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1. Personal statment

After being trained as a medical geneticist, I realized that my passion lay in identification of molecular mechanisms crucial for neuronal function and how errors in that drove the major neurodegenerative diseases. This led to my interest in the role of RNA processing alterations in neuronal health and dysfunction. During my postdoctoral training, I applied approaches in genomics to study the impact of two RNA binding proteins, TDP-43 and FUS/TLS, on RNA splicing and gene regulation. My efforts have demonstrated that RNA processing alterations are crucial in the pathogenesis of neurodegenerative diseases including amyotrophic lateral sclerosis (ALS) and frontotemporal dementia (FTD).
With my young team, I now use a combination of genomic approaches in mouse models, human brain and neurons converted from patient fibroblasts to define the roles of RNA-binding proteins related to neurodegenerative diseases. The goal is to compare regulatory networks to identify mechanisms by which aggregation-prone RNA-binding proteins contribute to neuronal function and may trigger neuronal death. The molecular signatures we identify serve as a functional readout to screen therapeutic compounds, and one part of my rationale is that uncovering which RNAs are abnormally processed upon ALS-causing mutations will open opportunities for interventional strategies. We also investigate disease mechanisms linked to a hexanucleotide repeat expansion in the gene C9orf72 that was recently identified as the most common genetic cause of ALS and FTD. Along with my collaborators at UCSD and corporate partner ISIS Pharmaceuticals, we have already established that antisense oligonucleotides (ASOs) targeting C9orf72 transcripts efficiently reduce pathological RNA foci in patient cells. We are now pursuing our efforts to define therapeutic targets and provide crucial information for the design of a clinical trial using ASOs in C9orf72 ALS/FTD patients.

2. Education

2008 **Ph.D.** in Molecular and Cellular Biology (***University Louis Pasteur, Strasbourg*****)**

2004 **Board certification in Medical Genetics** (***Medical School of Strasbourg, France***)

2003 **M.D.** (***Medical School of Strasbourg, France***)

2002 **M.S.** in Molecular and Cellular Biology (***University Louis Pasteur, Strasbourg***)

2002 Statistical methods training at the Centre d'Enseignement de la Statistique Appliquée à la Médecine (CESAM; ***University of Paris VI, France***)

1993-1999 Medical studies (***Medical School of Marseille, France***)

3. Professional experience

2013- **Assistant Investigator, Ludwig Institute for Cancer Research** (***University of California, San Diego, USA***)

2013- **Assistant Professor, Department of Neurosciences** (***University of California, San Diego, USA***)

2008-2013 Department of Cellular and Molecular Medicine and Ludwig Institute for Cancer Research, **Professor Don Cleveland** (*Ludwig Institute for Cancer Research, University of California, San Diego, USA*)

Postdoctoral Researcher: *Determining RNA metabolism alterations in amyotrophic lateral sclerosis*

2004-2008 Institut de Génétique et de Biologie Moléculaire et Cellulaire (*IGBMC*), **Professor Michel Koenig** (*University Louis Pasteur, Strasbourg*)

PhD thesis: *Identification and characterization of novel genes for autosomal recessive ataxia*

2004-2008 **Medical Geneticist** in Department of Genetics directed by Professor Jean-Louis Mandel (*Medical School of Strasbourg, France*)

2004 Department of Neurology, *Columbia University, College of Physicians and Surgeons*, **Professor Michio Hirano** (New York, USA)

Residency training, 1 year: *Cellular models for mitochondrial neuro-gastrointestinal encephalomyopathy*
Participation in Professor Hirano's clinical activity at the Eleanor and Lou Gehrig MDA/ALS multidisciplinary care center

2003 Clinical Genetics and Cytogenetics Department, Professor Hélène Dollfus (*Medical School of Strasbourg, France*)

Resident in Medical Genetics, 6 months

2003 Molecular Genetics Department, Professor Jean-Louis Mandel (*Medical School of Strasbourg, France*)

Resident in Medical Genetics, 6 months

2002 Institut de Génétique et de Biologie Moléculaire et Cellulaire (*IGBMC*), Professor Michel Koenig (*University of Strasbourg, France*)

