Stéphanie CHERQUI, Ph.D

Business Address University of California, San Diego
Department of Pediatrics
Division of Genetics
9500 Gilman Drive, MC 0734
La Jolla, California 92093-0734, USA
Telephone (858) 822-1023
Fax (858) 246 1125
E-mail address scherqui@ucsd.edu
Website https://healthsciences.ucsd.edu/som/pediatrics/research/labs/cherqui/Pages/default.aspx
Citizenships: French, USA

UNIVERSITY EDUCATION

1998 - 2002 **Ph.D. in Human Genetics, University of René Descartes, Paris, France.**
Inserm Unit 983, Necker-Enfants Malades Hospital

1997 - 1998 **Master's degree in Molecular Genetics of Developmental and Oncogenic Diseases**
University of René Descartes, Paris, France.

1993 - 1997 **Bachelor of Science degree in Molecular Biology and Genetics**
University of Denis Diderot, Paris, France.

RESEARCH ACTIVITY

2016 - present **Associate Professor | University of California, San Diego (UCSD), Department of Pediatrics, Division of Genetics, La Jolla, California, USA**
Research projects Developing stem cell and gene therapy strategies for degenerative multi-systemic disorders metabolic and neurodegenerative disorders, and understand the molecular and cellular mechanisms by which hematopoietic stem cell progeny can lead to tissue repair in non-hematopoietic genetic diseases.

2012 - 2016 **Assistant Professor | University of California, San Diego (UCSD), Department of Pediatrics, Division of Genetics, La Jolla, California, USA**
Research projects Developing stem cell and gene therapy strategies for degenerative multi-systemic disorders such as cystinosis, and understand the molecular and cellular mechanisms by which hematopoietic stem cell progeny can lead to tissue repair in non-hematopoietic genetic diseases.

2009 - 2012 **Assistant Professor | The Scripps Research Institute (TSRI), Department of Molecular and Experimental Medicine, La Jolla, California, USA**
Research Project Stem cell and Gene Therapy for Cystinosis.

2006 - 2009 **Staff scientist | TSRI, Department of Molecular and Experimental Medicine, La Jolla, California, USA**
Research Project Treatment of cystinosis nephropathy using genetically modified adult stem cells in murine cystinosis model.

2002 - 2006 **Research Associate | TSRI, Department of Molecular and Experimental Medicine, California, USA**
Project entitled The use of endothelial progenitors and proangiogenic growth factors to enhance angiogenesis.
Supervisor Daniel R. Salomon, M.D

1997 - 2002 **Ph.D, Inserm U983, Hospital Necker-Enfants Malades, Paris, France**
Thesis entitled "Characterization of cystinosin, the protein encoded by CTNS, the gene underlying nephropatic cystinosis, and generation of a mouse model".
Supervisor Corinne Antignac, M.D, Ph.D

POSITIONS and HONOR

Positions

2009 - 2012 Assistant Professor, *Tenure-Track*, The Scripps Research Institute, Department of Molecular and Experimental Medicine, La Jolla, California, USA

2012 - 2016 Assistant Professor, *In Residence*, University of California, San Diego, Department of Genetics, Division of Genetics, La Jolla, California, USA

2016 -present Associate Professor, *In Residence*, University of California, San Diego, Department of Genetics, Division of Genetics, La Jolla, California, USA

Major Committee Assignments:

2010 - present Scientific Review Board member of the Cystinosis Research Foundation

2010 - present Cure Cystinosis International Registry (CCIR) Scientific Council member

2011 - present Member of the Board of Trustees of the Cystinosis Research Foundatio

2014 - present UCSD Biomedical Sciences Graduate Program Admissions Committee member

2014 - present Chair of the Cystinosis Stem Cell and Gene Therapy Consortium

2011 - 2017 American Society of Gene and Cell Therapy (ASGCT) Gene & Cell Therapy of Genetic and Metabolic Diseases Committee member

2017 – present Chair of American Society of Gene and Cell Therapy (ASGCT) Gene & Cell Therapy of Genetic and Metabolic Diseases Committee

2018 - present Member of the University of California, San Diego Human Gene Transfer/Human Gene Editing Institutional Biosafety Committee (UCSD HGT/HGE IBC)

Honors and Fellowships:

Prize of the Association for Information and Research on Genetic Renal Diseases
Awarded October 3th 2001, Montpellier, France.

Prize of the Philippe Foundation
Awarded February 10th 2003, New York, USA.

September 1998- September 2001:

Graduate student fellowship from the Minister of National Education, Research and Technology (France).

September 2001- September 2002:
Graduate student fellowship from Vaincre les Maladies Lysosomales Foundation (France).

September 2001- September 2002:
Post-doctoral fellowship from Inserm (USA).

October 2003- October 2005:
Post-doctoral fellowship from Juvenile Diabetes Research Foundation (USA).

Assembly Resolution from the California Legislature Assembly for Dr. Cherqui's Research on Cystinosis
Awarded October 5th 2012, Sacramento, California.

