

**Lisa Pavinato, PhD**

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**PROFESSIONAL APPOINTMENTS**

07/2024 – present **Postdoctoral fellow**, Institute of Oncology Research (IOR), affiliated to Università della Svizzera italiana (USI), and member of Bios+, Bellinzona, Switzerland (Advisor: Prof. Arianna Baggiolini)

02/2023 – 06/2024 **Postdoctoral fellow**, University of Turin, Turin, Italy (Advisor: Prof. Alfredo Brusco)

03/2023 – 06/2023 **Visiting postdoctoral fellow**, Institute of Oncology Research (IOR), affiliated to Università della Svizzera italiana (USI), and member of Bios+, Bellinzona, Switzerland (Advisor: Prof. Arianna Baggiolini). Supported by EMBO and CorEUStem grants.

**EDUCATION AND TRAINING**

10/2018 – 02/2023 **University of Turin**, Turin, Italy.  
Ph.D. Program in Biomedical Sciences and Oncology, curriculum Medical Genetics.  
PhD defense: 07.02.2023. PhD advisor: Prof. Alfredo Brusco

01/2023 **University of Salento**, Lecce, Italy.  
National Italian Qualification for the profession of Biologist.

10/2020 – 07/2021 **University of Cologne**, Cologne, Germany.  
Visiting Ph.D. Student in the laboratory of Neurogenetics at Center for Molecular Medicine Cologne. Supported by DAAD-Fellowship. Advisor: Prof. Dr. Brunhilde Wirth

10/2016 – 10/2018 **University of Turin**, Turin, Italy.  
Master degree in Medical Biotechnologies. Final mark 110/110 with honours and recommendation for publication. Experimental thesis advisor: Prof. Paola Defilippi

10/2013 – 10/2016 **University of Turin**, Turin, Italy.  
Bachelor's degree in biotechnologies.

**PRIZES AND AWARDS**

06/2024 ESHG conference travel grant

05/2024 Award GiovedìScienza XXIII edition, 2nd place

04/2024 *Recti Eques* Award for Rare Diseases and Paladini Italiani della Salute Acknowledgment (supported by Italian Ministry of Health)

10/2023 Best oral presentation award at Italian Society of Human Genetics Conference

06/2023 Early Career Award candidate ESHG conference

10/2022 The company of Biologists DMM Travel grant for ASHG conference

06/2022 Early Career Award candidate ESHG conference

11/2021 Best oral presentation award Italian Society of Human Genetics Conference

10/2021 Charles J. Epstein Trainee Award for Excellence in Human Genetics Research, pre-doctoral finalist

11/2020 Best poster presentation award at Italian Society of Human Genetics Conference

**PERSONAL GRANTS**

06/2023-06/2023 CorEUStem Short Term Scientific Mission Grant - ref. n° E-COST-GRANT-CA20140-495ffa48

03/2023-05/2023 EMBO Scientific Exchange Grant - ref. n° 9957

10/2020-07/2021      Deutscher Akademischer Austauschdienst (DAAD) Research Grant, one-year grant for Doctoral Candidates, 2020/21 - ref. n° 91781256

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#### FUNDED PROJECTS

2022-2023	Emma and Ernesto Rulfo Association. Personal role: co-main applicant with Prof. A. Brusco. Amount: € 20.000
2022-2025	Italian Ministry of Health and Education (PRIN). Personal role: participant.
2021-2022	Emma and Ernesto Rulfo Association. Personal role: co-main applicant with Prof. A. Brusco. Amount: € 20.000
2020-2021	Emma and Ernesto Rulfo Association. Personal role: co-main applicant with Prof. A. Brusco. Amount: € 30.000

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#### PLATFORM TALKS

10/2024	Proteintech Webinar Series. Virtual.
10/2023	Italian Society of Human Genetics conference. Rimini, Italy.
09/2023	Italian Society of Neuroscience conference. Turin, Italy.
06/2023	European Society of Human Genetics conference. Glasgow, UK.
10/2022	American Society of Human Genetics conference. Los Angeles, California, USA.
09/2022	Italian Society of Human Genetics conference. Trieste, Italy.
06/2022	European Society of Human Genetics conference. Wien, Austria.
11/2021	Italian Society of Human Genetics conference. Virtual.
10/2021	American Society of Human Genetics conference. Virtual.

