Stanford

Pasqualina Colella

Sr Res Scientist-Basic Life, Pediatrics - Genetics

Bio

BIO

Dr. Pasqualina Colella is a scientist with extensive expertise in cell therapy and gene therapy approaches for the treatment of inherited diseases. She is currently Research Scientist at Stanford University, where she is developing innovative cell therapy approaches to address neuropathic lysosomal storage diseases (LSDs) using hematopoietic stem cell transplantation (HSCT) and genome editing.

Dr. Colella received her summa cum laude Bachelor of Science degree in Biology from the University of Naples Federico II (Italy) and her PhD in Human Genetics from The Open University (UK) at the Telethon Institute of Genetics and Medicine (TIGEM, Italy). During her PhD, she focused on investigating AAV gene therapy for inherited blindness and developed novel strategies to effectively express large (> 5 kb) genes in the retina for treating Stargardt disease and Usher Syndrome type I B, both of which cause untreatable forms of blindness.

After the PhD, Dr. Colella secured prestigious Postdoctoral fellowships from the Marie Sk#odowska-Curie Actions (EU) and Genethon (France). As Postdoc she developed innovative in vivo AAV gene therapy approaches based on liver gene transfer or multi-tissue gene expression to target the multi-organ manifestations of Pompe disease, a LSD that presents with neuromuscular impairment.

Dr. Colella is committed to researching innovative ways to combat genetic diseases and making a positive impact on the lives of patients.

Publications

PUBLICATIONS

- **CNS Repopulation by Hematopoietic-Derived Microglia-Like Cells Corrects Progranulin deficiency.** *Research square*
  Colella, P., Sayana, R., Suarez-Nieto, M. V., Sarno, J., Nyame, K., Xiong, J., Vera, L. N., Basurto, J. A., Corbo, M., Limaye, A., Davis, K. L., Abu-Remaileh, M., Gomez-Ospina, et al
  2023
- **Editorial: Ex-vivo and in-vivo genome engineering for metabolic and neurometabolic diseases.** *Frontiers in genome editing*
  Colella, P., Meneghini, V., Baldo, G., Gomez-Ospina, N.
  2023; 5: 1248904
- **Successful treatment of severe MSUD in Bckdhb-/- mice with neonatal AAV gene therapy.** *Journal of inherited metabolic disease*
  Pontoizeau, C., Gaborit, C., Tual, N., Simon-Sola, M., Rotaru, I., Benoist, M., Colella, P., Lamazière, A., Brassier, A., Arnoux, J. B., Rötig, A., Ottolenghi, C., de Lonlay, et al
  2023
- **Improved engraftment and therapeutic efficacy by human genome-edited hematopoietic stem cells with Busulfan-based myeloablation.** *Molecular therapy. Methods & clinical development*
  Poletto, E., Colella, P., Pimentel Vera, L. N., Khan, S., Tomatsu, S., Baldo, G., Gomez-Ospina, N.

2022; 25: 392-409

- **Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice.** *Nature communications*
  Pontoizeau, C., Simon-Sola, M., Gaborit, C., Nguyen, V., Rotaru, I., Tual, N., Colella, P., Girard, M., Biferi, M. G., Arnoux, J. B., Rötig, A., Ottolenghi, C., de Lonlay, et al
  2022; 13 (1): 3278

- **Hepatic expression of GAA results in enhanced enzyme bioavailability in mice and non-human primates.** *Nature communications*
  Costa-Verdera, H., Collaud, F., Riling, C. R., Sellier, P., Nordin, J. M., Preston, G. M., Cagin, U., Fabregue, J., Barral, S., Moya-Nilges, M., Krijnse-Locker, J., van Wittenberghe, L., Daniele, et al
  2021; 12 (1): 6393

- **Gene therapy with secreted acid alpha-glucosidase rescues Pompe disease in a novel mouse model with early-onset spinal cord and respiratory defects.** *EBioMedicine*
  Colella, P., Sellier, P., Gomez, M. J., Biferi, M. G., Tanniou, G., Guerchet, N., Cohen-Tannoudji, M., Moya-Nilges, M., van Wittenberghe, L., Daniele, N., Gjata, B., Krijnse-Locker, J., Collaud, et al
  2020; 61: 103052