Master training: *Homozygosity mapping of Joubert syndrome and Marinesco-Sjogren syndrome*

2001 Cytogenetics Department, Doctor Eric Jeandidier (*Hospital of Mulhouse, France*)
Resident in Medical Genetics, 6 months

2001 Molecular Genetics Department, Professor Jean-Louis Mandel (*Medical School of Strasbourg, France*)
Resident in Medical Genetics, 6 months

2000 Pediatrics Department, Professor Michel Fishbach (*Medical School of Strasbourg, France*)
Resident in Medical Genetics, 6 months

1999 Oncology Department, Professor Jean-Pierre Bergerat (*Medical School of Strasbourg, France*)
Resident in Medical Genetics, 6 months

1996 Molecular Genetics Laboratory, Professor Nicolas Levy (*Medical School of Marseille, France*)
Molecular biology training: *Molecular diagnosis of Charcot-Marie-Tooth type 1A and Smith Magenis diseases*

4. Honors and Awards

2013 **The Frick Foundation 2013 Award** (conjointly with Dr. Luc Dupuis)

2013 **Ray Thomas Edwards Foundation Recognition Award**

2011 **Muscular Dystrophy Association** (MDA) **Career Development Grant**

2011 **NIH traveling fellowship for Young investigators**

2010 Cold Spring Harbor course "**Advanced Sequencing Technologies and Applications**"

2009 **The Milton-Safenowitz Postdoctoral Fellowship** from the **Amyotrophic Lateral Sclerosis Association** (ALSA)

2004 **Alphonse Laveran Prize** for MD thesis Medical School of Strasbourg, France.

2003 **Association Française contre les Myopathies** (AFM) Fellowship for training at Columbia University

2002 **Fondation pour la Recherche Médicale** (FRM) Fellowship for Master training

2002 **Association Française contre les Myopathies** Fellowship for Master training

5. Professional Service and Memberships

2014 Member, American Neurological Association (ANA)

2013 Scientist Reviewer for the Department of Defense Congressionally Directed Medical Research

2013 Scientific Reviewer for the Amyotrophic Lateral Sclerosis Society of Canada

2011- International Scientific Committee for the Italian Agency for Research on Amyotrophic Lateral Sclerosis (AriSLA)

2011- Review Editorial Board of Frontiers in Neurogenomics

2010- Member, Society for Neuroscience

2008- Member, American Society of Human Genetics

2008- Scientific Referee for:

Nature	Nature Genetics	PLOS Genetics
Cell	Neuron	Lancet Neurology
PNAS	Nature Reviews Neurology	Annals of Neurology
Brain Research	Human Molecular Genetics	Journal of Neurology
JAMA Neurology	Journal of Neuroscience	Molecular Neurodeg

2004-2008 Member, Committee for Pre-implantation and Prenatal Diagnosis, Medical school of Strasbourg

2004-2008 Member, National Network for Molecular Diagnosis of Neurological and Muscular Diseases

2004-2008 Member, National Network for Genetic Diagnosis of Cystic Fibrosis

2000-2008 Member, European Society of Human Genetics

6. Teaching activities

2005 Lectures in Medical Genetics and Molecular Biology for 2nd year medical students at the *Medical School of Strasbourg* (12h)

2005 Lecture on Genetic Diseases for the Residency Program in Fetal Medicine at the *Medical School of Strasbourg* (2h)

2006 Lectures in Medical Genetics and Molecular Biology for 2nd year medical students at the *Medical School of Strasbourg* (12h)

2006 Lecture on Genetic Diseases for the Residency Program in Fetal Medicine at the *Medical School of Strasbourg* (2h)

2006 Lectures in Medical Genetics for the Residency Program in Biology at the *Medical School of Strasbourg* (4h)

2007 Lectures in Medical Genetics and Molecular Biology for 2nd year medical students at the *Medical School of Strasbourg* (12h)