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#### MEMBERSHIPS AND EXTERNAL ACTIVITIES

2024-present	Invited Human Disease Genes curator for <i>CAPRN1</i> and <i>RPH3A</i> genes.
2022-present	LeidenOpenVariationDatabase (LOVD) curator, for <i>CAPRN1</i> gene.
2022-present	Member of The European Network for Stem Cell Core Facilities (COST Action CorEuStem CA20140 WG1).
2022-present	Member of European Society of Human Genetics.
2022-present	Member of Italian Society of Human Genetics.
2021-2023	Ph.D. students' representative.
2020-present	Member of the Autism Sequencing Consortium.
2020-present	Member of American Society of Human Genetics.

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#### SCIENTIFIC REVIEWING AND MODERATOR ACTIVITIES

10/2023	Session moderator at American Society of Human Genetics Conference. Washington D.C., USA.
2022	Reviewer for ESHG conference abstracts.
2020-present	Reviewer for several journals, i.e. Brain, Trends in Cell Biology, etc.

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#### TEACHING AND STUDENTS' SUPERVISION

07/2024 – present	Supervision of two PhD students and teaching of complex 3D hPSC-based assembloid models.
01/2021 – present	Lecturer for Neurogenetics; Master's course in Neuroscience at University of Turin, Turin, Italy.
01/2021 – present	Expert for supplementary teaching; graduate school in Medical Genetics at University of Turin, Turin, Italy.
09/2023 – 06/2024	Supervision of on postgraduate trainee and two Master's students, teaching of hPSC-based models.
01/2023 – 12/2023	Thesis co-supervisor of Dr Valeria Gattolin, who graduated with the maximum rating.

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#### OUTREACH ACTIVITIES

06/2024	Attendance to “Oratory Art” and “Dissemination of Science” courses.
05/2024	Talk for GiovedìScienza, organized for High School students by CentroScienza Onlus.
2023-present	Ideator and curator of the dissemination page PilloleDiGenetica (Italian only).
2019-2023	Collaboration in the activities of European Researchers Night.
02/2023	Talk and activities for “Women in STEM” project for High School students.