Rady Children's Hospital and UCSD Physician of Excellence Award for your Basic Science Research
2018 Awards of Excellence Honoree

REVIEWER

Journals
- Nature Communications
- EMBO Molecular Medicine
- Stem Cells
- Human Molecular Genetics
- Journal of the American Society of Nephrology
- Kidney International
- FEBS journal
- Pediatric Nephrology
- Pediatric Research
- Cell transplantation
- Journal of inherited Metabolic Disease
- BMC Nephrology
- Clinical Pharmacology in Drug Development
- American Journal of Kidney Diseases
- Biotechnology journal
- Biochemistry and Biophysics Reports

Grants
- *Ad hoc* reviewer for NIH Therapeutic Approaches to Genetic Diseases (TAG) Study Section
- *Ad hoc* reviewer for NIH National Center for Advancing Translational Sciences (NCATS)
- Cystinosis Research Foundation
- Cystinosis Ireland

Conference
- American Society of Gene and Cell Therapy (ASGCT)

TEACHING

2010 - 2015	*Ad hoc* Lecturer for the Graduate Program at the Scripps Research Institute.
2013 - present	Lecturer for the Medical Genetics didactic course, University of California San Diego
2013 - present	Genetics in Medicine courses, University of California San Diego

2013 - present	Faculty member of the UCSD Biomedical Sciences Graduate Program
2014 - present	Faculty member of the UCSD Genetics Training Program
2016 - present	Lecturer for the Path225/BIOMED227 Seminars/Molecular Pathology course, BISP170 Seminars series, CMM250 Core Course in Stem Cell Biology, Medicine and Ethics, UCSD MD/PhD program seminar series.
2016 - present	Chair of MED271 course - Gene Therapy: Principles and Clinical Applications

INVITED LECTURES

Hematopoietic Stem Cell Gene Therapy for Cystinosis: Clinical Translation and Mechanism of Action. **Western Society of Pediatric Nephrology**; Rady Children's Hospital, San Diego, CA. January 26, 2019.

From Cystinosis to Friedreich's ataxia: Therapeutic Mechanism and Clinical Translation of Hematopoietic Stem Cell Gene Therapy. **Stem Cell Institute, University of Minnesota**; Minneapolis, MN. January 10, 2019. Seminar

Clinical Translation of Hematopoietic Stem Cell Gene Therapy for Cystinosis. **Cell and Gene Meeting on the Mesa**; San Diego California. October 5, 2018. Chair of the Session "Ex vivo Gene Therapy: Using Blood Stem Cells to treat Genetic Disorders".

Involvement of tunneling nanotubes for paracrine tissue preservation after hematopoietic stem cell transplantation: model of the cornea in cystinosis. **Alcon Research Institute Podos Colloquium**; Honolulu, Hawaii. April 28, 2018

Stem Cell Gene Therapy for Cystinosis: Update and Description of the Upcoming Clinical Trial. **Cystinosis Research Foundation Family Conference**; Newport Beach, California. April 20, 2018.

From Cystinosis to Friedreich's ataxia: Therapeutic Mechanism and Clinical Translation of Hematopoietic Stem Cell Gene Therapy. **Division of Regenerative Medicine Seminar Series, UCSD**. April 9, 2018. Seminar

Hematopoietic Stem Cell Gene Therapy for Cystinosis: Clinical Translation, Mechanism of Action and Application to Mitochondrial Diseases. **President's Lecture Series, LA BioMed**; Los Angeles, California. April 5, 2018. Seminar.

Toxicology, Manufacturing and Clinical description of the stem cell gene therapy product for cystinosis. March 1-2, 2018. **Sixth International CRF symposium**; Newport Beach, California. Co-chair and Organizer of the symposium.

From Cystinosis to Friedreich's ataxia: Therapeutic Mechanism and Clinical Translation of Hematopoietic Stem Cell Gene Therapy. **Friedreich's Ataxia Patient Symposium**, organized by Friedreich's ataxia Research Alliance (FARA) and UCLA; Torrance, Los Angeles, California. February 23, 2018.

Therapeutic mechanism and clinical translation of hematopoietic stem cell gene therapy for cystinosis and Friedreich's ataxia. **The Center for Cellular and Molecular Therapeutics** (CCMT) at the Children's Hospital of Philadelphia and University of Pennsylvania; Philadelphia, Pennsylvania. February 1, 2018. Seminar.

Involvement of tunneling nanotubes in tissue preservation after hematopoietic stem cell transplantation in degenerative genetic disorders. **ASCB/EMBO Meeting**; Philadelphia, Pennsylvania. December 2, 2017.

Hematopoietic Stem Cell Gene Therapy for Cystinosis: Clinical Translation, Mechanism of Action and Application to Mitochondrial Diseases. **Scintillon Institute**; San Diego, CA. September 8, 2017. Seminar.