2007 Lecture on Genetic Diseases for the Residency Program in Fetal Medicine at the *Medical School of Strasbourg* (2h)

2007 Lectures in Medical Genetics for the Residency Program in Biology at the *Medical School of Strasbourg* (4h)

2004-2007 Mentoring of Residents in Medical Genetics in the Genetic Department at the *Hospital of Strasbourg*

2008- Mentoring of five undergraduate students in the Independent Research Program in Biological Sciences (BISP199) at *Univ. of California at San Diego*

Jacqueline Moran	BISP199	2009-2010
Devesh Vashishtha	BISP199	2010
Kathryn Baker	BISP199	2010-2011
Max Dukarevich	BISP199	2011
Kevin Clutario	BISP199	2010-2012

2011 Lectures for the Neurosciences Graduate program at *Univ. of California at San Diego* (Class NEU268 "Molecular and Cellular Neurobiology" and Class NEU221 "Advance Topics in Neurosciences") (2h)

2014 Lectures for the Neurosciences Graduate program at *Univ. of California at San Diego* (Class NEU221 "Neurobiology of Diseases: Mechanisms of Neurodegeneration") (4h)

2014 Mentoring of Melanie Jambeau, student in Master 2 from University of Lorraine (France)

7. Invited Presentations

Targeted degradation of sense and antisense C9orf72 nuclear foci as therapy for ALS and FTD. International Center for Genetic Engineering and Biotechnology Conference, RNA metabolism: Changing Paradigms in Neurodegeneration, May 2014, Trieste, Italy.

Disease mechanisms and therapeutic strategies for two overlapping neurodegenerative diseases: ALS and FTD.

Shiley-Marcos Alzheimer's Disease Research Center, Symposium Alzheimer's Disease and Related Dementias, May 2014, San Diego, USA.

Targeted degradation of sense and antisense C9orf72 nuclear foci as therapy for ALS and FTD.
2014 Chicago Symposium on Translational Neuroscience, May 2014, Chicago, USA.

Genomewide analysis of RNA processing alterations in ALS to screen drug candidates.
Target ALS Annual Meeting, April 2014, New York, USA.

Disease mechanisms and therapeutic approach for ALS/FTD from C9orf72 expansion
California ALS Research Summit, January 2014, Fresno, USA.

Targeted degradation of sense and antisense C9orf72 nuclear foci as therapy for ALS and FTD.
24th International symposium on ALS/MND, December 2013, Milan, Italy.

Targeted degradation of sense and antisense C9orf72 nuclear foci as therapy for ALS and FTD.
9th symposium Fondation André-Delambre, September 2013, Quebec, Canada.

RNA processing Alterations in Neurodegeneration.
Invited seminar, hosted by Professor Laura Ranum, Center for NeuroGenetics, University of Florida, March 2013, USA.

Antisense oligonucleotides target nuclear RNA foci in ALS and frontotemporal dementia with C9orf72 expansion.
13th Annual Robert Packard Center for ALS Research Symposium, February 2013, Baltimore, USA.

RNA processing Alterations in Neurodegeneration.
Invited seminar, hosted by Professor Michael Snyder, Department of Genetics and the Center for Genomics and Personalized Medicine, Stanford University School of Medicine, January 2013, USA.

Convergent roles of FUS/TLS and TDP-43 in processing RNAs with long introns.
23rd International symposium on ALS/MND, December 2012, Chicago, USA.

Antisense oligonucleotides target nuclear RNA foci in ALS and frontotemporal dementia with C9orf72 expansion.
Neurodegenerative diseases meeting, November 2012, Cold Spring Harbor, USA.

Mechanisms and Therapy for ALS/FTD from C9orf72.
Symposium at the Society for Neuroscience Annual Meeting, October 2012, New Orleans, USA.

TDP-43 and FUS/TLS: emerging roles in RNA processing and neurodegeneration.
Invited seminar, hosted by Professor Kristian Baker, Center for RNA Molecular Biology, Case Western Reserve University, October 2012, Cleveland, USA.

Antisense Oligonucleotide therapy for Huntington's disease.
24 hours for Huntington's disease, October 2012, Case Western Reserve University, Cleveland, USA.

