Baldassari S, Iacomino M, Riva A, Balagura G, Piccolo G, Minetti C, Roberto D, Xia F, Razak R, Lawrence E, Hussein M, Chang EY, Holick M, Calì E, Aliberto E, De-Sarro R, Gambardella A, Network UD, Group SS, Emrick L, McCaffery PJA, Clagett-Dame M, Marcogliese PC, Bellen HJ, Lalani SR, Zara F, Striano P, Salpietro V. Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum. 2022 Feb 26. doi: 10.1007/s12311-022-01379-3.
3. Akin L, Rizzoti K, Gregory LC, Corredor B, Le Quesne Stabej P, Williams H, Buonocore F, Mouilleron S, Capra V, McGlacken-Byrne SM, Martos-Moreno GÁ, Azmanov DN, Kendirci M, Kurtoglu S, Suntharalingham JP, Galichet C, Gustincich S, Tasic V, Achermann JC, Accogli A, Filipovska A, Tuilpakov A, Maghnie M, Gucev Z, Gonen ZB, Pérez-Jurado LA, Robinson I, Badge RL, Argente J, Dattani MT. Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. Genet Med.2022 Feb;24(2):384-397. doi: 10.1016/j.gim.2021.09.019.
4. Asselta R, Paraboschi EM, Stravalaci M, Invernizzi P, Bonfanti P, Biondi A, Pagani I, Pedotti M, Doni A, Scavello F, Mapelli SN, Sironi M, Perucchini C, Varani L, Matkovic M, Cavalli A, Cesana D, Gallina P, Pedemonte N, Capurro V, Clementi N, Mancini N, Bayarri-Olmos R, Garred P, Rappuoli R, Duga S, Bottazzi B, Uguccioni M, Vicenzi E, Mantovani A, Garlanda C. Reply to: Hultström et al., Genetic determinants of mannose-binding lectin activity predispose to thromboembolic complications in critical COVID-19. Mannose-binding lectin genetics in COVID-19. Nat Immunol. 2022 Jun;23(6):865-867.
5. 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.
6. Baldassari S, Cervetto C, Amato S, Fruscione F, Balagura G, Pelassa S, Musante I, Iacomino M, Traverso M, Corradi A, Scudieri P, Maura G, Marcoli M, Zara F: Vesicular Glutamate Release from Feeder-FreehiPSC-Derived Neurons. Int J Mol Sci. 2022 Sep 11;23(18):10545. doi: 10.3390/ijms231810545.
7. Baldassarri M, Zguro K, Tomati V, Pastorino C, Fava F, Croci S, Bruttini M, Picchiotti N, Furini S, Pedemonte N, Gabbi C, Renieri A, Fallerini C; Gen-Covid Multicenter Study. Gain- and Loss-of-Function CFTR Alleles Are Associated with COVID-19 Clinical Outcomes. Cells. 2022 Dec 16;11(24):4096. doi: 10.3390/cells11244096.
8. Borgo C, D'Amore C, Capurro V, Tomati V, Sondo E, Cresta F, Castellani C, Pedemonte N, Salvi M. Targeting the E1 ubiquitin-activating enzyme (UBA1) improves elexacaftor/tezacaftor/ivacaftor efficacy towards F508del and rare misfolded CFTR mutants. Cell Mol Life Sci. 2022 Mar 16;79(4):192. doi: 10.1007/s00018-022-04215-3
9. Braccia C, Christopher JA, Crook OM, Breckels LM, Queiroz RML, Liessi N, Tomati V, Capurro C, Bandiera T, Baldassari S, Pedemonte N, Lilley KS, Armirotti A. CFTR Rescue by Lumacaftor (VX-809) Induces an Extensive Reorganization of Mitochondria in the Cystic Fibrosis Bronchial Epithelium. Cells 2022, 11(12), 1938. doi: 10.3390/cells11121938 2022
10. Brusa I, Sondo E, Falchi F, Pedemonte N, Roberti M, Cavalli A. Proteostasis Regulators in Cystic Fibrosis: Current Development and Future Perspectives. J Med Chem. 2022 Apr 14;65(7):5212-5243. doi: 10.1021/acs.jmedchem.1c01897
11. Calì E, Lin SJ, Rocca C, Sahin Y, Al Shamsi A, El Chehadeh S, Chaabouni M,Mankad K, Galanaki E, Efthymiou S, Sudhakar S, Athanasiou-Fragkouli A, Çelik T, Narlı N, Bianca S, Murphy D, De Carvalho Moreira FM; SYNaPS Study Group, Accogli A, Petree C, Huang K, Monastiri K, Edizadeh M, Nardello R, Ognibene M, De Marco P, Ruggieri M, Zara F, Striano P, Şahin Y, Al-Gazali L, Abi Warde MT, Gerard B, Zifarelli G, Beetz C, Fortuna S, Soler M, Valente EM, Varshney G, Maroofian R, Salpietro V, Houlden H.: A homozygous MED11 C-terminal variant causes a lethal neurodegenerative disease. Genet Med. 2022 Oct;24(10):2194-2203. doi: 10.1016/j.gim.2022.07.013.