Hematopoietic Stem Cell Gene Therapy for Cystinosis: Mechanism of Action and Clinical Translation. **2017 CIRM Bridges Annual Meeting**; Coronado, California. July 25, 2017

Stem Cell Gene Therapy for Cystinosis: Description of the Upcoming Clinical Trial. **Cystinosis Research Network conference**; Snowbird, Utah. July 15, 2017.

Role of phagocytic cells in tissue repair: model of lysosomal and mitochondrial genetic disorders. **2017 Phagocytes Gordon Research Conference**; Waterville Valley, New Hampshire. June 11-16, 2017.

Stem Cell Therapy in Cystinosis. **Pediatric Academic Societies Meeting**; San Francisco, California. May 6, 2017.

Hematopoietic Stem Cell Gene Therapy for the Lysosomal Storage Disorder Cystinosis: Clinical Translation, Mechanism of Action and Application to Mitochondrial Diseases. **Center for Genetic and Genomic Therapies**; UC San Diego, California. April 18, 2017. Seminar

Stem Cell Gene Therapy for Cystinosis: Description of the Upcoming Clinical Trial. **Cystinosis Research Foundation conference**; Newport Beach, California. March 31, 2017.

Hematopoietic Stem Cell Gene Therapy for the Lysosomal Storage Disorder Cystinosis: Clinical Translation and Application to Mitochondrial Diseases. **8th Annual Pediatric Research Symposium**; UC San Diego, California. January 13, 2017.

Gene and cell therapies: How far is the cure? Septembre, 2016. **17th Congress of the International Pediatric Nephrology Association** (IPNA); Iguaçu, Brazil.

Cellules souches et thérapie génique pour la cystinose: a l'approche d'un essai Clinique. April 14, 2016. **Néphrologie Pédiatrique Conférence**; Imagine Institute, Paris France.

Towards the Clinical Application of Stem Cell and Gene Therapy for Cystinosis. March 3-4, 2016. **Fourth International CRF symposium**; Newport Beach, California. Co-chair and Organizer of the symposium.

Animal models and gene therapy for Cystinosis. **American Society of Nephrology conference**; San Diego, California. November 4, 2015. ASN Engineering Genomes to Model Disease, Target Mutations, and Personalize Therapy Course.

Towards the clinical application of lentiviral hematopoietic stem cell gene therapy for cystinosis. **Stem Cell meeting on the Mesa's Scientific Symposium**; La Jolla, California. October 9, 2015

Hematopoietic stem cell gene therapy for cystinosis: new mechanisms of stem cell-mediated tissue repair and clinical application. **SoCal Stem Cell Consortium**; La Jolla, California. September 10, 2015. Seminar.

Towards the Clinical Application of an Emerging Model of Hematopoietic Stem Cell-Mediated Tissue Repair to Treat the Multi-Systemic Disorder Cystinosis. **Sanford Consortium for Regenerative Medicine Research Symposium**; La Jolla, California. May 19, 2015.

New Therapies on the Horizon: Gene Modified Auto Stem Cell Transplantation. **American Transplant Congress**; Philadelphia, Pennsylvania. May 3, 2015.

MISSION ~~IM~~POSSIBLE: Hematopoietic stem cell-mediated tissue repair: Rescue of the kidney and the eye in the cystinosis model. **UCSD Pediatrics Faculty Research Seminar Series**; La Jolla, California. April 2014. Seminar.

Hematopoietic stem cell transplantation for cystinosis: evidence of lysosomal cross-correction. **Gordon Research Conference on Lysosomal Diseases**; Galveston, Texas. March 16, 2015.

Emerging Technologies. **1st Annual Rare Disease Conference**; University of California, Davis, California. March 5, 2015. Panelist.

Hematopoietic Stem cell and Gene Therapy for Cystinosis. **American Society of Nephrology conference**; Philadelphia, Pennsylvania. November 12, 2014. ASN Human Genetics in Nephrology Course.

Hematopoietic Stem Cell Transplantation and Gene Therapy for Cystinosis. **8th International Cystinosis Congress**; Manchester, United Kingdom. July 25, 2014.

Stem cell and AAV-mediated gene therapies: two different approaches for the treatment of the lysosomal storage disorder Cystinosis. **GENETHON**; Paris, France. July 23, 2014. Seminar.

Hematopoietic stem cell and gene therapy for cystinosis: new mechanisms of stem cell-mediated tissue repair and implications for multi-systemic disease treatment. **Albert Einstein College of Medicine**; New York, New York. April 28, 2014. Seminar.

Stem Cell and Gene Therapy for Cystinosis: Update and Mechanism Studies. **Third International CRF symposium**; Newport Beach, California. March 6-7, 2014. Co-chair and Organizer of the symposium.

Hematopoietic stem cell gene therapy for cystinosis: a new hope for a multi-systemic degenerative disorder. **10th World Symposium on Lysosomal Diseases**; San Diego, California. February 12, 2014.