Determining RNA processing alterations in amyotrophic lateral sclerosis patients with C9orf72 hexanucleotide expansion.
7th International Conference on Unstable Microsatellites and Human Disease, June 2012, Mount Ste. Odile, Strasbourg, France.

TDP-43 and FUS/TLS: emerging roles in RNA processing and neurodegeneration.
Invited seminar, hosted by Professor Christopher Henderson, Motor Neuron Center, Columbia University, September 2011, New York, USA.

TDP-43: emerging roles in RNA processing and neurodegeneration.
Invited seminar, hosted by Professor Ruth Luthi-Carter, Ecole Polytechnique Fédérale de Lausanne (EPFL), Brain Mind Institute, July 2011, Lausanne, Switzerland.

RNA processing alterations in the pathogenesis of ALS.
The 2011 California ALS Research Summit, September 2011, Los Angeles, USA.

TDP-43 and FUS/TLS: emerging roles in RNA processing and neurodegeneration.
7th symposium Fondation André-Delambre, September 2011, Quebec, Canada.

Role of RNA processing in ALS.
21st International symposium on ALS/MND, December 2010, Orlando, USA.

RNA targets for TDP-43:identifying the basis for neuronal vulnerability in ALS.
6th symposium Fondation André-Delambre, September 2010, Quebec, Canada.

ADCK3, une kinase mitochondriale ancestrale, est mutée dans une nouvelle forme d'ataxie recessive associée à un deficit en coenzyme Q_{10}.
Assises de Génétique Humaine, January 2008, Lille, France.

Mitochondrial ADCK3, an ancestral prokaryotic kinase involved in coenzyme Q biosynthesis, is mutated in a new form of recessive ataxia.
American Society of Human Genetics, October 2007, San Diego, USA.

Mitochondrial CABC1, an ancestral prokaryotic kinase involved in Coenzyme Q biosynthesis, is mutant in a new form of recessive ataxia.
European Society of Human Genetics, June 2007, Nice, France.

The Marinesco-Sjogren Disease.
SPATAX Meeting June 2006, Paris, France.

Homozygosity mapping of Marinesco-Sjogren syndrome to 5q31.
European human Genetic Conference, May 2003, Birmingham, England.

8. Invitation to Workshops

Designing phase I/IIA clinical trials for Huntington's disease.
Milton Wexler Interdisciplinary Workshop at the Hereditary Disease Foundation, December 2011, New York

Does RNA participate directly in the pathogenesis of Huntington's disease ?
Organized by Pr. Allan Tobin and Pr Ethan Signer at the CHDI Foundation, November 2011, Los Angeles

How can RNA profiling best provide pathogenic insights and pharmacodynamic biomakers for Huntington's disease ?
Organized by Pr. Allan Tobin and Pr Ethan Signer at the CHDI Foundation, May 2011, Los Angeles

9. Publications **(35 published; 2136 career citations)**

1. **Lagier-Tourenne C.,** Chaigne D., Gong J., Flori J., Mohr M., Ruh D., Christmann D., Flament J., Mandel J.L., Koenig M.*, et al.* (2002). Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjogren syndrome. **J Med Genet** *39*, 838-843.

2. Biancalana V., Caron O., Gallati S., Baas F., Kress W., Novelli G., D'Apice M.R., **Lagier-Tourenne C.,** Buj-Bello, A., Romero, N.B., Mandel, J.L. (2003). Characterisation of mutations in 77 patients with X-linked myotubular myopathy, including a family with a very mild phenotype. **Hum Genet** *112*, 135-142.

3. **Lagier-Tourenne C.,** Tranebaerg L., Chaigne D., Gribaa M., Dollfus H., Silvestri G., Betard C., Warter J.M., and Koenig M. (2003). Homozygosity mapping of Marinesco-Sjogren syndrome to 5q31. **Eur J Hum Genet** *11*, 770-778.

4. **Lagier-Tourenne C.,** Ginglinger E., Alembik Y., De Saint Martin A., Peter M.O., Dulucq P., Jonveaux P., and Jeandidier E. (2004). Two cousins with partial trisomy 12q and monosomy 12p recombinants of a familial pericentric inversion of the chromosome 12. **Am J Med Genet** *125A*, 77-85.