12. Campbell C, McCormack M, Patel S, Stapleton C, Bobbili D, Krause R, DepondtC, Sills GJ, Koeleman BP, Striano P, Zara F, Sander JW, Lerche H, Kunz WS, Stefansson K, Stefansson H, Doherty CP, Heinzen EL, Scheffer IE, Goldstein DB, O'Brien T, Cotter D, Berkovic SF; EpiPGX Consortium, Sisodiya SM, Delanty N, Cavalleri GL. A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia. 2022 Jun;63(6):1563-1570. doi: 10.1111/epi.17228.
13. Cappiello F, Casciaro B, Loffredo MR, Puglisi E, Lin Q, Yang D, Conte G, d'Angelo I, Ungaro F, Ferrera L, Barbieri R, Cresti L, Pini A, Di YP, Mangoni ML.: Pulmonary Safety Profile of Esc Peptides and Esc-Peptide-Loaded Poly(lactide-co-glycolide) Nanoparticles: A Promising Therapeutic Approach for Local Treatment of Lung Infectious Diseases Pharmaceutics 2022 Oct 26;14(11):2297. doi: 10.3390/pharmaceutics14112297.
14. "Conteduca G, Baldo C, Arado A, Traverso M, Testa B, Malacarne M, Coviello D, Zara F, Baldassari S.: Generation of induced pluripotent stem cell lines from a patient with Sotos syndrome carrying 5q35 microdeletion. Stem Cell Res. 2022 Dec 21;66:103007. doi: 10.1016/j.scr.2022.103007.
15. Crocco M, Verrico A, Milanaccio C, Piccolo G, De Marco P, Gaggero G, Iurilli V, Di Profio S, Malerba F, Panciroli M, Giordano P, Calevo MG, Casalini E, Di Iorgi N, Garrè ML. Dyslipidemia in Children Treated with a BRAF Inhibitor for Low-Grade Gliomas: A New Side Effect? Cancers 2022 May 29;14(11):2693. doi: 10.3390/cancers14112693.
16. Croci C, Traverso M, Baratto S, Iacomino M, Pedemonte M, Caroli F, Scala M, Bruno C, Fiorillo C.: Congenital myopathy associated with a novel mutation in MEGF10 gene, myofibrillar alteration and progressive course Acta Myol. 2022 Sep 30;41(3):111-116. doi: 10.36185/2532-1900-076.
17. Cuccurullo C, Miele G, Piccolo G, Bilo L, Accogli A, D'Amico A, Fratta M, Guerrisi S, Iacomino M, Salpietro V, Ugga L, Striano P, Coppola A.: Hydranencephaly in CENPJ-related Seckel syndrome. Eur J Med Genet. 2022 Dec;65(12):104659. doi: 10.1016/j.ejmg.2022.104659.
18. D'Onofrio G, Riva A, Di Rosa G, Cali' E, Efthymiou S, Gitto E, Madia F, Accogli A, Zara F, Houlden H, Salpietro V, Striano P, Soler D. Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review, Brain Dev. 2022 Apr 9;S0387-7604(22)00057-2. doi: 10.1016/j.braindev.2022.03.010
19. Farinha CM, Brodsky JL, Pedemonte N. Fundamental and translational research in Cystic Fibrosis - why we still need it. J Cyst Fibros. 2023 Mar;22 Suppl 1:S1-S4. doi: 10.1016/j.jcf.2022.12.010.
20. Fossa P, Uggeri M, Orro A, Urbinati C, Rondina A, Milanesi M, Pedemonte N, Pesce E, Padoan R, Ford RC, Meng X, Rusnati M, D'Ursi P. Virtual Drug Repositioning as a Tool to Identify Natural Small Molecules That Synergize with Lumacaftor in F508del-CFTR Binding and Rescuing. Int J Mol Sci. 2022 Oct 14;23(20):12274. doi: 10.3390/ijms232012274.