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## PUBLICATIONS IN PEER-REVIEWED SCIENTIFIC JOURNALS

1. *Biallelic PI4KA Mutations Disrupt B-Cell Metabolism and Cause B-Cell Lymphopenia and Hypogammaglobulinemia.* | Saettini F, Guerra F, Mauri M, Salter CG, Adam MP, Adams D, Baple EL, Barredo E, Bhatia S, Borkhardt A, Brusco A, Bugarin C, Chinello C, Crosby AH, D'Souza P, Denti V, Fazio G, Giuliani S, Kuehn HS, Amel H, Elmi A, Lo B, Malighetti F, Mandrile G, Martín-Nalda A, Mefford HC, Moratto D, Emam Mousavi F, Nelson Z, Gutiérrez-Solana LG, Macnamara E, Michaud V, O'Leary M, Pagani L, **Pavinato L**, Santamaria PV, Planas-Serra L, Quadri M, Raspall-Chaure M, Rebellato S, Rosenzweig SD, Roubertie A, Holzinger D, Deal C, Vockley CW, Savino AM, L Stoddard J, Uhlig HH, Pujol A, Magni F, Paglia G, Cazzaniga G, Piazza R, Barberis M, Biondi A. *J Clin Immunol.* | 2024-09-23 | journal-article. DOI: 10.1007/s10875-024-01793-8
2. *DNA methylation analysis in patients with neurodevelopmental disorders improves variant interpretation and reveals complexity* | S. Trajkova, J. Kerkhof, M. Rossi Sebastiano, **L. Pavinato**, E. Ferrero, C. Giovenino, D. Carli, E. Di Gregorio, R. Marinoni, G. Mandrile, F. Palermo, S. Carestiato, S. Cardaropoli, V. Pullano, A. Rinninella, E. Giorgio, T. Pippucci, P. Dimartino, J. Rzasa, K. Rooney, H. McConkey, A. Petlichkovski, B. Pasini, E. Sukarova-Angelovska, C. M. Campbell, K. Metcalfe, S. Jenkinson, S. Banka, A. Mussa, G. Battista Ferrero, B. Sadikovic, A. Brusco  
*HGG Advances* | 2023-07-2024 | journal-article | DOI: 10.1016/j.xhgg.2024.100309
3. *Identification of the DNA methylation signature of Mowat-Wilson syndrome.* | S. Caraffi, L. van der Laan, K. Rooney, S. Trajkova, R. Zuntini, R. Relator, S. Haghshenas, M. Levy, C. Baldo, G. Mandrile, C. Lauzon-Young, D. Maria Cordelli, I. Ivanovski, A. Fetta, E. Sukarova-Angelovska, A. Brusco, **L. Pavinato**, V. Pullano, M. Zollino, H. McConkey, M. Tartaglia, G. Ferrero, B. Sadikovic.  
*European Journal of Human Genetics* | 2024-02-13. DOI: 10.1038/s41431-024-01548-4
4. *Missense variants in RPH3A cause defects in excitatory synaptic function and are associated with a clinically variable neurodevelopmental disorder* | **L. Pavinato**, J. Stanic, M. Barzasi, A. Gurgone, G. Chiantia, V. Cipriani, I. Eberini, L. Palazzolo, M. Di Luca, A. Costa, A. Marcantoni, E. Biamino, M. Spada, S. M. Hiatt, W. V. Kelley, L. Vestito, S. M. Sisodiya, Genomics England Research Consortium, S. Efthymiou, P. Chand, R. Kaiyrzhanov, A. Bruxelles, S. Cardaropoli, M. Tartaglia, S. De Rubeis, J. D. Buxbaum, D. Smedley, G. B. Ferrero, M. Giustetto, F. Gardoni, A. Brusco  
*Genetics in Medicine* | 2023-11-06 | journal-article | DOI: 10.1016/j.gim.2023.100922
5. *Spinocerebellar ataxia 38: structure–function analysis shows ELOVL5 G230V is proteotoxic, conformationally altered and a mutational hotspot* | E. Ferrero, E. Di Gregorio, M. Ferrero, E. Ortolan, Y.-A. Moon, A. Di Campli, **L. Pavinato**, C. Mancini, D. Tripathy, M. Manes, E. Hoxha, C. Costanzi, E. Pozzi, M. Rossi Sebastiano, N. Mitro, F. Tempia, D. Caruso, B. Borroni, M. Basso, M. Sallese & A. Brusco  
*Human Genetics* | 2023-08 | journal-article | DOI: 10.1007/s00439-023-02572-y
6. *Skewed X-chromosome inactivation in unsolved neurodevelopmental disease cases can guide re-evaluation For X-linked genes* | C. Giovenino, S. Trajkova, **L. Pavinato**, S. Cardaropoli, V. Pullano, E. Ferrero, E. Sukarova-Angelovska, S. Carestiato, P. Salmin, A. Rinninella, A. Battaglia, L. Bertoli, A. Fadda, F. Palermo, D. Carli, A. Mussa, P. Dimartino, A. Bruxelles, T. Froukh, G. Mandrile, B. Pasini, S. De Rubeis, J. D. Buxbaum, T. Pippucci, M. Tartaglia, M. Rossato, M. Delledonne, G. B. Ferrero & A. Brusco  
*European Journal of Human Genetics* | 2023-03-06 | journal-article | DOI: 10.1038/s41431-023-01324-w
7. *De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes* | M. Scala, N. Drouot, S. C. MacLennan, M. W. Wessels, M. Krygier, **L. Pavinato**, A. Telegrafi, S. A. de Man, M. van Slegtenhorst, M. Iacomino, F. Madia, P. Scudieri, T. Giacomini, G. Nobile, M. M. Mancardi, G. Balagura, G. B. Ferrero, G. Galloni, A. Verrotti, M. Umair, A. Khan, J. Liebelt,