5. **Lagier-Tourenne C.,** Boltshauser E., Breivik N., Gribaa M., Betard C., Barbot C., and Koenig M. (2004). Homozygosity mapping of a third Joubert syndrome locus to 6q23. **J Med Genet** *41*, 273-277.

6. **Lagier-Tourenne C.,** Chaigne D., Christmann D., Dollfus H. (2004). Affections malformatives et héréditaires des voies optiques d'expression neuro-ophtalmologique. Neuro-Ophtalmologie, Rapport de la Société Française d'Ophtalmologie, Edition Masson, 323-345.

7. Hirano M., **Lagier-Tourenne C.,** Valentino M.L., Marti R., and Nishigaki Y. (2005). Thymidine phosphorylase mutations cause instability of mitochondrial DNA. **Gene** *354*, 152-6.

8. Anttonen A.K., Mahjneh I., Hamalainen R.H., **Lagier-Tourenne C.,** Kopra O., Waris L., Anttonen M., Joensuu T., Kalimo H., Paetau A., Tranebjaerg L., Chaigne D., Koenig M., Eeg-Olofsson O., Udd B., Somer M., Somer H., Lehesjoki A.E. (2005). The gene disrupted in Marinesco-Sjogren syndrome encodes SIL1, an HSPA5 cochaperone. **Nat Genet** *37*, 1309-1311.

9. Curbo S., **Lagier-Tourenne C.,** Carrozzo R., Palenzuela L., Lucioli S., Hirano M., Santorelli F., Arenas J., Karlsson A., and Johansson M. (2006). Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. **Genomics** *87*, 410-416.

10. Valente E.M., Brancati F., Silhavy J.L., Castori M., Marsh S.E., Barrano G., Bertini E., Boltshauser E., Zaki M.S., Abdel-Aleem A., Abdel-Salam G.M., Bellacchio E., Battini R., Cruse R.P., Dobyns W.B., Krishnamoorthy K.S., **Lagier-Tourenne C.,** Magee A., Pascual-Castroviejo I., Salpietro C.D., Sarco D., Dallapiccola B., Gleeson J.G., International JSRD Study Group. (2006). AHI1 gene mutations cause specific forms of Joubert syndrome-related disorders. **Ann Neurol** *59*, 527-534.

11. **Lagier-Tourenne C.,** Hirano M. (2006). Neurogenetics: Scientific and Clinical Advances. Chapter: Mitochondrial Disorders. Edited by David Lynch, 261-288.

12. Gribaa M., Salih M., Anheim M., **Lagier-Tourenne C.,** H'Mida D., Drouot N., Mohamed A., Elmalik S., Kabiraj M., Al-Rayess M., Almubarak M., Betard C., Goebel H., Koenig M. (2007). A new form of childhood onset, autosomal recessive spinocerebellar ataxia and epilepsy is localized at 16q21-q23. **Brain** *130*, 1921-1928.

13. Dam A.H., Koscinski I., Kremer J.A., Moutou C., Jaeger A.S., Oudakker A.R., Tournaye H., Charlet N., **Lagier-Tourenne C.,** van Bokhoven H., Viville S. (2007). Homozygous mutation in SPATA16 is associated with male infertility in human globozoospermia. **Am J Hum Genet** *81*, 813-820.

14. **Lagier-Tourenne C.,** Tazir M., Lopez L.C., Quinzii C.M., Assoum M., Drouot N., Busso C., Makri S., Ali-Pacha L., Benhassine T., Anheim M., Lynch D.R., Thibault C., Plewniak F., Bianchetti L., Tranchant C., Poch O., DiMauro S., Mandel J.L., Barros M.H., Hirano M., Koenig M. (2008). ADCK3, an ancestral kinase, is mutated in a form of recessive ataxia associated with coenzyme Q10 deficiency. **Am J Hum Genet** *82*, 661-672.