- **Rescue of Advanced Pompe Disease in Mice with Hepatic Expression of Secretable Acid #-Glucosidase.** *Molecular therapy : the journal of the American Society of Gene Therapy*
  Cagin, U., Puzzo, F., Gomez, M. J., Moya-Nilges, M., Sellier, P., Abad, C., Van Wittenberghe, L., Daniele, N., Guerchet, N., Gjata, B., Collaud, F., Charles, S., Sola, et al
  2020; 28 (9): 2056-2072

- **Engineering monocyte/macrophage-specific glucocerebrosidase expression in human hematopoietic stem cells using genome editing** *Nature Communications*
  Scharenberg, S. G., Poletto, E., Lucot, K. L., Colella, P., Sheikali, A., Montine, T. J., Porteus, M. H., Gomez-Ospina, N.
  2020; 11: 1-14

- **Role of Regulatory T Cell and Effector T Cell Exhaustion in Liver-Mediated Transgene Tolerance in Muscle.** *Molecular therapy. Methods & clinical development*
  Poupiot, J., Costa Verdera, H., Hardet, R., Colella, P., Collaud, F., Bartolo, L., Davoust, J., Sanatine, P., Mingozzi, F., Richard, I., Ronzitti, G.
  2019; 15: 83-100

- **Gene Therapy for Pompe Disease: The Time is now.** *Human gene therapy*
  Colella, P., Mingozzi, F.
  2019; 30 (10): 1245-1262

- **Dual muscle-liver transduction imposes immune tolerance for muscle transgene engraftment despite preexisting immunity.** *JCI insight*
  Bartolo, L., Li Chung Tong, S., Chappert, P., Urbain, D., Collaud, F., Colella, P., Richard, I., Ronzitti, G., Demengeot, J., Gross, D. A., Mingozzi, F., Davoust, J.
  2019; 4 (11)

- **AAV Gene Transfer with Tandem Promoter Design Prevents Anti-transgene Immunity and Provides Persistent Efficacy in Neonate Pompe Mice** *MOLECULAR THERAPY-METHODS & CLINICAL DEVELOPMENT*
  Colella, P., Sellier, P., Verdera, H., Puzzo, F., van Wittenberghe, L., Guerchet, N., Daniele, N., Gjata, B., Marmier, S., Charles, S., Sola, M., Ragone, I., Leborgne, et al
  2019; 12: 85–101

- **Emerging Issues in AAV-Mediated In Vivo Gene Therapy.** *Molecular therapy. Methods & clinical development*
  Colella, P., Ronzitti, G., Mingozzi, F.
  2018; 8: 87-104

- **Rescue of GSDIII Phenotype with Gene Transfer Requires Liver- and Muscle-Targeted GDE Expression** *MOLECULAR THERAPY*
  Vidal, P., Pagliarani, S., Colella, P., Verdera, H., Jauze, L., Gjorgjieva, M., Puzzo, F., Marmier, S., Collaud, F., Sola, M., Charles, S., Lucchiari, S., van Wittenberghe, et al
  2018; 26 (3): 890–901

- **Rescue of Pompe disease in mice by AAV-mediated liver delivery of secretable acid alpha-glucosidase** *SCIENCE TRANSLATIONAL MEDICINE*
  Puzzo, F., Colella, P., Biferi, M. G., Bali, D., Paulk, N. K., Vidal, P., Collaud, F., Simon-Sola, M., Charles, S., Hardet, R., Leborgne, C., Meliani, A., Cohen-Tannoudji, et al
  2017; 9 (418)