21. Gemelli C, Traverso M, Trevisan L, Fabbri S, Scarsi E, Carlini B, Prada V, Mongini T, Ruggiero L, Patrone S, Gallone S, Iodice R, Pisciotta L, Zara F, Origone P, Rota E, Minetti C, Bruno C, Schenone A, Mandich P, Fiorillo C, Grandis M.Gemelli C, et al. Among authors: zara f. An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic hyperCKemia. Muscle Nerve. 2022 Jan;65(1):96-104. doi: 10.1002/mus.27448.
22. Geraldo AF, Luis A, Alves CAPF, Tortora D, Guimarães J, Reimão S, Pavanello M, de Marco P, Scala M, Capra V, Rossi A, Schwartz ES, Mankad K, Severino M. Spinal involvement in pediatric familial cavernous malformation syndrome. Neuroradiology 2022 Apr 22. doi: 10.1007/s00234-022-02958-1.
23. Geroldi A, Trevisan L, Gaudio A, Gotta F, Patrone S, Origone P, Grandis M, Gemelli C, Schenone A, Accogli A, Zara F, Mandich P, Bellone E: A misleading presentation of Mohr-Tranebjaerg syndrome: What is hidden behind an axonal neuropathy? Parkinsonism Relat Disord. 2022 Sep; 102:54-56. doi:10.1016/j.parkreldis.
24. Ghigo A, Murabito A, Sala V, Pisano AR, Bertolini S, Gianotti A, Caci E, Montresor A, Premchandar A, Pirozzi F, Ren K, Della Sala A, Mergiotti M, Richter W, de Poel E, Matthey M, Caldrer S, Cardone RA, Civiletti F, Costamagna A, Quinney NL, Butnarasu C, Visentin S, Ruggiero MR, Baroni S, Crich SG, Ramel D, Laffargue M, Tocchetti CG, Levi R, Conti M, Lu XY, Melotti P, Sorio C, De Rose V, Facchinetti F, Fanelli V, Wenzel D, Fleischmann BK, Mall MA, Beekman J, Laudanna C, Gentzsch M, Lukacs GL, Pedemonte N, Hirsch E. Airway administration of a PI3Kgamma mimetic peptide triggers CFTR gating, bronchodilation and reduced inflammation in obstructive airway diseases. Sci Transl Med. 2022 Mar 30;14(638):eabl6328. doi: 10.1126/scitranslmed.abl6328.
25. Giacomini T, Scala M, Nobile G, Severino M, Tortora D, Nobili L, Accogli A, Torella A, Capra V, Mancardi MM, Nigro V; TUDP study group. De novo POLR2A p.(Ile457Thr) variant associated with early-onset encephalopathy and cerebellar atrophy: expanding the phenotypic spectrum. Brain Dev. 2022 Aug;44(7):480-485. doi: 10.1016/j.braindev.2022.04.002
26. Golec A, Pranke I, Scudieri P, Hayes K, Dreano E, Dunlevy F, Hatton A, Downey DG, Galietta L, Sermet I. Isolation, cultivation, and application of primary respiratory epithelial cells obtained by nasal brushing, polyp samples, or lung explants. STAR Protoc. 2022 May 27;3(2):101419. doi: 10.1016/j.xpro.2022.101419.
27. Gorrieri G, Zara F, Scudieri P. SLC26A9 as a Potential Modifier and Therapeutic Target in Cystic Fibrosis Lung Disease. Biomolecules. 2022 Jan 25;12(2):202. doi: 10.3390/biom12020202.
28. Guidone D, Buccirossi M, Scudieri P, Genovese M, Sarnataro S, De Cegli R, Cresta F, Terlizzi V, Planelles G, Crambert G, Sermet I, Galietta LJ.: Airway surface hyperviscosity and defective mucociliary transport by IL-17/TNF-α are corrected by β-adrenergic stimulus JCI Insight. 2022 Nov 22;7(22):e164944. doi: 10.1172/jci.insight.164944.
29. Calhoun JD, Aziz MC, Happ HC, Gunti J, Gleason C, Mohamed N, Zeng K, Hiller M, Bryant E, Mithal DS, Bellinski I, Kinsley L, Grimmel M, Schwaibold EMC, Smith-Hicks C, Chassevent A, Scala M, Accogli A, Torella A, Striano P, Capra V, Bird LM, Ben-Sahra I, Ekhilevich N, Hershkovitz T, Weiss K, Millichap J, Gerard EE, Carvill GL. mTORC1 functional assay reveals SZT2 loss-of-function variants and a founder in-frame deletion. Brain. 2022 Jun 30;145(6):1939-1948. doi: 10.1093/brain/awab451.