- A. Brusco, B. S. Lipska Ziętkiewicz, J. J. Saris, N. Charlet-Berguerand, F. Zara, P. Striano, A. Piton Human Mutation. 2022-09 | journal-article. <https://doi.org/10.1002/humu.24414>
8. *Rare coding variation illuminates the allelic architecture, risk genes, cellular expression patterns, and phenotypic context of autism* | J. M. Fu, F. K. Satterstrom, M. Peng, H. Brand, R. L. Collins, S. Dong, B. Wamsley, L. Klei, L. Wang, S. P. Hao, C. R. Stevens, C. Cusick, M. Babadi, E. Banks, B. Collins, S. Dodge, S. B. Gabriel, L. Gauthier, S. K. Lee<sup>12</sup>, L. L. A. Ljungdahl, B. Mahjani, L. Sloofman, A. Smirnov, M. Barbosa, C. Betancur, A. Brusco, B. H. Y. Chung, E. H. Cook, M. L. Cuccaro, E. Domenici, G. B. Ferrero, J. J. Gargus, G. E. Herman, I. Hertz-Picciotto, P. Maciel, D. S. Manoach, M. R. Passos Bueno, A. M. Persico, A. Renieri, J. S. Sutcliffe, F. Tassone, E. Trabetti, G. Campos, S. Cardaropoli, D. Carli, M. C.Y. Chan, C. Fallerini, E. Giorgio, A. C. Girard, E. Hansen Kiss, S. Lun Lee, C. Lintas, Y. Ludena, R. Nguyen, **L. Pavinato**, M. Pericak-Vance, I. Pessah, R. Schmidt, M. Smith, C. I. C. Souza, S. Trajkova, J. Y. T. Wang, M. H. C. Yu, The Autism Sequencing Consortium (ASC), Broad Institute Center for Common Disease Genomics (Broad-CCDG), iPSYCH-BROAD Consortium, D. J. Cutler, S. De Rubeis, J. D. Buxbaum, M. J. Daly, B. Devlin, K. Roeder, S. J. Sanders, M. E. Talkowski Nature Genetics | 2022-08-18 | journal-article. <https://doi.org/10.1038/s41588-022-01104-0>
  9. *p140Cap regulates the composition and localization of the NMDAR complex in synaptic lipid rafts* | C. Angelini, A. Morellato, A. Alfieri, **L. Pavinato**, T. Cravero, O. T. Bianciotto, V. Salemme, D. Natalini, G., A. Raspanti, T. Garofalo, D. Valdembrì, G. Serini, A. Marcantoni, A. Becchetti, M. Giustetto, E. Turco, P. Defilippi. The Journal of Neuroscience | 2022-08-11 | journal-article | DOI: 10.1523/JNEUROSCI.1775-21.2022
  10. *CAPRIN1 haploinsufficiency causes a neurodevelopmental disorder with language impairment, ADHD and ASD* | **L. Pavinato**, A. Delle Vedove, D. Carli, M. Ferrero, S. Carestato, J. L. Howe, E. Agolini, D. A. Coviello, I. van de Laar, P. Y. Billie Au, E. Di Gregorio, A. Fabbiani, S. Croci, M. A. Mencarelli, L. P. Bruno, A. Renieri, D. Veltra, C. Sofocleous, L. Faivre, B. Mazel, H. Safraou, A-S. Denommé-Pichon, M. A. van Slegtenhorst, N. Giesbertz, R. H. van Jaarsveld, A. Childers, R. C. Rogers, A. Novelli, S. De Rubeis, J. D. Buxbaum, S. W. Scherer, G. B. Ferrero, B. Wirth, A. Brusco Brain | 2022-07-27 | journal-article. <https://doi.org/10.1093/brain/awac278>
  11. *The Emerging Roles of Long Non-Coding RNAs in Intellectual Disability and Related Neurodevelopmental Disorders* | C. Iaci, L. Prandi, **L. Pavinato**, A. Brusco, M. Maldotti, I. Molineris, S. Oliviero, G. R. Merlo International Journal of Molecular Sciences | 2022-05-30 | review | <https://doi.org/10.3390/ijms23116118>
  12. *Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis.* | **L. Pavinato**, M. Villamor-Payà, M. Sanchiz-Calvo, C. Andreoli, M. Gay, M. Vilaseca, G. Arauz-Garofalo, T. Pippucci, V. Prota, D. Carli, E. Giggio, F. C. Radio, V. Antona, M. Giuffrè, K. Ranguin, C. Colson, S. De Rubeis, P. Dimartino, J. D. Buxbaum, G. B. Ferrero, M. Tartaglia, S. Martinelli, T. H. Stracker, A. Brusco Journal of Medical Genetics. 2022-02 | journal-article. DOI: 10.1136/jmedgenet-2020-107281
  13. *Drosophila functional screening of de novo variants in autism uncovers deleterious variants and facilitates discovery of rare neurodevelopmental diseases.* | P. C. Marcogliese, S.L. Deal, J. Andrews, J. M. Harnish, V. H. Bhavana, H. K. Graves, S. Jangam, X. Luo, N. Liu, D. Bei, Y-H. Chao, B. Hull, P-T Lee, H. Pan, P. Bhadane, M-C Huang, C. M. Longley, H-T Chao, H-I Chung, N. A. Haelterman, O. Kanca, S. N. Manivannan, L. Z. Rossetti, R. J. German, A. Gerard, E. M. C. Schwaibold, S. Fehr, R. Guerrini, A. Vetro, E. England, C. N. Murali, T. S. Barakat, M. F. van Dooren, M. Wilke, M. van Slegtenhorst, G. Lesca, I. Sabatier, N Chatron, C. A. Brownstein, J. A. Madden, P. B. Agrawal, B. Keren, T. Courtin, L. Perrin, M. Brugger, T. Roser, S. Leiz, F. T. Mau-Them, J. Delanne, E. Sukarova-Angelovska, S. Trajkova, E. Rosenhahn, V. Strehlow, K. Platzer, R. Keller, **L. Pavinato**, A. Brusco, J. A. Rosenfeld, R. Marom, M. F. Wangler, S. Yamamoto Cell Reports. 2022-03-15 | journal-article. DOI: 10.1016/j.celrep.2022.110517
  14. *Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy.* | E. Verdura, A. Rodríguez-Palmero, V. Vélez-Santamaria, L. Planas-Serra, I. de la Calle, M. Raspall-Chaure, A. Roubertie, M. Benkirane, F. Saettini, **L. Pavinato**, G. Mandrile, M. O'Leary, E. O'Heir, E. Barredo, A. Chacón, V. Michaud, C. Goizet, M. Ruiz, A. Schlüter, I. Rouvet, J. Sala-Coromina, C. Fossati, M. Iascone, F. Canonico, A. Marcé-Grau, P. de Souza, D. R. Adams, C. Casasnovas, H. L. Rehm, H. C Mefford, L.