Hematopoietic stem cell gene therapy for cystinosis: a new hope for a multi-systemic degenerative disorder. **Pediatric Grand Rounds**, November 15, 2013, *and* **Pathology Research Lecture Series**, January 13, 2014. University of California San Diego, La Jolla, CA. Seminars.

Cell replacement strategies in nephropathic cystinosis. **6th Congress of the International Pediatric Nephrology Association**; Shangai, China. September 1st, 2013. Symposium.

Gene therapy in cystinosis. **Cystinosis Research Network Family Conference**; Washington, District of Columbia. July 2013. Seminar.

Cystinosis: from the gene to a potential therapy. **Augustana College**; Sioux falls, South Dakota. April 8, 2013. Seminar.

Stem cell transfer of cystinosin. **ACGM Annual Clinical Genetics Meeting**; Phoenix, Arizona. March 20, 2013. Symposium.

Approaches to the Genetic Rescue of Cystinosis. **American Society of Nephrology conference**; San Diego, California. November 4, 2012. Symposium.

Stem Cell and Gene Therapy-based Strategies for the Lysosomal Storage Disorder Cystinosis. **The Center for Cellular and Molecular Therapeutics** (CCMT) at the Children's Hospital of Philadelphia and University of Pennsylvania; Philadelphia, Pennsylvania. May 2012. Seminar.

HSC Gene Therapy for Cystinosis: New Hope for a Multi-Compartment Lysosomal Storage Disorder. **15th Annual Meeting of the American Society of Gene and Cell Therapy**; Philadelphia, Pennsylvania. May 2012. Scientific Symposium: Towards Clinical Trials for Genetic and Metabolic Diseases.

Stem Cell and Gene Therapy for Cytinosis: Update and Mechanism studies. **Second International CRF symposium**; Newport Beach, California. March 2012. Co-chair and Organizer of the symposium.

Stem cell and gene therapy for cystinosis. **IX Congress of ALANEPE** (Latin American Congress of Pediatric Nephrology); São Paulo, Brasil. October 2011.

The CRF Cystinosis Gene Therapy Consortium. **The Cystinosis Research Network Conference**; San Francisco, California. July 2011.

Impact of bone marrow cell transplantation on nephropathy in the mouse model for cystinosis**.** **Western Society of Pediatric Nephrology Conference**; San Diego, California. February 2011. Plenary presentation.

Bone marrow transplantation for cystinosis. **The 6th International Cystinosis Conference**; Lignano Sabbiadoro, Italy. September 2010. Plenary presentation.

Impact of bone marrow cell transplantation on nephropathy in the mouse model for cystinosis. **The 11th International Workshop on Developmental Nephrology**; New Paltz, New York. August 2010.

Stem cell and gene therapy for cystinosis. **International Cystinosis Research Symposium**; Irvine, California. April, 2010.

Impact of bone marrow cell transplantation on nephropathy in the mouse model for cystinosis. **Pediatric Nephrology seminar**; Necker-Enfants Malades hospital, Paris, France. March, 2010.

Cystinosis: from the gene to a potential therapy. **Canadian Association of Pediatric Nephrologists (CAPN) Scientific and Educational Forum** during the American Society of Nephrology Annual Conference; San Diego, California. October, 2010. Plenary presentation.

Successful treatment of the murine model of cystinosis using bone marrow cell transplantation. **National Institute of Health seminar**; Bethesda, Maryland. September 2009. Seminar.

Successful treatment of the murine model of cystinosis using bone marrow cell transplantation. **Cystinosis Research Network Conference**; Stone Mountain, Georgia. July 2009.

Stem cell-based therapy for the treatment of cystinosis in the murine model. **International Cystinosis Research Symposium**; Irvine, California. April 2008.

PUBLICATIONS

Lobry T, Miller R, Nevo N, Rocca CJ, Zhang J, Catz SD, Moore F, Thomas L, Pouly D, Bailleux A, Guerrera IC, Gubler MC, Wilson CW, Mak RH, Montier T, Antignac C, **Cherqui S.** (2019) Interaction between galectin-3 and cystinosin reveals a role of inflammation in kidney pathogenesis in cystinosis. *Kidney International*. [Epub ahead of print].