30. 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 M, Jansen A, Scalais E, Srivastava S, Tan WH, Olson HE, Loddenkemper T, Poduri A, Helbig KL, Helbig I, Fitzgerald MP, Goldberg EM, Roser T, Borggraefe I, Brünger T, May P, Lal D, Lederer D, Rubboli G, Heyne HO, Lesca G, Hedrich UBS, Benda J, Gardella E, Lerche H, Møller RS: 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.
31. Koko M, Motelow JE, Stanley KE, Bobbili DR, Dhindsa RS, May P; Canadian Epilepsy Network; Epi4K Consortium; Epilepsy Phenome/Genome Project; EpiPGX Consortium; EuroEPINOMICS-CoGIE Consortium. Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. Epilepsia. 2022 Mar;63(3):723-735. doi: 10.1111/epi.17166.
32. Mangano GD, Riva A, Fontana A, Salpietro V, Mangano GR, Nobile G, Orsini A, Iacomino M, Battini R, Astrea G, Striano P, Nardello R. De novo GRIN2A variants associated with epilepsy and autism and literature review. Epilepsy Behav. 2022 Apr;129:108604. doi: 10.1016/j.yebeh.2022.108604.
33. Martina MG, Sannio F, Crespan E, Pavone M, Simoncini A, Barbieri F, Perini C, Pesce E, Maga G, Pedemonte N, Docquier JD, Radi M. Towards Innovative Antibacterial-Correctors for Cystic Fibrosis Targeting the Lung Microbiome with a Multifunctional Effect. ChemMedChem. 2022 May 31. doi: 10.1002/cmdc.202200277.
34. Marzia Ognibene , Patrizia De Marco , Stefano Parodi , Mariaclaudia Meli , Andrea Di Cataldo, Federico Zara and Annalisa Pezzolo. Genomic Analysis Made It Possible to Identify Gene-Driver Alterations Covering the Time Window between Diagnosis of Neuroblastoma 4S and the Progression to Stage 4. Int. J. Mol. Sci. 2022, 23, 6513. doi: 10.3390/ijms23126513
35. Marzia Ognibene, Davide Cangelosi, Stefania Sorrentino, Sabrina Zanardi, Federico Zara, Annalisa Pezzolo, Stefano Parodi. E2F3 gene expression is a potential negative prognostic marker for localised and MYCN not-amplified neuroblastoma: Results of in silico analysis of 786 samples. Pediatr Blood Cancer.2022;e29800. doi: 1002/pbc.29800.
36. Mendonça LO, Grossi A, Caroli F, de Oliveira RA, Kalil J, Castro FFM, Pontillo A, Ceccherini I, Barros MAMT, Gattorno M. A case report of a novel compound heterozygous mutation in a Brazilian patient with deficiency of Interleukin-1 receptor antagonist (DIRA). Pediatr Rheumatol Online J. 2020 Aug 20;18(1):67. doi: 10.1186/s12969-020-00454-5.
37. Morabito LA, Allegri AEM, Capra AP, Capasso M, Capra V, Garaventa A, Maghnie M, Briuglia S, Wasniewska MG. Osteogenesis imperfecta/Ehlers–Danlos overlap syndrome and Neuroblastoma - Case Report and Review of literature. Genes (Basel). 2022 Mar 25;13(4):581. doi: 10.3390/genes13040581.
38. Naim A, Accogli A, Amadori E, D'Onofrio G, Madia F, Tortora D, Zara F, Striano P, Salpietro V, Severino M: Abnormal course of the corticospinal tracts in KIF5C-related encephalopathy. Eur J Med Genet. 2022 Sep 16;65(11):104622. doi: 10.1016/j.ejmg.2022.104622.
39. Nishikawa M, Scala M, Umair M, Ito H, Waqas A, Striano P, Zara F, Costain G, Capra V, Nagata KI. The gain-of-function p.F28S variant in RAC3 disrupts neuronal differentiation, migration, and axonogenesis during cortical development, leading to neurodevelopmental disorder". J Med Genet. 2022 May 20:jmedgenet-2022-108483. doi: 10.1136/jmedgenet-2022-108483.