15. Anheim M., **Lagier-Tourenne C.**, Stevanin G., Fleury M., Durr A., Namer IJ., Denora P., Brice A., Mandel J.L., Koenig M., Tranchant C. (2009). SPG11 spastic paraplegia. Anew cause of juvenile parkinsonism. **J Neurol** *256*, 104-108.

16. Cossée M*., **Lagier-Tourenne C*.,** Seguela C., Mohr M., Leturcq F., Gundesli H., Chelly J., Tranchant C., Koenig M., Mandel J.L. (2009). Use of SNP array analysis to identify a novel TRIM32 mutation in limb-girdle muscular dystrophy type 2H. **Neuromusc Disord** *19*, 255-260.

17. **Lagier-Tourenne C.,** Cleveland D.W. (2009). Rethinking ALS: the FUS about TDP-43. **Cell** *136,* 1001-4.

18. Travaglini L., Brancati F., Attie-Bitach T., Audollent S., Bertini E., Kaplan J., Perrault I., Iannicelli M., Mancuso B., Rigoli L., Rozet J.M., Swistun D., Tolentino J., Dallapiccola B., Gleeson J.G., Valente E.M., International JSRD Study Group, Zankl A., Leventer R., Grattan-Smith P., Janecke A., D'Hooghe M., Sznajer Y., Van Coster R., Demerleir L., Dias K., Moco C., Moreira A., Kim C.A., Maegawa G., Petkovic D., Abdel-Salam G.M., Abdel-Aleem A., Zaki M.S., Marti I., Quijano-Roy S., Sigaudy S., de Lonlay P., Romano S., Touraine R., Koenig M., **Lagier-Tourenne C.**, *et al.* (2009). Expanding CEP290 mutational spectrum in ciliopathies. **Am J Med Genet** *149A*, 2173-2180.

19. Braida C., Stefanatos RK., Adam B., Mahajan N., Smeets HJ., Niel F., Koenig M., **Lagier-Tourenne C.**, Mandel JL., Faber CG., de Die-Smulders CE., Spaans F., Monckton DG. (2010). Variant CCG and GGC repeats within the CTG expansion dramatically modify mutational dynamics and likely contribute toward unusual symptoms in some myotonic dystrophy type 1 patients. **Hum Mol Genet** *19,* 1399-412.

20. **Lagier-Tourenne C.*,** Polymenidou M.*, Cleveland DW. (2010). TDP-43 and FUS/TLS: emerging roles in RNA processing and neurodegeneration. **Hum Mol Genet** *19, R46-64.*

21. Quinzii CM, Lopez LC, Gilkerson RW, Dorado B, Coku J, Naini AB, **Lagier-Tourenne C.**, Schuelke M, Salviati L, Carrozzo R, Santorelli F, Rahman S, Tazir M, Koenig M, Dimauro S, Hirano M. (2010). Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ10 deficiency. **FASEB J** *24,* 3733-43.

22. Ling SC, Albuquerque CP, Han JS, **Lagier-Tourenne C.**, Tokunaga S, Zhou H, Cleveland DW. (2010). ALS-associated mutations in TDP-43 increase its stability and promote TDP-43 complexes with FUS/TLS. **PNAS** *107,*13318-23.

23. **Lagier-Tourenne C.,** Cleveland DW. (2010). Neurodegeneration: An expansion in ALS genetics. **Nature** *466,* 1052-3.

24. H'mida-Ben Brahim D., M'zahem A., Assoum M., Bouhlal Y., Fattori F., Anheim M., Ali-Pacha L., Ferrat F., Chaouch M., **Lagier-Tourenne C.**, Drouot N., Thibaut C., Benhassine T., Sifi Y., Stoppa-Lyonnet D., N'guyen K., Pouget J., Hamri A., Hentati F., Amouri R., Santorelli FM., Tazir M., Koenig M. (2011). Molecular diagnosis of known recessive ataxias by homozygosity mapping with SNP arrays. **J Neurol** *258,* 56-67.

25. Assoum M., Salih M.A., Drouot N., H'Mida-Ben Brahim D., **Lagier-Tourenne C.**, AlDrees A., Elmalik S.A., Ahmed T.S., Seidahmed M.Z., Kabiraj M.M., Koenig M. (2010). Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. **Brain** *133,* 2439-47.