Rocca CJ and **Cherqui S.** (2018) Potential use of stem cells as a therapy for cystinosis. *Ped Nephrol.* Review. [Epub ahead of print]. PMID: 29789935

Rocca CJ, Goodman SM, Dulin JN, Haquang JH, Gertsman I, Blondelle J, Smith JLM, Heyser CJ, **Cherqui S**. (2017) Hematopoietic stem cell transplantation prevents development of Friedreich's Ataxia in a humanized mouse model. *Sci Transl Med*. 9(413). PMID: 29070698

Ariazi J, Benowitz A, De Biasi V, Den Boer ML, **Cherqui S**, Cui H, Douillet N, Eugenin EA, Favre D, Goodman G, Gousset K, Hanein D, Israel DI, Kimura S, Kirkpatrick KB, Kuhn N, Jeong C, Lou E, Mailliard R, Maio S, Okafo G, Osswald M, Pasquier J, Polak R, Pradel G, de Rooij B, Schaeffer P, Skeberdis VA, Smith IF, Tanveer A, Volkmann N, Wu Z, and Zurzolo C. (2017) Tunneling Nanotubes and Gap Junctions–Their Role in Long-Range Intercellular Communication during Development, Health, and Disease Conditions. *Front Mol Neurosci*. Review. [Epub ahead of print] PMID: 29089870

Zhang J, Johnson JL, He J, Napolitano G, Ramadass M, Rocca C, Kiosses WB, Bucci C, Xin Q, Gavathiotis E, Cuervo AM, **Cherqui S**, Catz SD. (2017) Cystinosin, the small GTPase Rab11, and the Rab7 effector RILP regulate intracellular trafficking of the chaperone-mediated autophagy receptor LAMP2A. *J Biol Chem*. 292(25): 10328-10346. PMID: 28465352

Zhang J, Johnson JL, He J, Napolitano G, Ramadass M, Rocca CJ, Kiosses WB, Bucci C, Xin Q, Gavathiotis E, Cuervo AM, **Cherqui S**, Catz SD. (2017) Cystinosin, the small GTPase Rab11, and the Rab7 effector RILP regulate intracellular trafficking of the chaperone-mediated autophagy receptor LAMP2A. *J Biol Chem*. Jun 23;292(25):10328-10346. PMID: 28465352

Cherqui S, Courtoy PJ. (2017) The renal Fanconi syndrome in cystinosis: pathogenic insights and therapeutic perspectives. *Nat Rev Nephrol*. 13(2):115-131. Review. PMID: 27990015

Gaide Chevronnay HP, Jansen V, Van Der Smissen P, Rocca CJ, Liao XH, Refetoff S, Pierreux CE, **Cherqui S***, and Courtoy P*. (2016) Hematopoietic stem cell transplantation can normalize thyroid function in a cystinosis mouse model. *Endocrinology.* 57(4):1363-1371. PMID: 26812160 *co-senior author

Cheung WW, **Cherqui S**, Ding W, Esparza M, Zhou P, Shao J, Lieber RL, Mak RH. (2016) Muscle wasting and adipose tissue browning in infantile nephropathic cystinosis. *J Cachexia Sarcopenia Muscle*. 7(2):152-64. PMID: 27493869

Gaide Chevronnay HP, Janssens V, Van Der Smissen P, Liao, Y. Abid XH, Nevo N, Antignac C, Refetoff S, **Cherqui S**, Pierreux CE, Courtoy PJ. (2015) A mouse model suggests two mechanisms for thyroid alterations in infantile cystinosis: decreased thyroglobulin synthesis due to endoplasmic reticulum stress/unfolded protein response and impaired lysosomal processing. *Endocrinology*. 156(6):2349-2364. PMID: 25811319

Rocca CJ, Kreymerman A, Ur SN, Frizzi KE, Naphade S, Lau AJ, Tran T, Calcutt NA, Goldberg JL, **Cherqui S**. (2015) Treatment of inherited eye defects by systemic hematopoietic stem cell transplantation*. Invest Ophthalmol Vis Sci.* 56(12):7214-7223. PMID: 26540660

Napolitano G, Johnson JL, He J, Rocca CJ, Monfregola J, Pestonjamasp K, **Cherqui S**, Catz SD. (2015) Impairment of chaperone-mediated autophagy leads to selective lysosomal degradation defects in the lysosomal storage disease cystinosis. *EMBO Mol Med*. 7(2): 158-174. PMID: 25586965

Naphade S, Sharma J, Gaide Chevronnay HP, Shook MA, Yeagy BA, Rocca CJ, Ur SN, Lau AJ, Courtoy PJ, **Cherqui S**. (2015) Lysosomal cross-correction by hematopoietic stem cell-derived macrophages via tunneling nanotubes. *Stem Cells*. 33(1):301-309. PMID: 25186209

Emma F, Nesterova G, Langman C, Labbé A, **Cherqui S**, Goodyer P, Janssen MC, Greco M, Topaloglu R, Elenberg E, Dohil R, Trauner D, Antignac C, Cochat P, Kaskel F, Servais A, Wühl E, Niaudet P, Van't Hoff W, Gahl W, Levtchenko E. (2014) Nephropathic cystinosis: an international consensus document. *Nephrol Dial Transplant*. Review. 29 Suppl 4:iv87-94. PMID: 25165189

Rocca CJ, Ur SN, Harrison F, **Cherqui S**. (2014) rAAV9 combined with renal vein injection is optimal for kidney-targeted gene delivery: conclusion of a comparative study. *Gene Ther*. 21(6):618-628. PMID: 24784447