40. Orsini A, Ferrari D, Riva A, Santangelo A, Macrì A, Freri E, Canafoglia L, D'Aniello A, Di Gennaro G, Massimetti G, Minetti C, Zara F, Michelucci R, Tumber A, Vincent A, Minassian BA, Striano P. Ocular phenotype and electroretinogram abnormalities in Lafora disease and correlation with disease stage. J Neurol. 2022 Jul;269(7):3597-3604. doi: 10.1007/s00415-022-10974-7.
41. Orsini A, Santangelo A, Bravin F, Bonuccelli A, Peroni D, Battini R, Foiadelli T, Bertini V, Valetto A, Iacomino M, Nigro V, Torella AL, Scala M, Capra V, Vari MS, Fetta A, Di Pisa V, Montanari F, Epifanio R, Bonanni P, Giorda R, Operto F, Pastorino G, Sarigecili E, Sardaroglu E, Okuyaz C, Bozdogan S, Musante L, Faletra F, Zanus C, Ferretti A, Vigevano F, Striano P, Cordelli DM. Expanding Phenotype of Poirier-Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes (Basel). 2022 Jan 30;13(2):276. doi: 10.3390/genes13020276.
42. Panicucci C, Schiaffino MC, Nesti C, Derchi M, Trocchio G, Severino M, Stagnaro N, Priolo E, Zara F, Santorelli FM, Bruno C.: Long term follow-up in two siblings with Sengers syndrome: Case report Ital J Pediatr. 2022 Oct 17;48(1):180. doi: 10.1186/s13052-022-01370-y.
43. Borgia P, Baldassari S, Pedemonte N, Alkhunaizi E, D'Onofrio G, Tortora D, Calì E, Scudieri P, Balagura G, Musante I, Diana MC, Pedemonte M, Vari MS, Iacomino M, Riva A, Chimenz R, Mangano GD, Mohammadi MH, Toosi MB, Ashrafzadeh F, Imannezhad S, Karimiani EG, Accogli A, Schiaffino MC, Maghnie M, Soler MA, Echiverri K, Abrams CK, Striano P, Fortuna S, Maroofian R, Houlden H, Zara F, Fiorillo C, Salpietro V. Genotype-phenotype correlations and disease mechanisms in PEX13-related Zellweger spectrum disorders. Orphanet J Rare Dis. 2022 Jul 19;17(1):286. doi: 10.1186/s13023-022-02415-5.
44. Parenti I, Leitão E, Kuechler A, Villard L, Goizet C, Courdier C, Bayat A, Rossi A, Julia S, Bruel AL, Tran Mau-Them F, Nambot S, Lehalle D, Willems M, Lespinasse J, Ghoumid J, Caumes R, Smol T, El Chehadeh S, Schaefer E, Abi-Warde MT, Keren B, Afenjar A, Tabet AC, Levy J, Maruani A, Aledo-Serrano Á, Garming W, Milleret-Pignot C, Chassevent A, Koopmans M, Verbeek NE, Person R, Belles R, Bellus G, Salbert BA, Kaiser FJ, Mazzola L, Convers P, Perrin L, Piton A, Wiegand G, Accogli A, Brancati F, Benfenati F, Chatron N, Lewis-Smith D, Thomas RH, Zara F, Striano P, Lesca G, Depienne C.The different clinical facets of SYN1-related neurodevelopmental disorders.Front Cell Dev Biol. 2022 Dec 8;10:1019715. doi: 10.3389/fcell.2022.1019715.
45. Parodi A, Righetti G, Pesce E, Salis A, Tomati V, Pastorino C, Tasso B, Benvenuti M, Damonte G, Pedemonte N, Cichero E, Millo E. Journey on VX-809-Based Hybrid Derivatives towards Drug-like F508del-CFTR Correctors: From Molecular Modeling to Chemical Synthesis and Biological Assays. Pharmaceuticals (Basel). 2022 Feb 23;15(3):274. doi: 10.3390/ph15030274
46. Pedemonte N. Nasal epithelial cells as a gold-standard predictive model for personalized medicine in cystic fibrosis. J Physiol. 2022 Mar;600(6):1285-1286. doi: 10.1113/JP282586
47. Philp AR, Miranda F, Gianotti A, Mansilla A, Scudieri P, Musante I, Vega G, Figueroa CD, Galietta LJV, Sarmiento JM, Flores CA. KCa3.1 differentially regulates trachea and bronchi epithelial gene expression in a chronic-asthma mouse model. Physiol Genomics. 2022 Jun 6. doi: 10.1152/physiolgenomics.00134.2021.