- González Gutierrez-Solana, A. Brusco, M. Koenig, A. Macaya, A. Pujol  
Brain | 2021-08-20 | journal-article. DOI: 10.1093/brain/awab124
15. *KCNK18 biallelic variants associated with intellectual disability and autism spectrum disorder alter TRESK channel activity.* | **L. Pavinato**, E. Nematian-Ardestani, A. Zonta, S. De Rubeis, J. D. Buxbaum, C. Mancini, A. Bruselles, M. Tartaglia, M. Pessia, S. J. Tucker, M. C. D'Adamo, A. Brusco  
International Journal of Molecular Sciences. 2021-06-04 | journal-article. DOI: 10.3390/ijms22116064
16. *Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review.* | **L. Pavinato**, S. Trajkova, E. Grosso, E. Giorgio, A. Bruselles, F. C. Radio, T. Pippucci, P. Dimartino, M. Tartaglia, A. Petlichkovski, S. De Rubeis, J. D. Buxbaum, G. B. Ferrero, R. Keller, A. Brusco  
American Journal of Medical Genetics Part A. 2021-03-06 | journal-article. DOI: 10.1002/ajmg.a.62157
17. *New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review.* | S. Trajkova, E. Di Gregorio, G. B. Ferrero, D. Carli, **L. Pavinato**, G. Delplanq, P. Kuentz, A. Brusco  
Brain Sciences | 2020-10-28 | journal-article. DOI: 10.3390/brainsci10110788
18. *Quando il disturbo del neurosviluppo ha un substrato genetico: un caso di sindrome di Kleefstra.* | L. Sorasio, L. Franceschi, **L. Pavinato**, A. Peduto.  
Medico e Bambino, Volume XXIV, Aprile 2021, Numero 4. | caso contributivo. DOI:  
<https://doi.org/10.53126/MEBXXIV114>
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#### PREPRINTS AND SUBMITTED WORKS

1. *CAPRIN1 in neurological disorders; Handbook of the Biology and Pathology of Mental Disorders* | A. Brusco and **L. Pavinato**  
Springer Nature | book chapter | accepted for publication.
  2. *Contribution of autosomal rare and de novo variants to sex differences in autism* | M. Koko, K. Satterstrom, **Autism Sequencing Consortium**, APEX consortium, V. Warriar, H. Martin.  
Preprint: <https://doi.org/10.1101/2024.04.13.24305713>
  3. *Skipping of exon 20 in EP300: A Novel Variant Linked to Rubinstein-Taybi Syndrome with Atypical and Severe Clinical Manifestations* | L. Pavinato, S. Carestiano *et al.* | under review
  4. *A novel systems biology approach for prioritization of whole exome sequencing datasets leads to the discovery of new ASD-associated genes* | V. Remori *et al.* | under review
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