Prencipe G, Caiello I, **Cherqui S**, Whisenant T, Petrini S, Emma F and De Benedetti F. (2014) Cystine crystals are an inflammasome activating danger signal: possible implications for the pathogenesis of cystinosis. *J Am Soc Nephrol*. 25(6):1163-1169. PMID: 24525029

Chevronnay HP, Janssens V, Van Der Smissen P, N'Kuli F, Nevo N, Guiot Y, Levtchenko E, Marbaix E, Pierreux CE, **Cherqui S**, Antignac C and Courtoy PJ. (2014) Time-course of pathogenic and adaptive mechanisms in cystinotic mice kidneys. *J Am Soc Nephrol*. 25(6):1256-1269. PMID: 24525030

Cherqui S. (2014) Is Genetic Rescue of Cystinosis an Achievable Treatment Goal? *Nephrol Dial Transplant.* Review. 29(3):522-528. PMID: 23861466

Johnson JL, Napolitano G, Monfregola J, Rocca CJ, **Cherqui S** and Catz SD. (2013) Upregulation of the Rab27a-dependent trafficking and secretory mechanisms improves lysosomal transport, alleviates endoplasmic reticulum stress and reduces lysosome overload in cystinosis. *Mol Cell Biol*. 33(15):2950-2962. PMID: 23716592

Harrison F, Yeagy BA, Rocca CJ, Kohn DB, Salomon DR, **Cherqui S**. (2013) Hematopoietic stem cell gene therapy in the mouse model of cystinosis. Molecular Therapy. *Mol Ther*. 21(2):433-444. PMID: 23089735

Cherqui S. (2012) Cysteamine therapy: a treatment for cystinosis, not a cure. *Kidney Intern*. 81(2):127-129. Commentary article. PMID: 22205430

Simpson J, Nien CJ, Flynn K, Jester B, **Cherqui S**, Jester J. (2011) Quantitative in vivo and ex vivo confocal microscopy analysis of corneal cystine crystals in the Ctns knockout mouse. *Mol Vis*. 17:2212-20. PMID: 21897743

Taniguchi N, Carames B, Hsu E, **Cherqui S**, Kawakami Y, Lotz M. (2011) Expression patterns and function of chromatin protein HMGB2 during Mesenchymal stem cell differentiation. *J Biol Chem*. 286(48):41489-41498. PMID: 21890638

Yeagy BA and **Cherqui S.** (2011) Kidney repair and stem cells: a complex and controversial process. *Pediatr Nephrol*. 26(9):1427-1434. Review. PMID: 21336814

Yeagy BA, Harrison F, Gubler M.C, Koziol JA, Salomon DR, **Cherqui S.** (2011) Kidney preservation by bone marrow cell transplantation depends on the level of stem cell engraftment in hereditary nephropathy. *Kidney Intern*. 79(11):1198-1206. **Cover photo**. PMID: 21248718 [Featured in: Pinkernell K. (2011) Cellular therapies: what is still missing? *Kidney International*. 79(11):1161-1163.]

Syres K, Harrison F, Tadlock M, Jester J, Simpson J, Roy S, Salomon DR, **Cherqui S.** (2009) Successful treatment of the mouse model of cystinosis using bone marrow cell transplantation. *Blood*. 114:2530-2541. **Cover photo**. PMID: 19506297 [Featured in: Terryn S, Devuyst O, Antignac C. (2010) Cell therapy for cystinosis. *Nephrol Dial Transplant*. 25(4):1059-1066.]

Cherqui S, Kingdon KM, Thorpe C, Kurian SM, Salomon DR. (2007) Lentiviral gene delivery of vMIP-II via both transplanted mature endothelial and progenitor cells enhances angiogenesis *in vivo*. *Mol Ther*. 15(7):1264-1272. PMID: 17505479

Martina Y, Marcucci KT, **Cherqui S**, Szabo A, Drysdale T, Srinivisan U, Wilson CA, Patience C, Salomon D.R. (2006) Mice transgenic for a human porcine endogenous retrovirus receptor are susceptible to productive viral infection. *J Virol*. 80(7):3135-3146. PMID: 16537582

Cherqui S, Kurian S.M, Schussler O, Hewel J.A, Yates J.R, Salomon DR. (2005). Isolation and angiogenesis by endothelial progenitors in fetal liver. *Stem cells*. 24(1):44-54. PMID: 16099996

Martina Y, Kurian S, **Cherqui S**, Evanoff G, Wilson C, Salomon DR. (2005) Pseudotyping of porcine endogenous retrovirus by xenotropic murine leukemia virus in a pig islet xenotransplantation model. *American Journal of Transplantation*. 5(8):1837-1847. PMID: 15996230

Chol M, Nevo N, **Cherqui S**, Antignac C, Rusrin P. (2004) Glutathione precursors replenish decreased glutathione pool in cystinotic cell lines. *Biochem Biophys Res Commun*. 324(1):231-235. PMID: 15465007