48. Piccolo G, Verrico A, Morana G, Piatelli G, De Marco P, Iurilli V, Antonelli M, Gaggero G, Ramaglia A, Crocco M, Caruggi S, Milanaccio C, Garrè ML, Pavanello M.: Early molecular diagnosis of BRAF status drives the neurosurgical management in BRAF V600E-mutant pediatric low-grade gliomas: a case report. BMC Pediatr. 2022 Nov 29;22(1):685. doi: 10.1186/s12887-022-03711-6.
49. Principi E, Sondo E, Bianchi G, Ravera S, Morini M, Tomati V, Pastorino C, Zara F, Bruno C, Eva A, Pedemonte N, Raffaghello L. Targeting of Ubiquitin E3 Ligase RNF5 as a Novel Therapeutic Strategy in Neuroectodermal Tumors. Cancers (Basel). 2022 Apr 1;14(7):1802. doi:10.3390/cancers14071802
50. Raffaghello L, Principi E, Baratto S, Panicucci C, Pintus S, Antonini F, Del Zotto G, Benzi A, Bruzzone S, Scudieri P, Minetti C, Gazzerro E, Bruno C. P2X7 Receptor Antagonist Reduces Fibrosis and Inflammation in a Mouse Model of Alpha-Sarcoglycan Muscular Dystrophy. Pharmaceuticals (Basel). 2022 Jan 13;15(1):89. doi: 10.3390/ph15010089.
51. Riva A, Nobile G, Giacomini T, Ognibene M, Scala M, Balagura G, Madia F, Accogli A, Romano F, Tortora D, Severino M, Scudieri P, Baldassari S, Musante I, Uva P, Salpietro V, Torella A, Nigro V, Capra V, Nobili L, Striano P, Mancardi MM, Zara F, Iacomino M. A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Front Pediatr. 2022 Apr 29;10:847549. doi: 10.3389/fped.2022.847549
52. Romano F, Falco M, Cappuccio G, Brunetti-Pierri N, Lonardo F, Torella A, Digilio MC, Dentici ML, Alfieri P, Agolini E, Novelli A, Garavelli L, Accogli A; TUDP, Striano P, Scarano G, Nigro V, Scala M, Capra V. Genotype-phenotype spectrum and correlations in Xia-Gibbs syndrome: Report of five novel cases and literature review. Birth Defects Res. 2022 Jun 18. doi: 10.1002/bdr2.2058.
53. Romano F, Madia F, De Marco P, Ognibene M, Guerrisi S, Scala M, Iacomino M, Baldassari S, Vercellino N, Manunza F, Tallone R, Pavanello M, Piatelli G, Garaventa A, Zara F, Capra V.: Clinical and genetic analysis of patients with segmental overgrowth features and somatic mammalian target of rapamycin (mTOR) pathway disruption: Possible novel clinical issues. Birth Defects Res 2022 Dec 1;114(20):1440-144. doi: 10.1002/bdr2.2113
54. Ruscitti F, Cerminara M, Iascone M, Pezzoli L, Rosti G, Romano F, Ronchetto P, Martucciello G, Buratti S, Buffelli F, Bocciardi R, Puliti A, Divizia MT. An example of parenchymal renal sparing in the context of complex malformations due to a novel mutation in the PBX1 gene. Birth Defects Res. 2022 Jul 15;114(12):674-681. doi: 10.1002/bdr2.2065. Epub 2022 Jun 25. PMID: 35751431.Salpietro V, Galassi Deforie V, Efthymiou S, O'Connor E, Marcé-Grau A, Maroofian R, Striano P, Zara F, Morrow MM; SYNAPS Study Group, Reich A, Blevins A, Sala-Coromina J, Accogli A, Fortuna S, Alesandrini M, Au PYB, Singhal NS, Cogne B, Isidor B, Hanna MG, Macaya A, Kullmann DM, Houlden H, Männikkö R. : De novo KCNA6 variants with attenuated KV 1.6 channel deactivation in patients with epilepsy. Epilepsia. 2022 Nov 1. doi: 10.1111/epi.17455.
55. Scala M, De Grandis E, Nobile G, Iacomino M, Madia F, Capra V, Nobili L, Zara F, Striano P.: Biallelic ZBTB11 variants associated with complex neuropsychiatric phenotype featuring Tourette syndrome. Brain. 2022 Sep 7:awac323. doi: 10.1093/brain/awac323.