- **Transposon-mediated Generation of Cellular and Mouse Models of Splicing Mutations to Assess the Efficacy of snRNA-based Therapeutics.** *Molecular therapy. Nucleic acids*
  Barbon, E., Ferrarese, M., van Wittenberghe, L., Sanatine, P., Ronzitti, G., Collaud, F., Colella, P., Pinotti, M., Mingozzi, F.
  2016; 5 (11): e392
- **Long-term exposure to Myozyme results in a decrease of anti-drug antibodies in late-onset Pompe disease patients.** *Scientific reports*
  Masat, E., Laforêt, P., De Antonio, M., Corre, G., Perniconi, B., Taouagh, N., Mariampillai, K., Amelin, D., Mauhin, W., Hogrel, J. Y., Caillaud, C., Ronzitti, G., Puzzo, et al
  2016; 6: 36182
- **Improved dual AAV vectors with reduced expression of truncated proteins are safe and effective in the retina of a mouse model of Stargardt disease.** *Human molecular genetics*
  Trapani, I., Toriello, E., de Simone, S., Colella, P., Iodice, C., Polishchuk, E. V., Sommella, A., Colecchi, L., Rossi, S., Simonelli, F., Giunti, M., Bacci, M. L., Polishchuk, et al
  2015; 24 (23): 6811-25
- **Efficient gene delivery to the cone-enriched pig retina by dual AAV vectors.** *Gene therapy*
  Colella, P., Trapani, I., Cesi, G., Sommella, A., Manfredi, A., Puppo, A., Iodice, C., Rossi, S., Simonelli, F., Giunti, M., Bacci, M. L., Auricchio, A.
  2014; 21 (4): 450-6
- **Effective delivery of large genes to the retina by dual AAV vectors.** *EMBO molecular medicine*
  Trapani, I., Colella, P., Sommella, A., Iodice, C., Cesi, G., de Simone, S., Marrocco, E., Rossi, S., Giunti, M., Palfi, A., Farrar, G. J., Polishchuk, R., Auricchio, et al
  2014; 6 (2): 194-211
- **Myosin7a deficiency results in reduced retinal activity which is improved by gene therapy.** *PloS one*
  Colella, P., Sommella, A., Marrocco, E., Di Vicino, U., Polishchuk, E., Garcia Garrido, M., Seeliger, M. W., Polishchuk, R., Auricchio, A.
  2013; 8 (8): e72027
- **Photoreceptor degeneration in mice: adeno-associated viral vector-mediated delivery of erythropoietin.** *Methods in molecular biology (Clifton, N.J.)*
  Colella, P., Auricchio, A.
  2013; 982: 237-63
- **Gene therapy of inherited retinopathies: a long and successful road from viral vectors to patients.** *Human gene therapy*
  Colella, P., Auricchio, A.
  2012; 23 (8): 796-807
- **TFEB links autophagy to lysosomal biogenesis.** *Science (New York, N.Y.)*
  Settembre, C., Di Malta, C., Polito, V. A., Garcia Arencibia, M., Vetrini, F., Erdin, S., Erdin, S. U., Huynh, T., Medina, D., Colella, P., Sardiello, M., Rubinsztein, D. C., Ballabio, et al
  2011; 332 (6036): 1429-33
- **Non-erythropoietic erythropoietin derivatives protect from light-induced and genetic photoreceptor degeneration.** *Human molecular genetics*
  Colella, P., Iodice, C., Di Vicino, U., Annunziata, I., Surace, E. M., Auricchio, A.
  2011; 20 (11): 2251-62
- **AAV-mediated gene supply for treatment of degenerative and neovascular retinal diseases.** *Current gene therapy*
  Colella, P., Auricchio, A.
  2010; 10 (5): 371-80
- **Ocular gene therapy: current progress and future prospects.** *Trends in molecular medicine*
  Colella, P., Cotugno, G., Auricchio, A.
  2009; 15 (1): 23-31
- **AP20187-mediated activation of a chimeric insulin receptor results in insulin-like actions in skeletal muscle and liver of diabetic mice.** *Human gene therapy*
  Cotugno, G., Formisano, P., Giacco, F., Colella, P., Beguinot, F., Auricchio, A.
  2007; 18 (2): 106-17