26. Polymenidou M.*, **Lagier-Tourenne C.***, Hutt K.R.*, Huelga S.C., Moran J., Liang T.Y., Ling S.C., Sun E., Wancewicz E., Mazur C., Kordasiewicz H., Sedaghat Y., Donohue J.P., Shiue L., Bennett C.F., Yeo G.W., Cleveland D.W. (2011). Long pre-mRNA depletion and RNA missplicing contribute to neuronal vulnerability from loss of TDP-43. **Nat Neurosci** *14*, 459-468.

27. Polymenidou M.*, **Lagier-Tourenne C.***, Hutt K.R.*, Bennett C.F., Cleveland D.W, Yeo G.W. (2012). Misregulated RNA processing in amyotrophic lateral sclerosis. **Brain Res** *1462*, 3-15.

28. **Lagier-Tourenne C.***, Polymenidou M.*, Hutt K.R.*, Vu A.Q, Clutario K.M., Baughn M., Huelga S.C., Ling S.C., Liang T.Y., Mazur C., Wancewicz E., Salim A., Watt A., Freier S., Hicks G.G, Donohue J.P., Shiue L., Bennett C.F., Ravits J., Cleveland D.W., Yeo G.W. (2012). Divergent roles of ALS-linked proteins FUS/TLS and TDP-43 intersect in processing long pre-mRNAs. **Nat Neurosci** *15*, 1488-1497.

29. Thauvin-Robinet C., et al., collaborating working group on p.Arg117His. (2013). CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. **J Med Genet** *50*, 220-227.

30. Arnold E.S., Ling S.C., Huelga S.C., **Lagier-Tourenne C.**, Polymenidou M., Ditsworth D., Kordasiewicz H.B., McAlonis-Downes M., Platoshyn O., Parone P.A., Da Cruz S., Clutario K.M., Swing D., Tessarollo L., Marsala M., Shaw C.E., Yeo G.W., Cleveland D.W. (2013). ALS-linked TDP-43 mutations produce aberrant RNA splicing and adult-onset motor neuron disease without aggregation or loss of nuclear TDP-43. **PNAS** *110*, E736-45.

31. **Lagier-Tourenne C.*,** Baughn M.*, Rigo F., Sun S., Liu P., Li H-R., Jiang J., Watt A., Chun S., Katz M., Qiu J., Sun Y., Ling S-C., Zhu Q., Polymenidou M., Drenner K., Artates J.W., McAlonis M.M., Markmiller S., Hutt R.R., Pizzo D.P., Cady J., Harms M.B., Baloh R.H., VandenBerg S.R., Yeo G.W, Fu X.D., Bennett C.F., Cleveland D.W., Ravits J. (2013) Targeted degradation of sense and antisense *C9orf72* RNA foci as therapy for amyotrophic lateral sclerosis and frontotemporal dementia. **PNAS** *110*:E4530-9.

32. Meyer K., Ferraiuolo L., Miranda C.J., Likhite S., McElroy S., Renusch S., Ditsworth D., **Lagier-Tourenne C.**, Smith R.A., Ravits J., Burghes A.H., Shaw P.J., Cleveland D.W., Kolb S.J., Kaspar B.K. (2014). Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. **PNAS** *111:*829-32.

33. Crotti A., Benner C., Kerman B.E., Gosselin D., **Lagier-Tourenne C.**, Zuccato C., Cattaneo E., Gage F.H., Cleveland D.W., Glass C.K. (2014). Mutant Huntingtin promotes autonomous microglia activation via myeloid lineage-determining factors. **Nat Neurosci** *17*, 513-521.

34. Hu J., Liu J., Yu D., Aiba Y., Lee S., Pendergraff H., Boubaker J., Artates J.W., **Lagier-Tourenne C.**, Lima W.F., Swayze E.E., Prakash T.P., Corey D.R. (2014). Exploring the effect of sequence length and composition on allele-selective inhibition of human huntingtin expression by single-stranded silencing RNAs. **Nucleic Acid Ther** Apr 2.