56. Scala M, Drouot N, MacLennan SC, Wessels MW, Krygier M, Pavinato L, Telegrafi A, de Man SA, van Slegtenhorst M, Iacomino M, Madia F, Scudieri P, Uva P, Giacomini T, Nobile G, Mancardi MM, Balagura G, Galloni GB, Verrotti A, Umair M, Khan A, Liebelt J, Schmidts M, Langer T, Brusco A, Lipska-Ziętkiewicz BS, Saris JJ, Charlet-Berguerand N, Zara F, Striano P, Piton A. De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Hum Mutat. 2022 May 24. doi: 10.1002/humu.24414.
57. 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. Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology. 2022 May 17;98(20):e2046-e2059. doi: 10.1212/WNL.0000000000200660.
58. Scudieri P, Pusch M.: Take a big sip and shrink it with ASOR. Pflugers Arch. 2022 Nov;474(11):1121-1122. doi: 10.1007/s00424-022-02750-3. Epub 2022 Sep 29.
59. Sondo E, Cresta F, Pastorino C, Tomati V, Capurro V, Pesce E, Lena M, Iacomino M, Baffico AM, Coviello D, Bandiera T, Zara F, Galietta LJV, Bocciardi R, Castellani C, Pedemonte N. The L467F-F508del Complex Allele Hampers Pharmacological Rescue of Mutant CFTR by Elexacaftor/Tezacaftor/Ivacaftor in Cystic Fibrosis Patients: The Value of the Ex Vivo Nasal Epithelial Model to Address Non-Responders to CFTR-Modulating Drugs. Int J Mol Sci. 2022 Mar 15;23(6):3175. doi: 10.3390/ijms23063175
60. Steiner A, Reygaerts T, Pontillo A, Ceccherini I, Moecking J, Moghaddas F, Davidson S, Caroli F, Grossi A, Castro FFM, Kalil J, Gohr FN, Schmidt FI, Bartok E, Zillinger T, Hartmann G, Geyer M, Gattorno M, Mendonça LO, Masters SL. Recessive NLRC4-Autoinflammatory Disease Reveals an Ulcerative Colitis Locus. J Clin Immunol. 2022 Feb;42(2):325-335. doi: 10.1007/s10875-021-01175-4.
61. Stephenson SEM, Costain G, Blok LER, Silk MA, Nguyen TB, Dong X, Alhuzaimi DE, Dowling JJ, Walker S, Amburgey K, Hayeems RZ, Rodan LH, Schwartz MA, Picker J, Lynch SA, Gupta A, Rasmussen KJ, Schimmenti LA, Klee EW, Niu Z, Agre KE, Chilton I, Chung WK, Revah-Politi A, Au PYB, Griffith C, Racobaldo M, Raas-Rothschild A, Ben Zeev B, Barel O, Moutton S, Morice-Picard F, Carmignac V, Cornaton J, Marle N, Devinsky O, Stimach C, Wechsler SB, Hainline BE, Sapp K, Willems M, Bruel AL, Dias KR, Evans CA, Roscioli T, Sachdev R, Temple SEL, Zhu Y, Baker JJ, Scheffer IE, Gardiner FJ, Schneider AL, Muir AM, Mefford HC, Crunk A, Heise EM, Millan F, Monaghan KG, Person R, Rhodes L, Richards S, Wentzensen IM, Cogné B, Isidor B, Nizon M, Vincent M, Besnard T, Piton A, Marcelis C, Kato K, Koyama N, Ogi T, Goh ES, Richmond C, Amor DJ, Boyce JO, Morgan AT, Hildebrand MS, Kaspi A, Bahlo M, Friðriksdóttir R, Katrínardóttir H, Sulem P, Stefánsson K, Björnsson HT, Mandelstam S, Morleo M, Mariani M; TUDP Study Group, Scala M, Accogli A, Torella A, Capra V, Wallis M, Jansen S, Weisfisz Q, de Haan H, Sadedin S; Broad Center for Mendelian Genomics, Lim SC, White SM, Ascher DB, Schenck A, Lockhart PJ, Christodoulou J, Tan TY. Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome Am J Hum Genet. 2022 Apr 7;109(4):601-617. doi: 10.1016/j.ajhg.2022.03.002.PMID: 35395208
62. Stravalaci M, Pagani I, Paraboschi EM, Pedotti M, Doni A, Scavello F, Mapelli SN, Sironi M, Perucchini C, Varani L, Matkovic M, Cavalli A, Cesana D, Gallina P, Pedemonte N, Capurro V, Clementi N, Mancini N, Invernizzi P, Bayarri-Olmos R, Garred P, Rappuoli R, Duga S, Bottazzi B, Uguccioni M, Asselta R, Vicenzi E, Mantovani A, Garlanda C. Recognition and inhibition of SARS-CoV-2 by humoral innate immunity pattern recognition molecules. Nat Immunol. 2022 Feb;23(2):275-286. doi: 10.1038/s41590-021-01114-w
63. Tomati V, Costa S, Capurro V, Pesce E, Pastorino C, Lena M, Sondo E, Di Duca M, Cresta F, Cristadoro S, Zara F, Galietta LJV, Bocciardi R, Castellani C, Lucanto MC, Pedemonte N. Rescue by elexacaftor-tezacaftor-ivacaftor of the G1244E cystic fibrosis mutation's stability and gating defects are dependent on cell background. J Cyst Fibros. 2023 May;22(3):525-537. doi: 10.1016/j.jcf.2022.12.005.