Kalatzis V, **Cherqui S**, Nevo N, Gasnier B, Antignac C. (2004) Molecular pathogenesis of cystinosis: effect of *CTNS* mutations on the transport activity and subcellular localization of cystinosin. *Hum Mol Genet*. 13(13):1361-1371. PMID: 15128704

Cherqui S, Sevin C, Hamard G, Kalatzis V, Sich M, Pequignot M.O, Gogat K, Abitbol M, Broyer M, Gubler M.C, Antignac C. (2002) Intra-lysosomal cystine accumulation in mice lacking cystinosin, the protein defective in cystinosis. *Mol Cell Biol.* 22(21): 7622-7632. PMID: 12370309

Kalatzis V, **Cherqui S**, Antignac C, Gasnier B. (2001) Cystinosin, the protein defective in cystinosis, is a H^+-driven lysosomal cystine transporter. *EMBO J*. 20(21): 5940-5949. PMID: 11689434

Kalatzis V, **Cherqui S**, Jean G, Cordier B, Broyer M, Cochat P, Antignac C. (2001) Characterisation of a putative founder mutation accounts for high incidence of cystinosis in Brittany. *J Am Soc Nephrol.* 12(10): 2170-2174. PMID: 11562417

Cherqui S, Kalatzis V, Trugnan G, Antignac C. (2001) The targetting of cystinosin to the lysosomal membrane requires a tyrosine-based signal and a novel sorting motif. *J Biol Chem*. 276: 13314-13321. PMID: 11150305

Cherqui S, Kalatzis V, Forestier L, Poras I, Antignac C. (2000) Identification and Characterisation of the Murine Homologue of the Gene Responsible for Cystinosis, *Ctns*. *BMC Genomics*. 1(1):2. PMID: 11121245

Attard M, Jean G, Forestier L, **Cherqui S**, van't Hoff W, Broyer M, Antignac C, Town M. (1999) Severity of phenotype in cystinosis varies with mutations in the *CTNS* gene: predicted effect on the model of cystinosin. *Hum Mol Genet*. 8(13): 2507-2514. PMID: 10556299

Forestier L, Jean G, Attard M, **Cherqui S**, Lewis C, van't Hoff W, Broyer M, Town M, Antignac C. (1999) Molecular characterization of *CTNS* deletions in nephropathic cystinosis: development of a PCR-based detection assay. *Am J Hum Genet*. 65(2): 353-359. PMID: 10417278

Town M, Jean G, **Cherqui S**, Attard M, Forestier L, Whitmore SA, Callen DF, Gribouval O, Broyer M, Bates GP, van't Hoff W, Antignac C. (1998) A novel gene encoding an integral membrane protein is mutated in nephropathic cystinosis. *Nature Genet*. 18(4): 319-324. PMID: 9537412

BOOK CHAPTERS

Rocca C. and **Cherqui S**. (2019) Gene Transfer to the Mouse Kidney in Vivo. In Viral Vectors for Gene Therapy. 1937:227-234 - Methods in Molecular Biology, published by Humana Press, USA (part of the Springer Publishing Group)

Cherqui S, Kalatzis V, **Cherqui S**, Antignac C. (2015) Cystinosis. In Genetic Diseases of the kidney, Second Edition. Elsevier

PATENTS - COMPANY

Patent entitled “Methods of treating mitochondrial disorders” (#20378-201301) – Cherqui Inventor

Patent entitled “Methods of treating lysosomal disorders” (#20378-101530) - Cherqui co-Inventor

2016 - present Co-founder of GenStem Therapeutics, Inc.

INVESTIGATIONAL NEW DRUG

IND #018631 (CTNS-RD-04; Treatment of Cystinosis)

A Phase 1/2 Study to Determine the Safety and Efficacy of Transplantation with Autologous Human $CD34^+$ Hematopoietic Stem Cells (HSC) from Mobilized Peripheral Blood Stem Cells (PBSC) of Patients with Cystinosis Modified by *Ex Vivo* Transduction using the pCCL-CTNS Lentiviral Vector

FDA-approved in December 19, 2018

GRANTS

Ongoing:

California Institute of Regenerative Medicine (CIRM) CLIN1-09230 11/01/2016 - 01/31/2019
Ex vivo transduced autologous human CD34+ hematopoietic stem cells for treatment of cystinosis
To complete the pharmacology/toxicology studies required by the FDA, purchase the GMP lentivirus preparation, perform the manufacturing development and write the IND for autologous transplantation of *ex vivo* gene-modified hematopoietic stem cells using a lentivirus vector containing *CTNS* transgene.
Role: PI

NIH/NIDDK 2R01-DK090058-6 01/01/2016 - 08/31/2020
Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis
To investigate strategies to improve the stem cell-gene therapy approach for cystinosis and to investigate which patients' genetic profile will benefit the most from the future clinical trial.
Role: PI

Cystinosis Research Foundation 09/01/2015 - 08/31/2019
Mechanism of bone marrow stem cell-mediated therapy in the mouse model of cystinosis
To investigate the mechanisms by which transplantation of hematopoietic stem cells expressing a functional *Ctns* gene lead to cystine decrease and tissue preservation in the mouse model for cystinosis.
Role: PI

Cystinosis Research Foundation 01/01/2018 - 12/31/2018
Intra-dermal Imaging of Subjects with Cystinosis using Confocal Microscopy
To investigate intra-dermal images of the skin from patients with cystinosis as a method to estimate the quantity of cystine crystals in this tissue compartment.
Role: PI

Friedreich's Ataxia Research Alliance (FARA) 02/01/2018 - 01/31/2019
Stem Cell Gene Therapy for Friedreich's Ataxia
The major goal of this project is to investigate the mechanism of action for stem cell-mediated tissue rescue for Friedreich's Ataxia.
Role: PI

NIH/NINDS R01-NS108965 09/01/2018 – 08/31/2023
Stem Cell Gene Therapy for Friedreich's Ataxia
The major goal is to develop an ex vivo hematopoietic stem cell gene therapy approach for the treatment of Friedreich's ataxia.

Role: PI

NIH/NIDDK R01-DK110162-01A1 07/01/2017 – 06/30/2021
Molecular and Cellular Mechanisms of the Lysosomal Storage Disease Cystinosis
To develop new understanding of the molecular mechanism regulating Chaperone mediated autophagy in nephropathic cystinosis.
Role: co-PI (PI: Catz, The Scripps Research Institute)

NIH/NIDDK R21-EY028642-01A 09/01/2017 - 08/31/2019
Chaperone-mediated autophagy in corneal cystinosis
The major goal of this project is to develop new mechanistic approaches mechanistic to understand the molecular processes leading to corneal disease in cystinosis.
Role: co-PI (PI: Catz, The Scripps Research Institute)

California Institute of Regenerative Medicine (CIRM) DISC2-11131 09/01/2018 – 08/31/2020
Genetically Modified Hematopoietic Stem Cells for the Treatment of Danon Disease
The major goals of this project are to develop the autologous transplantation of *ex vivo* gene-modified hematopoietic stem cells using a lentivirus vector for Danon disease.
Role: co-PI (PI: Adler, UCSD)

Completed:

Cystinosis Research Foundation 09/01/13 - 08/31/17
Pharmacology/Toxicology studies for gene-modified stem cell transplantation for cystinosis
To evaluate the impact of cysteamine on the stem cell transplantation and to coordinate a medical panel for clinical trial and collect information for natural history report on cystinosis.
Role: PI

Sanford Stem Cell Clinical Center 09/01/15 - 10/31/16
A one arm, open label, single treatment safety and efficacy study of pCCL-CTNS modified CD34+ hematopoietic stem cells after autologous transplantation in patients with nephropathic cystinosis
To complete the pharmacology/toxicology studies required by the FDA, to prepare and submit the regulatory documents and to initiate the phase I clinical trial for autologous transplantation of *ex vivo* gene-modified hematopoietic stem cells using a lentivirus vector containing *CTNS* transgene.

NIH/NINDS R21-NS090066 08/15/14 - 07/31/16
Hematopoietic stem cell-based therapy for Friedrich Ataxia
To evaluate the impact of hematopoietic stem and progenitor cell transplantation on Friedrich Ataxia in the mouse model.
Role: PI

NIH/NIDDK R01-DK090058 01/01/11 - 12/31/15
Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis
To develop a multisystemic strategy for the treatment of cystinosis and establish the proof-of concept for the transplantation of autologous hematopoietic stem cells genetically modified *ex vivo* to express a functional *CTNS* gene using a lentiviral vector.
Role: PI

NIH/NIDDK R01-DK099338 07/01/13 - 06/30/15
Toxicology studies for gene-modified stem cell transplantation for cystinosis

To perform the pharmacology/toxicology studies required to obtain an IND for a phase I clinical trial for an autologous transplantation of *ex vivo* gene-modified hematopoietic stem cells using a lentivirus vector containing a functional *CTNS* transgene for cystinosis.
Role: PI

NIH/NIDDK R21-DK090548-01A1 09/01/11 - 08/31/13
Kidney-targeted gene delivery for cystinosis
To demonstrate that renal vein injection of adeno-associated virus (AAV) expressing a functional *CTNS* gene will treat or prevent the renal defects in cystinosis when delivered very early in the disease and ameliorate the renal disease if administered to older patients.
Role: PI

Cystinosis Research Foundation 07/01/09 - 06/30/11
Stem cell and gene therapy for cystinosis
To develop two strategies for the treatment of cystinosis:
- A multisystemic strategy: transplantation of bone marrow stem cells, comparison of whole bone marrow, hematopoietic stem cells and mesenchymal stem cells.
- A kidney-specific strategy: optimization of retrograde ureteral injection of self-complementary adeno-associated virus (scAAV).
Role: PI