64. Vitobello A, Mazel B, Lelianova VG, Zangrandi A, Petitto E, Suckling J, Salpietro V, Meyer R, Elbracht M, Kurth I, Eggermann T, Benlaouer O, Lall G, Tonevitsky AG, Scott DA, Chan KM, Rosenfeld JA, Nambot S, Safraou H, Bruel AL, Denommé-Pichon AS, Tran Mau-Them F, Philippe C, Duffourd Y, Guo H, Petersen AK, Granger L, Crunk A, Bayat A, Striano P, Zara F, Scala M, Thomas Q, Delahaye A, de Sainte Agathe JM,. Buratti J, Kozlov SV, Faivre L, Thauvin-Robinet C, Ushkaryov Y.: ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and alters synaptic activity and behavior in a mouse model. Am J Hum Genet. 2022 Aug 4;109(8):1436-1457. doi: 10.1016/j.ajhg.2022.06.011.
65. Xian J, Parthasarathy S, Ruggiero SM, Balagura G, Fitch E, Helbig K, Gan J, Ganesan S, Kaufman MC, Ellis CA, Lewis-Smith D, Galer P, Cunningham K, O'Brien M, Cosico M, Baker K, Darling A, Veiga de Goes F, El Achkar CM, Doering JH, Furia F, García-Cazorla Á, Gardella E, Geertjens L, Klein C, Kolesnik-Taylor A, Lammertse H, Lee J, Mackie A, Misra-Isrie M, Olson H, Sexton E, Sheidley B, Smith L, Sotero L, Stamberger H, Syrbe S, Thalwitzer KM, van Berkel A, van Haelst M, Yuskaitis C, Weckhuysen S, Prosser B, Son Rigby C, Demarest S, Pierce S, Zhang Y, Møller RS, Bruining H, Poduri A, Zara F, Verhage M, Striano P, Helbig I. Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain. 2022 Jun 3;145(5):1668-1683. doi: 10.1093/brain/awab327.
66. Zuccolini P, Ferrera L, Remigante A, Picco C, Barbieri R, Bertelli S, Moran O, Gavazzo P, Pusch M. The VRAC blocker DCPIB directly gates the BK channels and increases intracellular Ca2+ in melanoma and pancreatic duct adenocarcinoma cell lines. Br J Pharmacol. 2022 Jul;179(13):3452-3469. doi: 10.1111/bph.15810.
67. Geraldo AF, Alves CAPF, Luis A, Tortora D, Guimarães J, Abreu D, Reimão S, Pavanello M, de Marco P, Scala M, Capra V, Vaz R, Rossi A, Schwartz ES, Mankad K, Severino M.: Natural history of familial cerebral cavernous malformation syndrome in children: a multicenter cohort study.Neuroradiology. 2022 Oct 6. doi: 10.1007/s00234-022-03056-y.
68. Scala M, Nishikawa M, Ito H, Tabata H, Khan T, Accogli A, Davids L, Ruiz A, Chiurazzi P, Cericola G, Schulte B, Monaghan KG, Begtrup A, Torella A, Pinelli M, Denommé-Pichon AS, Vitobello A, Racine C, Mancardi MM, Kiss C, Guerin A, Wu W, Gabau Vila E, Mak BC, Martinez-Agosto JA, Gorin MB, Duz B, Bayram Y, Carvalho CMB, Vengoechea JE, Chitayat D, Tan TY, Callewaert B, Kruse B, Bird LM, Faivre L, Zollino M, Biskup S; Undiagnosed Diseases Network; Telethon Undiagnosed Diseases Program, Striano P, Nigro V, Severino M,Capra V, Costain G, Nagata KI. Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenoty BRAIN. 2022 Sep 14;145(9):3308-3327. doi: 10.1093/brain/awac106.PMID:35851598.